Rare Disease Conditions Clinical Data Models

Maintenance release v1.9.0

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1 Document History and Control

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Version History

Version	Date	Description
1.9.0	09/04/2018	From finalised Data Model Catalogue v 1.9

Areas to consider

(The below is a list of suggested functions to consider before assigning reviewers for your document. Not all will be applicable, but it's a good idea to work through them to make sure all areas have been considered.)

Areas to consider*(delete where applicable)*		
Science - Rare Disease		

Quick links page

Introduction

Purpose of this document

The aim of this document is to provide an up-to-date list of eligibility criteria for conditions approved for recruitment within the Genomics England Rare Diseases Programme.

Structure and background to eligibility statements

For each disease listed we provide an "eligibility statement" composed of the following key information:

1. Inclusion criteria – the clinical features, characteristics or investigations that probands with a given disease must have in order to be eligible for recruitment.
2. Exclusion criteria - the clinical features, characteristics or investigation findings that participants with a given disease must not have in order to be eligible for recruitment.
3. Prior genetic testing – this sets out both in general terms, and where appropriate more specifically, the genetic testing which participants with a given disease must have performed prior to recruitment.

Each eligibility statement has been informed by at least one clinician specialising in the field and incorporates comments provided during the consultation period with Genomic Medicine Centres. Therefore, we would like to take this opportunity to thank this community for providing their expertise and understanding of complex disorders so generously.

Given the rapid progress in the understanding of rare diseases worldwide, it is important that the eligibility statements continue to be reviewed and developed over time in light of new discoveries and changes in clinical practice. Therefore we will continue our engagement with the clinical community throughout the lifetime of the project.

Summary of changes

The follow table summarises the changes in Rare Disease version 1.9. Only additions have been made to the catalogue: two new diseases have been added to the existing Categories, and a new Category – 'Genomic Medicine Service Indications' – has been added containing a new subcategory 'Whole Genome Sequencing Indications' and 22 new diseases. These new items are highlighted in yellow. The Genomic Medicine Service Indications will be available as first line tests in the new NHS Genomic Medicine Service. Prior to that, local clinical teams can use them as a first line test or in parallel to, or following, current diagnostic testing according to the clinical setting, noting that the 100,000 Genomes Project pipeline does not yet report on all variant types that will be available in the NHS pipeline and is not accredited. Other tests that should be considered are listed in the 'Where in pathway' section of the Inclusion criteria.

Category	Subcategory	Disease
Neurology and neurodevelopmental disorders (10988)	Motor and sensory disorders of the PNS (10991)	Pain channelopathies (82148)
Renal and urinary tract disorders (11000)	Syndromes with prominent renal abnormalities (11001)	Familial IgA nephropathy and IgA vasculitis (82147)
Genomic medicine service indications (82157)	Whole genome sequencing indications (82159)	GMS R14 Acutely unwell infants with a likely monogenic disorder (82160)
		GMS R27 Congenital malformation and dysmorphism syndromes - likely monogenic (82161)
		GMS R69 Floppy infant with a likely central cause (82162)
		GMS R29 Moderate, severe or profound intellectual disability (82163)
		GMS R89 Ultra-rare and atypical monogenic disorders (82164)
		GMS R100 Rare syndromic craniosynostosis or isolated multisuture synostosis (82165)
		GMS R104 Skeletal dysplasia (82166)
		GMS R143 Neonatal diabetes (82167)
		GMS R98 Likely inborn error of metabolism - targeted testing not possible (82168)
		GMS R83 Arthrogryposis (82185)

		GMS R84 Cerebellar anomalies (82169)
		GMS R87 Cerebral malformation (82170)
		GMS R61 Childhood onset hereditary spastic paraplegia (82171)
		GMS R109 Childhood onset leukodystrophy (82172)
		GMS R59 Early onset or syndromic epilepsy (82173)
		GMS R54 Hereditary ataxia with onset in adulthood (82174)
		GMS R55 Hereditary ataxia with onset in childhood (82175)
		GMS R85 Holoprosencephaly - NOT chromosomal (82176)
		GMS R86 Hydrocephalus (82177)
		GMS R381 Other rare neuromuscular disorders (82178)
		GMS R88 Severe microcephaly (82179)
		GMS R193 Cystic renal disease (82180)

Rare Disease Conditions Phenotypes and Clinical Tests

Cardiovascular disorders (10950)

Arteriopathies (33332)

Familial cerebral small vessel disease (36469)

Familial cerebral small vessel disease phenotypes (36902)

Level 3 Title	Arteriopathies (33332)
Level 4 Title	Familial cerebral small vessel disease (36469)
Phenotypes	Entries ordered left to right in table

Migraine with aura (HP:0002077)	Migraine without aura (HP:0002083)	Stroke (HP:0001297)
Ischemic stroke (HP:0002140)	Stroke-like episodes (HP:0002401)	Transient ischemic attack (HP:0002326)
Cerebral hemorrhage (HP:0001342)	Cerebral ischemia (HP:0002637)	Abnormality of the cerebral white matter (HP:0002500)
Seizures (HP:0001250)	Generalized tonic seizures (HP:0010818)	Dementia (HP:0000726)
Subcortical dementia (HP:0007123)	Cognitive impairment (HP:0100543)	Retinopathy (HP:0000488)
Hearing impairment (HP:0000365)	Unilateral deafness (HP:0009900)	Sensorineural hearing impairment (HP:0000407)
Peripheral neuropathy (HP:0009830)	Reduced consciousness/confusion (HP:0004372)	Encephalopathy (HP:0001298)
Gait disturbance (HP:0001288)	Gait apraxia (HP:0010521)	Ataxia (HP:0001251)
Abnormality of the retinal vasculature (HP:0008046)	Depression (HP:0000716)	Bipolar affective disorder (HP:0007302)
Anxiety (HP:0000739)	Apathy (HP:0000741)	Personality changes (HP:0000751)
Hypertension (HP:0000822)	Hyperlipidemia (HP:0003077)	Diabetes mellitus (HP:0000819)
Coronary artery disease (HP:0001677)	Peripheral arterial disease (HP:0004950)	Atrial fibrillation (HP:0005110)

	Angina pectoris (HP:0001681)	Myocardial infarction (HP:0001658)	Chronic kidney disease (HP:0012622)
	Abnormality of cardiovascular system physiology (HP:0011025)	Abnormal echocardiogram (HP:0003116)	Carotid artery stenosis (HP:0100546)
	Arrhythmia (HP:0011675)		

Familial cerebral small vessel disease clinical tests (36472)

Level 3 Title	Arteriopathies (33332)
Level 4 Title	Familial cerebral small vessel disease (36469)
Clinical Tests	NB. Clinical Test Guidance: General imaging diagnostics refers to MRI brain. General biopsy refers to skin biopsy (where taken). Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	General Biopsy (33614.1)

Familial Hypercholesterolaemia (33666)

Familial Hypercholesterolaemia phenotypes (33667)

Level 3 Title	Arteriopathies (33332)
Level 4 Title	Familial Hypercholesterolaemia (33666)
Phenotypes	Entries ordered left to right in table

Hyperlipidemia (HP:0003077)	Hypercholesterolemia (HP:0003124)	Hyperbetalipoproteinemia (HP:0003141)
Hyperlipoproteinemia (HP:0010980)	Myocardial infarction (HP:0001658)	Hypertriglyceridemia (HP:0002155)
Tendon xanthomatosis (HP:0010874)	Corneal arcus (HP:0001084)	

Familial Hypercholesterolaemia clinical tests (33668)

Level 3 Title	Arteriopathies (33332)
Level 4 Title	Familial Hypercholesterolaemia (33666)
Clinical Tests	Entries ordered left to right in table

Lipids (33158.1)

Severe hypertriglyceridaemia (42185)

Severe hypertriglyceridaemia phenotypes (42187)

Level 3 Title	Arteriopathies (33332)
Level 4 Title	Severe hypertriglyceridaemia (42185)
Phenotypes	Entries ordered left to right in table

Hyperlipidemia (HP:0003077)	Hypercholesterolemia (HP:0003124)	Hyperbetalipoproteinemia (HP:0003141)
Hyperlipoproteinemia (HP:0010980)	Myocardial infarction (HP:0001658)	Hypertriglyceridemia (HP:0002155)
Xanthelasma (HP:0001114)	Eruptive xanthomas (HP:0001013)	

Severe hypertriglyceridaemia clinical tests (42188)

Level 3 Title	Arteriopathies (33332)
Level 4 Title	Severe hypertriglyceridaemia (42185)
Clinical Tests	Entries ordered left to right in table

Lipids (33158.1)

Connective Tissues Disorders and Aortopathies (10951)

Familial Thoracic Aortic Aneurysm Disease (11021)

Connective Tissues Disorders phenotypes (28653)

Level 3 Title	Connective Tissues Disorders and Aortopathies (10951)
Level 4 Title	Familial Thoracic Aortic Aneurysm Disease (11021)
Phenotypes	Entries ordered left to right in table

Thoracic aortic aneurysm (HP:0012727)	Sinus of Valsalva aneurysm (HP:0011645)	Dilatation of the ascending aorta (HP:0005111)
Ascending aortic dissection (HP:0004933)	Dilatation of the descending thoracic aorta (HP:0004959)	Descending aortic dissection (HP:0012499)
Dilatation of the abdominal aorta (HP:0005112)	Abnormality of the mitral valve (HP:0001633)	Abnormality of the aortic valve (HP:0001646)
Arterial tortuosity (HP:0005116)	Aneurysm (HP:0002617)	Pulmonary artery dilatation (HP:0004927)
Arteriovenous fistula (HP:0004947)	Joint hypermobility (HP:0001382)	Joint dislocation (HP:0001373)
Scoliosis (HP:0002650)	Disproportionate tall stature (HP:0001519)	Arachnodactyly (HP:0001166)
Congenital finger flexion contractures (HP:0005879)	Abnormality of the sternum (HP:0000766)	Tendon rupture (HP:0100550)
Equinus calcaneus (HP:0008138)	Pes planus (HP:0001763)	Dolichocephaly (HP:0000268)
Ptosis (HP:0000508)	Downslanted palpebral fissures (HP:0000494)	Hypertelorism (HP:0000316)
Retrognathia (HP:0000278)	Cleft palate (HP:0000175)	Bifid uvula (HP:0000193)
Crumpled ear (HP:0009901)	Myopia (HP:0000545)	Ectopia lentis (HP:0001083)
Retinal detachment (HP:0000541)	Hyperextensible skin (HP:0000974)	Striae distensae (HP:0001065)
Atypical scarring of skin (HP:0000987)	Abnormality of the uterus (HP:0000130)	Pneumothorax (HP:0002107)
Hernia (HP:0100790)	Rectal prolapse (HP:0002035)	

Connective Tissues Disorders clinical tests (30852)

<table>
<tr><td>Level 3 Title</td><td>Connective Tissues Disorders and Aortopathies (10951)</td></tr>
<tr><td>Level 4 Title</td><td>Familial Thoracic Aortic Aneurysm Disease (11021)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Familial Thoracic Aortic Aneurysm Disease clinical test guidance:
Imaging Diagnostics refers to Cardiac MRI and/or High resolution chest CT
Additional body measurements refers to arm span and upper and lower segment measurement
General non-imaging diagnostics refers to pulmonary function tests
Entries ordered left to right in table
<table><tr><td>Blood pressure (30245.1)</td><td>Echocardiogram (29800.2)</td><td>Beighton test (31476.1)</td></tr><tr><td>Additional body measurements (30247.2)</td><td>General Non-imaging Diagnostics (34838.1)</td><td>General Imaging Diagnostics (33633.1)</td></tr></table></td></tr>
</table>

Brugada syndrome (11022)

Arrhythmia phenotypes (31379)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Brugada syndrome (11022)
Phenotypes	Entries ordered left to right in table

Prolonged PR interval (HP:0012248)	Atrioventricular block (HP:0001678)	Prolonged QTc interval (HP:0005184)
Shortened QT interval (HP:0012232)	J wave (HP:0012272)	Abnormal ST segment (HP:0012249)
Abnormal T-wave (HP:0005135)	Bundle branch block (HP:0011710)	Atrial fibrillation (HP:0005110)
Ventricular extrasystoles (HP:0006682)	Ventricular tachycardia (HP:0004756)	Effort-induced polymorphic ventricular tachycardias (HP:0004758)
Torsade de pointes (HP:0001664)	Ventricular fibrillation (HP:0001663)	Sudden cardiac death (HP:0001645)
Syncope (HP:0001279)	Cardiac arrest (HP:0001695)	Palpitations (HP:0001962)
Sensorineural hearing impairment (HP:0000407)	Seizures (HP:0001250)	Periodic paralysis (HP:0003768)
Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)	

Arrhythmia clinical tests (30854)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Brugada syndrome (11022)
Clinical Tests	Entries ordered left to right in table

ECG diagnostics (30183.2)	Cardiac MRI (31429.2)	Holter monitor test (31432.3)
Signal averaged ECG (31437.2)	Exercise test - cardiac (31445.3)	Echocardiogram (29800.2)
Electrophysiological study (31449.2)	Sodium channel blocker challenge (31403.3)	Alcohol intake (30206.1)
Exercise status (31469.1)	Epinephrine challenge (31404.3)	Smoking status (31464.1)

Long QT syndrome (11023)

Arrhythmia phenotypes (31379)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Long QT syndrome (11023)
Phenotypes	Entries ordered left to right in table

Prolonged PR interval (HP:0012248)	Atrioventricular block (HP:0001678)	Prolonged QTc interval (HP:0005184)
Shortened QT interval (HP:0012232)	J wave (HP:0012272)	Abnormal ST segment (HP:0012249)
Abnormal T-wave (HP:0005135)	Bundle branch block (HP:0011710)	Atrial fibrillation (HP:0005110)
Ventricular extrasystoles (HP:0006682)	Ventricular tachycardia (HP:0004756)	Effort-induced polymorphic ventricular tachycardias (HP:0004758)
Torsade de pointes (HP:0001664)	Ventricular fibrillation (HP:0001663)	Sudden cardiac death (HP:0001645)
Syncope (HP:0001279)	Cardiac arrest (HP:0001695)	Palpitations (HP:0001962)
Sensorineural hearing impairment (HP:0000407)	Seizures (HP:0001250)	Periodic paralysis (HP:0003768)
Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)	

Arrhythmia clinical tests (30854)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Long QT syndrome (11023)
Clinical Tests	Entries ordered left to right in table

ECG diagnostics (30183.2)	Cardiac MRI (31429.2)	Holter monitor test (31432.3)
Signal averaged ECG (31437.2)	Exercise test - cardiac (31445.3)	Echocardiogram (29800.2)
Electrophysiological study (31449.2)	Sodium channel blocker challenge (31403.3)	Alcohol intake (30206.1)
Exercise status (31469.1)	Epinephrine challenge (31404.3)	Smoking status (31464.1)

Catecholaminergic Polymorphic Ventricular Tachycardia (11024)

Arrhythmia phenotypes (31379)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Catecholaminergic Polymorphic Ventricular Tachycardia (11024)
Phenotypes	Entries ordered left to right in table

Prolonged PR interval (HP:0012248)	Atrioventricular block (HP:0001678)	Prolonged QTc interval (HP:0005184)
Shortened QT interval (HP:0012232)	J wave (HP:0012272)	Abnormal ST segment (HP:0012249)
Abnormal T-wave (HP:0005135)	Bundle branch block (HP:0011710)	Atrial fibrillation (HP:0005110)
Ventricular extrasystoles (HP:0006682)	Ventricular tachycardia (HP:0004756)	Effort-induced polymorphic ventricular tachycardias (HP:0004758)
Torsade de pointes (HP:0001664)	Ventricular fibrillation (HP:0001663)	Sudden cardiac death (HP:0001645)
Syncope (HP:0001279)	Cardiac arrest (HP:0001695)	Palpitations (HP:0001962)
Sensorineural hearing impairment (HP:0000407)	Seizures (HP:0001250)	Periodic paralysis (HP:0003768)
Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)	

Arrhythmia clinical tests (30854)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Catecholaminergic Polymorphic Ventricular Tachycardia (11024)
Clinical Tests	Entries ordered left to right in table

ECG diagnostics (30183.2)	Cardiac MRI (31429.2)	Holter monitor test (31432.3)
Signal averaged ECG (31437.2)	Exercise test - cardiac (31445.3)	Echocardiogram (29800.2)
Electrophysiological study (31449.2)	Sodium channel blocker challenge (31403.3)	Alcohol intake (30206.1)
Exercise status (31469.1)	Epinephrine challenge (31404.3)	Smoking status (31464.1)

Unexplained sudden death in the young (38566)

Unexplained sudden death in the young phenotypes (40121)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Unexplained sudden death in the young (38566)
Phenotypes	Entries ordered left to right in table

Sudden cardiac death (HP:0001645)	Syncope (HP:0001279)	Seizures (HP:0001250)
Abnormality of the cardiovascular system (HP:0001626)		

Unexplained sudden death in the young clinical tests (40122)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Unexplained sudden death in the young (38566)
Clinical Tests	NB. Clinical Test Guidance: General non-imaging diagnostics refers to results of post-mortem analysis Entries ordered left to right in table

General Non-imaging Diagnostics (34838.1)

Idiopathic ventricular fibrillation (42161)

Idiopathic ventricular fibrillation phenotypes (42167)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Idiopathic ventricular fibrillation (42161)
Phenotypes	Entries ordered left to right in table

Ventricular fibrillation (HP:0001663)	Sudden cardiac death (HP:0001645)	Syncope (HP:0001279)

Arrhythmia clinical tests (30854)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Idiopathic ventricular fibrillation (42161)
Clinical Tests	Entries ordered left to right in table

ECG diagnostics (30183.2)	Cardiac MRI (31429.2)	Holter monitor test (31432.3)
Signal averaged ECG (31437.2)	Exercise test - cardiac (31445.3)	Echocardiogram (29800.2)
Electrophysiological study (31449.2)	Sodium channel blocker challenge (31403.3)	Alcohol intake (30206.1)
Exercise status (31469.1)	Epinephrine challenge (31404.3)	Smoking status (31464.1)

Short QT syndrome (55487)

Short QT syndrome phenotypes (68118)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Short QT syndrome (55487)
Phenotypes	Entries ordered left to right in table

Shortened QT interval (HP:0012232)	Syncope (HP:0001279)	Palpitations (HP:0001962)
Cardiac arrest (HP:0001695)	Ventricular arrhythmia (HP:0004308)	Atrial fibrillation (HP:0005110)
Prolonged PR interval (HP:0012248)	Atrioventricular block (HP:0001678)	Abnormal ST segment (HP:0012249)
J wave (HP:0012272)	Abnormal T-wave (HP:0005135)	Bundle branch block (HP:0011710)
Periodic paralysis (HP:0003768)	Sensorineural hearing impairment (HP:0000407)	Seizures (HP:0001250)
Intellectual disability (HP:0001249)		

Short QT syndrome clinical tests (55488)

Level 3 Title	Cardiac arrhythmia (10952)
Level 4 Title	Short QT syndrome (55487)
Clinical Tests	Entries ordered left to right in table

ECG diagnostics (30183.2)	Cardiac MRI (31429.2)	Holter monitor test (31432.3)
Signal averaged ECG (31437.2)	Exercise test - cardiac (31445.3)	Echocardiogram (29800.2)
Electrophysiological study (31449.2)	Sodium channel blocker challenge (31403.3)	Alcohol intake (30206.1)
Exercise status (31469.1)	Epinephrine challenge (31404.3)	Smoking status (31464.1)

Cardiomyopathy (10953)

Arrhythmogenic Right Ventricular Cardiomyopathy (11025)

Cardiomyopathy phenotypes (28640)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Arrhythmogenic Right Ventricular Cardiomyopathy (11025)
Phenotypes	Entries ordered left to right in table

Cardiomyopathy (HP:0001638)	Hypertrophic cardiomyopathy (HP:0001639)	Dilated cardiomyopathy (HP:0001644)
Left ventricular noncompaction cardiomyopathy (HP:0011664)	Right ventricular cardiomyopathy (HP:0011663)	Restrictive cardiomyopathy (HP:0001723)
Arrhythmia (HP:0011675)	Hypertension (HP:0000822)	Congestive heart failure (HP:0001635)
Stroke (HP:0001297)	Syncope (HP:0001279)	Sudden cardiac death (HP:0001645)
Angina pectoris (HP:0001681)	Palpitations (HP:0001962)	Edema of the lower limbs (HP:0010741)
Dyspnea (HP:0002094)	Failure to thrive (HP:0001508)	Skeletal myopathy (HP:0003756)
Visual impairment (HP:0000505)	Hearing impairment (HP:0000365)	Diabetes mellitus (HP:0000819)
Intellectual disability (HP:0001249)	Increased nuchal translucency (HP:0010880)	

Cardiomyopathy clinical tests (30856)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Arrhythmogenic Right Ventricular Cardiomyopathy (11025)
Clinical Tests	Entries ordered left to right in table

Blood pressure (30245.1)	ECG diagnostics (30183.2)	Echocardiogram (29800.2)
Cardiac MRI (31429.2)	Holter monitor test (31432.3)	Doppler Diastolic function assessment (29051.2)
Exercise status (31469.1)	Alcohol intake (30206.1)	Exercise test (30214.1)
Smoking status (31464.1)		

Left Ventricular Noncompaction Cardiomyopathy (15044)

Cardiomyopathy phenotypes (28640)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Left Ventricular Noncompaction Cardiomyopathy (15044)
Phenotypes	Entries ordered left to right in table

Cardiomyopathy (HP:0001638)	Hypertrophic cardiomyopathy (HP:0001639)	Dilated cardiomyopathy (HP:0001644)
Left ventricular noncompaction cardiomyopathy (HP:0011664)	Right ventricular cardiomyopathy (HP:0011663)	Restrictive cardiomyopathy (HP:0001723)
Arrhythmia (HP:0011675)	Hypertension (HP:0000822)	Congestive heart failure (HP:0001635)
Stroke (HP:0001297)	Syncope (HP:0001279)	Sudden cardiac death (HP:0001645)
Angina pectoris (HP:0001681)	Palpitations (HP:0001962)	Edema of the lower limbs (HP:0010741)
Dyspnea (HP:0002094)	Failure to thrive (HP:0001508)	Skeletal myopathy (HP:0003756)
Visual impairment (HP:0000505)	Hearing impairment (HP:0000365)	Diabetes mellitus (HP:0000819)
Intellectual disability (HP:0001249)	Increased nuchal translucency (HP:0010880)	

Cardiomyopathy clinical tests (30856)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Left Ventricular Noncompaction Cardiomyopathy (15044)
Clinical Tests	Entries ordered left to right in table

Blood pressure (30245.1)	ECG diagnostics (30183.2)	Echocardiogram (29800.2)
Cardiac MRI (31429.2)	Holter monitor test (31432.3)	Doppler Diastolic function assessment (29051.2)
Exercise status (31469.1)	Alcohol intake (30206.1)	Exercise test (30214.1)
Smoking status (31464.1)		

Dilated Cardiomyopathy (31340)

Cardiomyopathy phenotypes (28640)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Dilated Cardiomyopathy (31340)
Phenotypes	Entries ordered left to right in table

Cardiomyopathy (HP:0001638)	Hypertrophic cardiomyopathy (HP:0001639)	Dilated cardiomyopathy (HP:0001644)
Left ventricular noncompaction cardiomyopathy (HP:0011664)	Right ventricular cardiomyopathy (HP:0011663)	Restrictive cardiomyopathy (HP:0001723)
Arrhythmia (HP:0011675)	Hypertension (HP:0000822)	Congestive heart failure (HP:0001635)
Stroke (HP:0001297)	Syncope (HP:0001279)	Sudden cardiac death (HP:0001645)
Angina pectoris (HP:0001681)	Palpitations (HP:0001962)	Edema of the lower limbs (HP:0010741)
Dyspnea (HP:0002094)	Failure to thrive (HP:0001508)	Skeletal myopathy (HP:0003756)
Visual impairment (HP:0000505)	Hearing impairment (HP:0000365)	Diabetes mellitus (HP:0000819)
Intellectual disability (HP:0001249)	Increased nuchal translucency (HP:0010880)	

Cardiomyopathy clinical tests (30856)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Dilated Cardiomyopathy (31340)
Clinical Tests	Entries ordered left to right in table

Blood pressure (30245.1)	ECG diagnostics (30183.2)	Echocardiogram (29800.2)
Cardiac MRI (31429.2)	Holter monitor test (31432.3)	Doppler Diastolic function assessment (29051.2)
Exercise status (31469.1)	Alcohol intake (30206.1)	Exercise test (30214.1)
Smoking status (31464.1)		

Dilated Cardiomyopathy and conduction defects (11027)

Cardiomyopathy phenotypes (28640)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Dilated Cardiomyopathy and conduction defects (11027)
Phenotypes	Entries ordered left to right in table

Cardiomyopathy (HP:0001638)	Hypertrophic cardiomyopathy (HP:0001639)	Dilated cardiomyopathy (HP:0001644)
Left ventricular noncompaction cardiomyopathy (HP:0011664)	Right ventricular cardiomyopathy (HP:0011663)	Restrictive cardiomyopathy (HP:0001723)
Arrhythmia (HP:0011675)	Hypertension (HP:0000822)	Congestive heart failure (HP:0001635)
Stroke (HP:0001297)	Syncope (HP:0001279)	Sudden cardiac death (HP:0001645)
Angina pectoris (HP:0001681)	Palpitations (HP:0001962)	Edema of the lower limbs (HP:0010741)
Dyspnea (HP:0002094)	Failure to thrive (HP:0001508)	Skeletal myopathy (HP:0003756)
Visual impairment (HP:0000505)	Hearing impairment (HP:0000365)	Diabetes mellitus (HP:0000819)
Intellectual disability (HP:0001249)	Increased nuchal translucency (HP:0010880)	

Cardiomyopathy clinical tests (30856)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Dilated Cardiomyopathy and conduction defects (11027)
Clinical Tests	Entries ordered left to right in table

Blood pressure (30245.1)	ECG diagnostics (30183.2)	Echocardiogram (29800.2)
Cardiac MRI (31429.2)	Holter monitor test (31432.3)	Doppler Diastolic function assessment (29051.2)
Exercise status (31469.1)	Alcohol intake (30206.1)	Exercise test (30214.1)
Smoking status (31464.1)		

Hypertrophic Cardiomyopathy (11028)

Cardiomyopathy phenotypes (28640)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Hypertrophic Cardiomyopathy (11028)
Phenotypes	Entries ordered left to right in table

Cardiomyopathy (HP:0001638)	Hypertrophic cardiomyopathy (HP:0001639)	Dilated cardiomyopathy (HP:0001644)
Left ventricular noncompaction cardiomyopathy (HP:0011664)	Right ventricular cardiomyopathy (HP:0011663)	Restrictive cardiomyopathy (HP:0001723)
Arrhythmia (HP:0011675)	Hypertension (HP:0000822)	Congestive heart failure (HP:0001635)
Stroke (HP:0001297)	Syncope (HP:0001279)	Sudden cardiac death (HP:0001645)
Angina pectoris (HP:0001681)	Palpitations (HP:0001962)	Edema of the lower limbs (HP:0010741)
Dyspnea (HP:0002094)	Failure to thrive (HP:0001508)	Skeletal myopathy (HP:0003756)
Visual impairment (HP:0000505)	Hearing impairment (HP:0000365)	Diabetes mellitus (HP:0000819)
Intellectual disability (HP:0001249)	Increased nuchal translucency (HP:0010880)	

Cardiomyopathy clinical tests (30856)

Level 3 Title	Cardiomyopathy (10953)
Level 4 Title	Hypertrophic Cardiomyopathy (11028)
Clinical Tests	Entries ordered left to right in table

Blood pressure (30245.1)	ECG diagnostics (30183.2)	Echocardiogram (29800.2)
Cardiac MRI (31429.2)	Holter monitor test (31432.3)	Doppler Diastolic function assessment (29051.2)
Exercise status (31469.1)	Alcohol intake (30206.1)	Exercise test (30214.1)
Smoking status (31464.1)		

Congenital heart disease (10954)

Familial congenital heart disease (42212)

Congenital heart disease phenotypes (28642)

Level 3 Title	Congenital heart disease (10954)
Level 4 Title	Familial congenital heart disease (42212)
Phenotypes	Entries ordered left to right in table

Abnormality of cardiac atrium (HP:0005120)	Abnormality of the atrial septum (HP:0011994)	Abnormality of the ventricular septum (HP:0010438)
Abnormal atrioventricular connection (HP:0011546)	Abnormal ventriculo-arterial connection (HP:0011563)	Abnormality of cardiac ventricle (HP:0001713)
Abnormality of the aortic valve (HP:0001646)	Abnormality of the pulmonary valve (HP:0001641)	Abnormality of the tricuspid valve (HP:0001702)
Abnormality of the mitral valve (HP:0001633)	Abnormality of the pulmonary vasculature (HP:0004930)	Abnormality of the coronary arteries (HP:0006704)
Abnormality of the aorta (HP:0001679)	Right atrial isomerism (HP:0011536)	Left atrial isomerism (HP:0011537)
Cardiomyopathy (HP:0001638)	Arrhythmia (HP:0011675)	Abnormal anatomic location of the heart (HP:0004307)
Increased nuchal translucency (HP:0010880)	Small for gestational age (HP:0001518)	Large for gestational age (HP:0001520)
Failure to thrive (HP:0001508)	Short stature (HP:0004322)	Tall stature (HP:0000098)
Microcephaly (HP:0000252)	Macrocephaly (HP:0000256)	Abnormal facial shape (HP:0001999)
Intellectual disability (HP:0001249)	Abnormal respiratory motile cilium physiology (HP:0012261)	Polysplenia (HP:0001748)
Asplenia (HP:0001746)	Ectopia of the spleen (HP:0010452)	Intestinal malrotation (HP:0002566)

Syndromic congenital heart disease (42213)

Congenital heart disease phenotypes (28642)

Level 3 Title	Congenital heart disease (10954)
Level 4 Title	Syndromic congenital heart disease (42213)
Phenotypes	Entries ordered left to right in table

Abnormality of cardiac atrium (HP:0005120)	Abnormality of the atrial septum (HP:0011994)	Abnormality of the ventricular septum (HP:0010438)
Abnormal atrioventricular connection (HP:0011546)	Abnormal ventriculo-arterial connection (HP:0011563)	Abnormality of cardiac ventricle (HP:0001713)
Abnormality of the aortic valve (HP:0001646)	Abnormality of the pulmonary valve (HP:0001641)	Abnormality of the tricuspid valve (HP:0001702)
Abnormality of the mitral valve (HP:0001633)	Abnormality of the pulmonary vasculature (HP:0004930)	Abnormality of the coronary arteries (HP:0006704)
Abnormality of the aorta (HP:0001679)	Right atrial isomerism (HP:0011536)	Left atrial isomerism (HP:0011537)
Cardiomyopathy (HP:0001638)	Arrhythmia (HP:0011675)	Abnormal anatomic location of the heart (HP:0004307)
Increased nuchal translucency (HP:0010880)	Small for gestational age (HP:0001518)	Large for gestational age (HP:0001520)
Failure to thrive (HP:0001508)	Short stature (HP:0004322)	Tall stature (HP:0000098)
Microcephaly (HP:0000252)	Macrocephaly (HP:0000256)	Abnormal facial shape (HP:0001999)
Intellectual disability (HP:0001249)	Abnormal respiratory motile cilium physiology (HP:0012261)	Polysplenia (HP:0001748)
Asplenia (HP:0001746)	Ectopia of the spleen (HP:0010452)	Intestinal malrotation (HP:0002566)

Lymphatic disorders (33334)

Meige disease (34328)

Meige disease phenotypes (33570)

Level 3 Title	Lymphatic disorders (33334)
Level 4 Title	Meige disease (34328)
Phenotypes	Entries ordered left to right in table

Lymphedema (HP:0001004)	Predominantly lower limb lymphedema (HP:0003550)	Edema of the upper limbs (HP:0010742)
Edema of the lower limbs (HP:0010741)	Edema of the dorsum of feet (HP:0012098)	Facial edema (HP:0000282)
Hydrops fetalis (HP:0001789)	Pleural effusion (HP:0002202)	Pulmonary lymphangiectasia (HP:0006521)
Pericardial effusion (HP:0001698)	Intestinal lymphangiectasia (HP:0002593)	Chronic diarrhea (HP:0002028)
Abnormality of the lymphatic system (HP:0100763)	Abnormality of the lymphatic vessels (HP:0100766)	Hypoplasia of lymphatic vessels (HP:0003759)
Hypertrophy of the lower limb (HP:0010496)	Hypertrophy of the upper limb (HP:0010484)	Arteriovenous malformation (HP:0100026)
Vascular skin abnormality (HP:0011276)	Cellulitis (HP:0100658)	Cleft palate (HP:0000175)
Abnormality of the eye (HP:0000478)	Distichiasis (HP:0009743)	Abnormality of cardiovascular system morphology (HP:0030680)
Intellectual disability (HP:0001249)	Ptosis (HP:0000508)	Immunodeficiency (HP:0002721)
Verrucae (HP:0200043)	Recurrent viral skin infections (HP:0011371)	Intellectual disability, mild (HP:0001256)
Intellectual disability, moderate (HP:0002342)	Intellectual disability, severe (HP:0010864)	Macrocephaly (HP:0000256)
Microcephaly (HP:0000252)	Hearing impairment (HP:0000365)	Myelodysplasia (HP:0002863)
Leukemia (HP:0001909)		

Meige disease clinical tests (33571)

Level 3 Title	Lymphatic disorders (33334)
Level 4 Title	Meige disease (34328)
Clinical Tests	Entries ordered left to right in table

Full Blood Count (30318.2)	Serum immunoglobulins (30338.2)	Liver biochemistry (30328.2)

Milroy disease (37604)

Milroy disease phenotypes (37747)

Level 3 Title	Lymphatic disorders (33334)
Level 4 Title	Milroy disease (37604)

Phenotypes

Entries ordered left to right in table

Lymphedema (HP:0001004)	Predominantly lower limb lymphedema (HP:0003550)	Edema of the lower limbs (HP:0010741)
Edema of the upper limbs (HP:0010742)	Edema of the dorsum of feet (HP:0012098)	Facial edema (HP:0000282)
Abnormality of the lymphatic system (HP:0100763)	Abnormality of the lymphatic vessels (HP:0100766)	Hypoplasia of lymphatic vessels (HP:0003759)
Cellulitis (HP:0100658)	Hydrops fetalis (HP:0001789)	Pleural effusion (HP:0002202)
Pulmonary lymphangiectasia (HP:0006521)	Pericardial effusion (HP:0001698)	Intestinal lymphangiectasia (HP:0002593)
Chronic diarrhea (HP:0002028)	Hypertrophy of the lower limb (HP:0010496)	Hypertrophy of the upper limb (HP:0010484)
Arteriovenous malformation (HP:0100026)	Vascular skin abnormality (HP:0011276)	Distichiasis (HP:0009743)
Varicose veins (HP:0002619)	Venous insufficiency (HP:0005293)	Hydrocele testis (HP:0000034)
Abnormality of cardiovascular system morphology (HP:0030680)	Cleft palate (HP:0000175)	Ptosis (HP:0000508)
Immunodeficiency (HP:0002721)	Verrucae (HP:0200043)	Recurrent viral skin infections (HP:0011371)
Abnormality of the eye (HP:0000478)	Intellectual disability (HP:0001249)	Intellectual disability, mild (HP:0001256)
Intellectual disability, moderate (HP:0002342)	Intellectual disability, severe (HP:0010864)	Microcephaly (HP:0000252)
Macrocephaly (HP:0000256)	Hearing impairment (HP:0000365)	Myelodysplasia (HP:0002863)
Leukemia (HP:0001909)		

Milroy disease clinical tests (37606)

<table>
<tr><td>Level 3 Title</td><td>Lymphatic disorders (33334)</td></tr>
<tr><td>Level 4 Title</td><td>Milroy disease (37604)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical Test Guidance:
Full blood count should include full differential
Liver biochemistry is requested for serum albumin
Primary immunodeficiency investigations refers to CD4+ CD8+
Entries ordered left to right in table

<table><tr><td>Serum immunoglobulins (30338.2)</td><td>Full Blood Count (30318.2)</td><td>Liver biochemistry (30328.2)</td></tr><tr><td>Primary immunodeficiency investigations (33165.1)</td><td></td><td></td></tr></table></td></tr>
</table>

Lymphoedema distichiasis (37612)

Milroy disease phenotypes (37747)

Level 3 Title	Lymphatic disorders (33334)
Level 4 Title	Lymphoedema distichiasis (37612)
Phenotypes	Entries ordered left to right in table

Lymphedema (HP:0001004)	Predominantly lower limb lymphedema (HP:0003550)	Edema of the lower limbs (HP:0010741)
Edema of the upper limbs (HP:0010742)	Edema of the dorsum of feet (HP:0012098)	Facial edema (HP:0000282)
Abnormality of the lymphatic system (HP:0100763)	Abnormality of the lymphatic vessels (HP:0100766)	Hypoplasia of lymphatic vessels (HP:0003759)
Cellulitis (HP:0100658)	Hydrops fetalis (HP:0001789)	Pleural effusion (HP:0002202)
Pulmonary lymphangiectasia (HP:0006521)	Pericardial effusion (HP:0001698)	Intestinal lymphangiectasia (HP:0002593)
Chronic diarrhea (HP:0002028)	Hypertrophy of the lower limb (HP:0010496)	Hypertrophy of the upper limb (HP:0010484)
Arteriovenous malformation (HP:0100026)	Vascular skin abnormality (HP:0011276)	Distichiasis (HP:0009743)
Varicose veins (HP:0002619)	Venous insufficiency (HP:0005293)	Hydrocele testis (HP:0000034)
Abnormality of cardiovascular system morphology (HP:0030680)	Cleft palate (HP:0000175)	Ptosis (HP:0000508)
Immunodeficiency (HP:0002721)	Verrucae (HP:0200043)	Recurrent viral skin infections (HP:0011371)
Abnormality of the eye (HP:0000478)	Intellectual disability (HP:0001249)	Intellectual disability, mild (HP:0001256)
Intellectual disability, moderate (HP:0002342)	Intellectual disability, severe (HP:0010864)	Microcephaly (HP:0000252)
Macrocephaly (HP:0000256)	Hearing impairment (HP:0000365)	Myelodysplasia (HP:0002863)
Leukemia (HP:0001909)		

Milroy disease clinical tests (37606)

<table>
<tr><td>Level 3 Title</td><td>Lymphatic disorders (33334)</td></tr>
<tr><td>Level 4 Title</td><td>Lymphoedema distichiasis (37612)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Full blood count should include full differential
Liver biochemistry is requested for serum albumin
Primary immunodeficiency investigations refers to CD4+ CD8+
Entries ordered left to right in table

<table><tr><td>Serum immunoglobulins (30338.2)</td><td>Full Blood Count (30318.2)</td><td>Liver biochemistry (30328.2)</td></tr><tr><td>Primary immunodeficiency investigations (33165.1)</td><td></td><td></td></tr></table></td></tr>
</table>

Lipoedema disease (55456)

Lipoedema disease phenotypes (68112)

Level 3 Title	Lymphatic disorders (33334)
Level 4 Title	Lipoedema disease (55456)
Phenotypes	Entries ordered left to right in table

Lipedema (HP:0100695)	Abnormality of the lower limb (HP:0002814)	Adipocyte hypertrophy (HP:0030759)
Abnormality of subcutaneous fat tissue (HP:0001001)	Increased body mass index (HP:0045083)	Abnormality of the upper limb (HP:0002817)
Bruising susceptibility (HP:0000978)	Pain (HP:0012531)	Hypothermia (HP:0002045)
Telangiectasia (HP:0001009)	Joint hypermobility (HP:0001382)	Lymphedema (HP:0001004)

Lipoedema disease clinical tests (55458)

Level 3 Title	Lymphatic disorders (33334)
Level 4 Title	Lipoedema disease (55456)
Clinical Tests	NB clinical test guidance: Additional body measurements refers to Height on presentation and Weight at presentation Entries ordered left to right in table

Additional body measurements (30247.2)

Primary lymphoedema (55517)

Primary lymphoedema phenotypes (68126)

Level 3 Title	Lymphatic disorders (33334)
Level 4 Title	Primary lymphoedema (55517)
Phenotypes	Entries ordered left to right in table

Lymphedema (HP:0001004)	Predominantly lower limb lymphedema (HP:0003550)	Edema of the lower limbs (HP:0010741)
Edema of the upper limbs (HP:0010742)	Edema of the dorsum of feet (HP:0012098)	Facial edema (HP:0000282)
Abnormality of the lymphatic system (HP:0100763)	Abnormality of the lymphatic vessels (HP:0100766)	Hypoplasia of lymphatic vessels (HP:0003759)
Cellulitis (HP:0100658)	Hydrops fetalis (HP:0001789)	Pleural effusion (HP:0002202)
Pulmonary lymphangiectasia (HP:0006521)	Pericardial effusion (HP:0001698)	Intestinal lymphangiectasia (HP:0002593)
Chronic diarrhea (HP:0002028)	Hypertrophy of the lower limb (HP:0010496)	Hypertrophy of the upper limb (HP:0010484)
Arteriovenous malformation (HP:0100026)	Vascular skin abnormality (HP:0011276)	Distichiasis (HP:0009743)
Varicose veins (HP:0002619)	Venous insufficiency (HP:0005293)	Hydrocele testis (HP:0000034)
Abnormality of cardiovascular system morphology (HP:0030680)	Cleft palate (HP:0000175)	Ptosis (HP:0000508)
Immunodeficiency (HP:0002721)	Verrucae (HP:0200043)	Recurrent viral skin infections (HP:0011371)
Abnormality of the eye (HP:0000478)	Intellectual disability (HP:0001249)	Intellectual disability, mild (HP:0001256)
Intellectual disability, moderate (HP:0002342)	Intellectual disability, severe (HP:0010864)	Microcephaly (HP:0000252)
Macrocephaly (HP:0000256)	Hearing impairment (HP:0000365)	Myelodysplasia (HP:0002863)
Leukemia (HP:0001909)		

Primary lymphoedema clinical tests (55518)

Level 3 Title	Lymphatic disorders (33334)
Level 4 Title	Primary lymphoedema (55517)
Clinical Tests	NB. Clinical test guidance: Full blood count should include full differential Liver biochemestry is requested for serum albumin Primary immunodeficiency investigations refers to CD4+ CD8+ Entries ordered left to right in table

Serum immunoglobulins (30338.2)	Full Blood Count (30318.2)	Liver biochemistry (30328.2)

Pulmonary heart disease (55662)

Pulmonary arterial hypertension (55499)

Pulmonary arterial hypertension phenotypes (68120)

Level 3 Title	Pulmonary heart disease (55662)
Level 4 Title	Pulmonary arterial hypertension (55499)
Phenotypes	Entries ordered left to right in table

Pulmonary arterial hypertension (HP:0002092)	Dyspnea (HP:0002094)	Syncope (HP:0001279)
Right ventricular failure (HP:0001708)	Decreased DLCO (HP:0045051)	Left ventricular diastolic dysfunction (HP:0025168)
Interstitial pulmonary abnormality (HP:0006530)	Emphysema (HP:0002097)	Pulmonary embolism (HP:0002204)
Pulmonary arteriovenous malformation (HP:0006548)	Abnormality of the thyroid gland (HP:0000820)	Left-to-right shunt (HP:0012382)
Obstructive deficit on pulmonary function testing (HP:0030877)	Pulmonary capillary hemangiomatosis (HP:0005954)	Portal hypertension (HP:0001409)

Pulmonary arterial hypertension clinical tests (55500)

Level 3 Title	Pulmonary heart disease (55662)
Level 4 Title	Pulmonary arterial hypertension (55499)
Clinical Tests	Entries ordered left to right in table

Forced vital capacity (30180.2)

Ciliopathies (10963)

Congenital malformations caused by ciliopathies (15091)

Bardet-Biedl Syndrome (11046)

Bardet-Biedl Syndrome phenotypes (28526)

Level 3 Title	Congenital malformations caused by ciliopathies (15091)
Level 4 Title	Bardet-Biedl Syndrome (11046)
Phenotypes	Entries ordered left to right in table

Obesity (HP:0001513)	Neurodevelopmental delay (HP:0012758)	Intellectual disability (HP:0001249)
Behavioral abnormality (HP:0000708)	Rod-cone dystrophy (HP:0000510)	Retinal dystrophy (HP:0000556)
Optic atrophy (HP:0000648)	Astigmatism (HP:0000483)	Strabismus (HP:0000486)
Cataract (HP:0000518)	Brachydactyly syndrome (HP:0001156)	Polydactyly (HP:0010442)
Syndactyly (HP:0001159)	Abnormality of male external genitalia (HP:0000032)	Abnormality of the female genitalia (HP:0010460)
Glucose intolerance (HP:0000833)	Diabetes mellitus (HP:0000819)	Diabetes insipidus (HP:0000873)
Chronic kidney disease (HP:0012622)	Abnormal renal morphology (HP:0012210)	Cardiomyopathy (HP:0001638)
Abnormality of cardiovascular system morphology (HP:0030680)	Abnormality of the ear (HP:0000598)	Hyperlipidemia (HP:0003077)
Abnormality of the middle ear ossicles (HP:0004452)	Hypertension (HP:0000822)	Otitis media (HP:0000388)
Sensorineural hearing impairment (HP:0000407)	Conductive hearing impairment (HP:0000405)	Dental crowding (HP:0000678)
High palate (HP:0000218)	Hepatic fibrosis (HP:0001395)	Hepatic steatosis (HP:0001397)
Abnormality of the intrahepatic bile duct (HP:0011040)	Elevated hepatic transaminases (HP:0002910)	Autonomic bladder dysfunction (HP:0005341)
Ataxia (HP:0001251)	Spasticity (HP:0001257)	Aganglionic megacolon (HP:0002251)
Anosmia (HP:0000458)		

Bardet-Biedl Syndrome clinical tests (30868)

Level 3 Title	Congenital malformations caused by ciliopathies (15091)
Level 4 Title	Bardet-Biedl Syndrome (11046)
Clinical Tests	NB. Clinical test guidance: Imaging Diagnostics refers to hand, facial and other relevant medical photographs Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Joubert syndrome (36478)

Joubert syndrome phenotypes (36634)

Level 3 Title	Congenital malformations caused by ciliopathies (15091)
Level 4 Title	Joubert syndrome (36478)
Phenotypes	Entries ordered left to right in table

Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)
Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)	Microcephaly (HP:0000252)
Macrocephaly (HP:0000256)	Generalized hypotonia (HP:0001290)	Ataxia (HP:0001251)
Molar tooth sign on MRI (HP:0002419)	Cerebellar vermis hypoplasia (HP:0001320)	Occipital encephalocele (HP:0002085)
Polymicrogyria (HP:0002126)	Neonatal breathing dysregulation (HP:0002790)	Oculomotor apraxia (HP:0000657)
Coloboma (HP:0000589)	Retinal dystrophy (HP:0000556)	Abnormal facial shape (HP:0001999)
Accessory oral frenulum (HP:0000191)	Hamartoma of tongue (HP:0011802)	Cleft palate (HP:0000175)
Abnormality of cardiovascular system morphology (HP:0030680)	Hyperechogenic kidneys (HP:0004719)	Multiple renal cysts (HP:0005562)
Hypertension (HP:0000822)	Chronic kidney disease (HP:0012622)	Hepatic fibrosis (HP:0001395)
Elevated hepatic transaminases (HP:0002910)	Preaxial hand polydactyly (HP:0001177)	Postaxial hand polydactyly (HP:0001162)
Mesoaxial hand polydactyly (HP:0006159)	Preaxial foot polydactyly (HP:0001841)	Postaxial foot polydactyly (HP:0001830)
Mesoaxial foot polydactyly (HP:0010112)	Short stature (HP:0004322)	Obesity (HP:0001513)

Joubert syndrome clinical tests (36480)

Level 3 Title	Congenital malformations caused by ciliopathies (15091)
Level 4 Title	Joubert syndrome (36478)
Clinical Tests	N. Clinical test guidance: General imaging diagnostics refers to medical photographs and MRI brain Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Rare multisystem ciliopathy disorders (36488)

Joubert syndrome phenotypes (36634)

Level 3 Title	Congenital malformations caused by ciliopathies (15091)
Level 4 Title	Rare multisystem ciliopathy disorders (36488)
Phenotypes	Entries ordered left to right in table

Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)
Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)	Microcephaly (HP:0000252)
Macrocephaly (HP:0000256)	Generalized hypotonia (HP:0001290)	Ataxia (HP:0001251)
Molar tooth sign on MRI (HP:0002419)	Cerebellar vermis hypoplasia (HP:0001320)	Occipital encephalocele (HP:0002085)
Polymicrogyria (HP:0002126)	Neonatal breathing dysregulation (HP:0002790)	Oculomotor apraxia (HP:0000657)
Coloboma (HP:0000589)	Retinal dystrophy (HP:0000556)	Abnormal facial shape (HP:0001999)
Accessory oral frenulum (HP:0000191)	Hamartoma of tongue (HP:0011802)	Cleft palate (HP:0000175)
Abnormality of cardiovascular system morphology (HP:0030680)	Hyperechogenic kidneys (HP:0004719)	Multiple renal cysts (HP:0005562)
Hypertension (HP:0000822)	Chronic kidney disease (HP:0012622)	Hepatic fibrosis (HP:0001395)
Elevated hepatic transaminases (HP:0002910)	Preaxial hand polydactyly (HP:0001177)	Postaxial hand polydactyly (HP:0001162)
Mesoaxial hand polydactyly (HP:0006159)	Preaxial foot polydactyly (HP:0001841)	Postaxial foot polydactyly (HP:0001830)
Mesoaxial foot polydactyly (HP:0010112)	Short stature (HP:0004322)	Obesity (HP:0001513)

Joubert syndrome clinical tests (36480)

Level 3 Title	Congenital malformations caused by ciliopathies (15091)
Level 4 Title	Rare multisystem ciliopathy disorders (36488)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to medical photographs and MRI brain Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Respiratory ciliopathies (15092)

Primary ciliary dyskinesia (11047)

Primary ciliary dyskinesia phenotypes (27791)

Level 3 Title	Respiratory ciliopathies (15092)
Level 4 Title	Primary ciliary dyskinesia (11047)
Phenotypes	Entries ordered left to right in table

Neonatal respiratory distress (HP:0002643)	Cough (HP:0012735)	Recurrent sinopulmonary infections (HP:0005425)
Bronchiectasis (HP:0002110)	Chronic otitis media (HP:0000389)	Chronic bronchitis (HP:0004469)
Nasal obstruction (HP:0001742)	Chronic sinusitis (HP:0011109)	Infertility (HP:0000789)
Decreased fertility (HP:0000144)	Abnormality of cardiovascular system morphology (HP:0030680)	Dextrocardia (HP:0001651)
Abnormal anatomic location of the heart (HP:0004307)	Right atrial isomerism (HP:0011536)	Left atrial isomerism (HP:0011537)
Atrial septal defect (HP:0001631)	Complete atrioventricular canal defect (HP:0001674)	Ventricular septal defect (HP:0001629)
Tetralogy of Fallot (HP:0001636)	Situs inversus totalis (HP:0001696)	Polysplenia (HP:0001748)
Asplenia (HP:0001746)	Ectopia of the spleen (HP:0010452)	Intestinal malrotation (HP:0002566)
Extrahepatic biliary duct atresia (HP:0005242)	Aplasia/Hypoplasia of the corpus callosum (HP:0007370)	Hydrocephalus (HP:0000238)

Primary ciliary dyskinesia clinical tests (30869)

<table>
<tr><td>Level 3 Title</td><td>Respiratory ciliopathies (15092)</td></tr>
<tr><td>Level 4 Title</td><td>Primary ciliary dyskinesia (11047)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Culture refers to sputum cultures
Imaging diagnostics refers to chest CT, chest x-ray and/or abdominal ultrasound for laterality defects
Entries ordered left to right in table

<table><tr><td>Culture (28285.2)</td><td>Nasal Cilia Imaging (29830.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr></table></td></tr>
</table>

Non-CF bronchiectasis (11048)

Non-CF bronchiectasis phenotypes (29227)

Level 3 Title	Respiratory ciliopathies (15092)
Level 4 Title	Non-CF bronchiectasis (11048)
Phenotypes	Entries ordered left to right in table

Chronic lung disease (HP:0006528)	Recurrent respiratory infections (HP:0002205)	Recurrent bronchopulmonary infections (HP:0006538)
Recurrent pneumonia (HP:0006532)	Recurrent mycobacterial infections (HP:0011274)	Abnormality of the bronchi (HP:0002109)
Bronchiectasis (HP:0002110)	Asthma (HP:0002099)	Pulmonary fibrosis (HP:0002206)
Bronchiolitis obliterans (HP:0011946)	Aspiration (HP:0002835)	Hemoptysis (HP:0002105)
Sinusitis (HP:0000246)	Otitis media (HP:0000388)	Bronchial cartilage hypoplasia (HP:0006539)
Tracheobronchmegaly (HP:0010776)	Tracheobronchomalacia (HP:0002786)	Tracheoesophageal fistula (HP:0002575)
Pulmonary sequestration (HP:0100632)	Abnormality of the ribs (HP:0000772)	Immunodeficiency (HP:0002721)
Decreased antibody level in blood (HP:0004313)	Specific antibody deficiency (HP:0012475)	Failure to thrive (HP:0001508)
Hepatomegaly (HP:0002240)	Biliary cirrhosis (HP:0002613)	Abnormality of the pancreas (HP:0001732)
Exocrine pancreatic insufficiency (HP:0001738)	Malabsorption (HP:0002024)	Achalasia (HP:0002571)
Gastroesophageal reflux (HP:0002020)	Meconium ileus (HP:0004401)	Rectal prolapse (HP:0002035)
Cor pulmonale (HP:0001648)	Yellow nails (HP:0011367)	Male infertility (HP:0003251)
Elevated sweat chloride (HP:0012236)	Hypercalciuria (HP:0002150)	

Non-CF clinical tests (30870)

<table>
<tr><td>Level 3 Title</td><td>Respiratory ciliopathies (15092)</td></tr>
<tr><td>Level 4 Title</td><td>Non-CF bronchiectasis (11048)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Culture refers to sputum cultures
Imaging diagnostics refers to chest CT and chest x-ray
Entries ordered left to right in table

<table><tr><td>Serum immunoglobulins (30338.2)</td><td>Microbiology antibodies (33173.1)</td><td>Culture (28285.2)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td>Smoking status (31464.1)</td><td></td></tr></table></td></tr>
</table>

Dermatological disorders (10956)

Atopy (15084)

Severe multi-system atopic disease with high IgE (15085)

Atopic phenotypes (29224)

Level 3 Title	Atopy (15084)
Level 4 Title	Severe multi-system atopic disease with high IgE (15085)
Phenotypes	Entries ordered left to right in table

Eczema (HP:0000964)	Increased IgE level (HP:0003212)	Allergy (HP:0012393)
Cow milk allergy (HP:0100327)	Seasonal allergy (HP:0012395)	Allergic rhinitis (HP:0003193)
Chronic rhinitis (HP:0002257)	Asthma (HP:0002099)	Recurrent bacterial skin infections (HP:0005406)
Recurrent viral skin infections (HP:0011371)	Recurrent cutaneous fungal infections (HP:0011370)	Recurrent bacterial infections (HP:0002718)
Recurrent viral infections (HP:0004429)	Recurrent fungal infections (HP:0002841)	

Atopic clinical tests (30863)

Level 3 Title	Atopy (15084)
Level 4 Title	Severe multi-system atopic disease with high IgE (15085)
Clinical Tests	Entries ordered left to right in table

Full Blood Count (30318.2)	Serum immunoglobulins (30338.2)

Autoimmune skin disorders (33336)

Generalised pustular psoriasis (33646)

Generalised pustular psoriasis phenotypes (33952)

Level 3 Title	Autoimmune skin disorders (33336)
Level 4 Title	Generalised pustular psoriasis (33646)
Phenotypes	Entries ordered left to right in table

Psoriasis (HP:0003765)	Pustule (HP:0200039)	Episodic fatigue (HP:0012431)
Episodic fever (HP:0001954)	Elevated C-reactive protein level (HP:0011227)	Abnormal leukocyte count (HP:0011893)
Abnormality of the thyroid gland (HP:0000820)	Diabetes mellitus (HP:0000819)	Elevated mean arterial pressure (HP:0004972)
Celiac disease (HP:0002608)	Coronary artery disease (HP:0001677)	Palmoplantar pustulosis (HP:0100847)
Subungual hyperkeratosis (HP:0008392)	Recurrent loss of toenails and fingernails (HP:0008390)	Osteolysis (HP:0002797)
Arthralgia (HP:0002829)	Abnormality of the palm (HP:0100871)	Abnormality of the plantar skin of foot (HP:0100872)
Nail dystrophy (HP:0008404)		

Generalised pustular psoriasis clinical tests (33648)

Level 3 Title	Autoimmune skin disorders (33336)
Level 4 Title	Generalised pustular psoriasis (33646)
Clinical Tests	Entries ordered left to right in table

Inflammatory markers (33156.1)	Full Blood Count (30318.2)	Smoking status (31464.1)

Ectodermal dysplasias (33338)

Ectodermal dysplasia without a known gene mutation (33699)

Ectodermal dysplasia without a known gene mutation phenotypes (33733)

Level 3 Title	Ectodermal dysplasias (33338)
Level 4 Title	Ectodermal dysplasia without a known gene mutation (33699)
Phenotypes	Entries ordered left to right in table

Ectodermal dysplasia (HP:0000968)	Dry skin (HP:0000958)	Hypopigmentation of the skin (HP:0001010)
Hyperpigmentation of the skin (HP:0000953)	Periorbital wrinkles (HP:0000607)	Fragile skin (HP:0001030)
Palmoplantar hyperkeratosis (HP:0000972)	Hypohidrosis (HP:0000966)	Hyperhidrosis (HP:0000975)
Heat intolerance (HP:0002046)	Nail dysplasia (HP:0002164)	Anonychia (HP:0001798)
Hypodontia (HP:0000668)	Abnormality of dental morphology (HP:0006482)	Hypoplasia of dental enamel (HP:0006297)
Sparse hair (HP:0008070)	Abnormality of hair texture (HP:0010719)	Aplasia/Hypoplasia of the nipples (HP:0006709)
Ectrodactyly (HP:0100257)	Camptodactyly (HP:0012385)	Syndactyly (HP:0001159)
Photophobia (HP:0000613)	Ankyloblepharon (HP:0009755)	Lacrimal duct atresia (HP:0000564)
Bifid uvula (HP:0000193)	Cleft palate (HP:0000175)	Cleft upper lip (HP:0000204)
Conductive hearing impairment (HP:0000405)	Sensorineural hearing impairment (HP:0000407)	Global developmental delay (HP:0001263)
Intellectual disability (HP:0001249)	Abnormal nasal morphology (HP:0005105)	External ear malformation (HP:0008572)
Abnormal external genitalia (HP:0000811)	Recurrent infections (HP:0002719)	Immunodeficiency (HP:0002721)
Neoplasm of the skin (HP:0008069)	Microcephaly (HP:0000252)	

Ectodermal dysplasia without a known gene mutation clinical tests (33738)

Level 3 Title	Ectodermal dysplasias (33338)
Level 4 Title	Ectodermal dysplasia without a known gene mutation (33699)
Clinical Tests	Entries ordered left to right in table Serum immunoglobulins (30338.2)

Ichthyoses (33340)

Autosomal recessive congenital ichthyosis (33700)

Autosomal recessive congenital ichthyosis phenotypes (33950)

Level 3 Title	Ichthyoses (33340)
Level 4 Title	Autosomal recessive congenital ichthyosis (33700)
Phenotypes	Entries ordered left to right in table

Congenital ichthyosiform erythroderma (HP:0007431)	Congenital nonbullous ichthyosiform erythroderma (HP:0007479)	Ichthyosis (HP:0008064)
Palmoplantar hyperkeratosis (HP:0000972)	Alopecia (HP:0001596)	Hypotrichosis (HP:0001006)
Abnormality of hair texture (HP:0010719)	Subungual hyperkeratosis (HP:0008392)	Small nail (HP:0001792)
Nail dystrophy (HP:0008404)	Ectropion (HP:0000656)	Eclabion (HP:0012472)
Abnormality of the nares (HP:0005288)	Abnormality of the outer ear (HP:0000356)	Carious teeth (HP:0000670)
Open mouth (HP:0000194)	Abnormality of the teeth (HP:0000164)	Gingivitis (HP:0000230)
Anhidrosis (HP:0000970)	Hypohidrosis (HP:0000966)	Pruritus (HP:0000989)

Keratodermas (33342)

Palmoplantar keratoderma and erythrokeratodermas (33701)

Palmoplantar keratoderma and erythrokeratodermas phenotypes (33951)

Level 3 Title	Keratodermas (33342)
Level 4 Title	Palmoplantar keratoderma and erythrokeratodermas (33701)
Phenotypes	Entries ordered left to right in table

Diffuse palmoplantar keratoderma (HP:0007435)	Palmoplantar keratoderma (HP:0000982)	Punctate palmoplantar hyperkeratosis (HP:0007530)
Honeycomb palmoplantar keratoderma (HP:0007465)	Follicular hyperkeratosis (HP:0007502)	Hyperkeratosis with erythema (HP:0007390)
Skin plaque (HP:0200035)	Abnormal blistering of the skin (HP:0008066)	Palmoplantar blistering (HP:0007446)
Deep plantar creases (HP:0001869)	Palmoplantar hyperhidrosis (HP:0007410)	Palmar pits (HP:0010610)
Fragile skin (HP:0001030)	Milia (HP:0001056)	Epidermal cyst (HP:0200040)
Steatocystoma multiplex (HP:0012035)	Erythema (HP:0010783)	Blotching pigmentation of the skin (HP:0007610)
Recurrent fungal infections (HP:0002841)	Perioral hyperpigmentation (HP:0010802)	Abnormality of oral mucosa (HP:0011830)
Periodontitis (HP:0000704)	Carious teeth (HP:0000670)	Hypodontia (HP:0000668)
Abnormality of dental enamel (HP:0000682)	Hypotrichosis (HP:0001006)	Hypertrichosis (HP:0000998)
Alopecia (HP:0001596)	Pili torti (HP:0003777)	Coarse hair (HP:0002208)
Curly hair (HP:0002212)	Dry hair (HP:0011359)	Fine hair (HP:0002213)
Uncombable hair (HP:0030056)	Woolly hair (HP:0002224)	Woolly scalp hair (HP:0040149)
Brittle hair (HP:0002299)	Brittle scalp hair (HP:0004779)	Nail dystrophy (HP:0008404)
Leukonychia (HP:0001820)	Subungual hyperkeratosis (HP:0008392)	Thin nail (HP:0001816)

Familial disseminated superficial actinic porokeratosis (37644)

Familial disseminated superficial actinic porokeratosis phenotypes (37645)

Level 3 Title	Keratodermas (33342)
Level 4 Title	Familial disseminated superficial actinic porokeratosis (37644)
Phenotypes	Entries ordered left to right in table Porokeratosis (HP:0200044)

Familial disseminated superficial actinic porokeratosis clinical tests (37646)

Level 3 Title	Keratodermas (33342)
Level 4 Title	Familial disseminated superficial actinic porokeratosis (37644)
Clinical Tests	NB. Clinical test guidance: General biopsy refers to skin biopsy Entries ordered left to right in table General Biopsy (33614.1)

Neurocutaneous disorders (33344)

Undiagnosed neurocutaneous disorders (33686)

Undiagnosed neurocutaneous disorders phenotypes (33687)

Level 3 Title	Neurocutaneous disorders (33344)
Level 4 Title	Undiagnosed neurocutaneous disorders (33686)
Phenotypes	Entries ordered left to right in table

Abnormality of skin pigmentation (HP:0001000)	Cafe-au-lait spot (HP:0000957)	Hypopigmentation of the skin (HP:0001010)
Freckling (HP:0001480)	Abnormality of hair pigmentation (HP:0009887)	Seizures (HP:0001250)
Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)	Scoliosis (HP:0002650)
Abnormality of the skeletal system (HP:0000924)	Abnormality of the endocrine system (HP:0000818)	Abnormal heart morphology (HP:0001627)
Abnormal facial shape (HP:0001999)	Abnormality of the cerebrum (HP:0002060)	Abnormality of the cerebellum (HP:0001317)
Abnormality of the eye (HP:0000478)	Abnormal renal morphology (HP:0012210)	

Undiagnosed neurocutaneous disorders clinical tests (33980)

Level 3 Title	Neurocutaneous disorders (33344)
Level 4 Title	Undiagnosed neurocutaneous disorders (33686)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Skin adnexa disorders (36587)

Familial cicatricial alopecia (36588)

Familial cicatricial alopecia phenotypes (36636)

Level 3 Title	Skin adnexa disorders (36587)
Level 4 Title	Familial cicatricial alopecia (36588)
Phenotypes	Entries ordered left to right in table

Scarring alopecia of scalp (HP:0004552)	Temporal hypotrichosis (HP:0004524)	Congenital posterior occipital alopecia (HP:0007534)
Sparse and thin eyebrow (HP:0000535)	Loss of eyelashes (HP:0011457)	Sparse facial hair (HP:0007464)
Sparse axillary hair (HP:0002215)	Sparse pubic hair (HP:0002225)	Sparse body hair (HP:0002231)
Abnormality of the nail (HP:0001597)	High anterior hairline (HP:0009890)	

Familial cicatricial alopecia clinical tests (36590)

Level 3 Title	Skin adnexa disorders (36587)
Level 4 Title	Familial cicatricial alopecia (36588)
Clinical Tests	Entries ordered left to right in table

Smoking status (31464.1)	Alcohol intake (30206.1)

Familial hidradenitis suppurativa (41844)

Familial hidradenitis suppurativa phenotypes (42229)

Level 3 Title	Skin adnexa disorders (36587)
Level 4 Title	Familial hidradenitis suppurativa (41844)
Phenotypes	Entries ordered left to right in table Recurrent cutaneous abscess formation (HP:0100838)

Familial hidradenitis suppurativa clinical tests (41846)

Level 3 Title	Skin adnexa disorders (36587)
Level 4 Title	Familial hidradenitis suppurativa (41844)
Clinical Tests	Clinical testing guidance: General Imaging Diagnostics refers to refers to medical photographs where relevant Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Non-syndromic hypotrichosis (36849)

Non-syndromic hypotrichosis phenotypes (36905)

Level 3 Title	Skin adnexa disorders (36587)
Level 4 Title	Non-syndromic hypotrichosis (36849)

Phenotypes

Entries ordered left to right in table

Absent hair (HP:0002298)	Alopecia totalis (HP:0007418)	Congenital alopecia totalis (HP:0005597)
Sparse scalp hair (HP:0002209)	Hypotrichosis of the scalp (HP:0004782)	Progressive hypotrichosis (HP:0002296)
Slow-growing scalp hair (HP:0100038)	Abnormal hair quantity (HP:0011362)	Abnormal hair pattern (HP:0010720)
Congenital abnormal hair pattern (HP:0011361)	Congenital posterior occipital alopecia (HP:0007534)	Abnormality of the hairline (HP:0009553)
Abnormality of the frontal hairline (HP:0000599)	Abnormality of the posterior hairline (HP:0030141)	Sparse anterior scalp hair (HP:0004768)
Temporal hypotrichosis (HP:0004524)	Absent eyebrow (HP:0002223)	Aplasia/Hypoplasia of the eyebrow (HP:0100840)
Sparse and thin eyebrow (HP:0000535)	Sparse lateral eyebrow (HP:0005338)	Absent eyelashes (HP:0000561)
Sparse eyelashes (HP:0000653)	Abnormality of the eyelashes (HP:0000499)	Abnormality of hair texture (HP:0010719)
Brittle scalp hair (HP:0004779)	Fine hair (HP:0002213)	Woolly scalp hair (HP:0040149)
Uncombable hair (HP:0030056)	Abnormal hair laboratory examination (HP:0003328)	Pili canaliculi (HP:0002235)
Pili torti (HP:0003777)	Trichodysplasia (HP:0002552)	Trichorrhexis nodosa (HP:0009886)
Alopecia universalis (HP:0002289)	Generalized hypotrichosis (HP:0004528)	Coarse hair (HP:0002208)
Erythema (HP:0010783)	Pruritus (HP:0000989)	Fair hair (HP:0002286)
Generalized papillary lesions (HP:0007482)	Follicular hyperkeratosis (HP:0007502)	Sparse body hair (HP:0002231)

Non-syndromic hypotrichosis clinical tests (36864)

Level 3 Title	Skin adnexa disorders (36587)
Level 4 Title	Non-syndromic hypotrichosis (36849)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to medical photographs where relevant Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Skin fragility disorders (33346)

Epidermolysis bullosa (33684)

Epidermolysis bullosa phenotypes (33685)

Level 3 Title	Skin fragility disorders (33346)
Level 4 Title	Epidermolysis bullosa (33684)
Phenotypes	Entries ordered left to right in table

Abnormal blistering of the skin (HP:0008066)	Palmoplantar blistering (HP:0007446)	Palmoplantar keratoderma (HP:0000982)
Skin erosion (HP:0200041)	Milia (HP:0001056)	Atrophic scars (HP:0001075)
Fragile skin (HP:0001030)	Mitten deformity (HP:0004057)	Poikiloderma (HP:0001029)
Cutaneous photosensitivity (HP:0000992)	Hyperpigmentation of the skin (HP:0000953)	Nail dystrophy (HP:0008404)
Anonychia (HP:0001798)	Alopecia of scalp (HP:0002293)	Abnormality of hair texture (HP:0010719)
Glomerulonephritis (HP:0000099)	Renal insufficiency (HP:0000083)	Urethral stricture (HP:0012227)
Cardiomyopathy (HP:0001638)	Delayed puberty (HP:0000823)	Osteoporosis (HP:0000939)
Anemia (HP:0001903)	Growth delay (HP:0001510)	Failure to thrive (HP:0001508)
Gingival overgrowth (HP:0000212)	Ankyloglossia (HP:0010296)	Hypoplasia of dental enamel (HP:0006297)
Colitis (HP:0002583)	Esophagitis (HP:0100633)	Ectropion (HP:0000656)
Blepharitis (HP:0000498)	Corneal erosion (HP:0200020)	Interstitial pulmonary abnormality (HP:0006530)
Recurrent infections (HP:0002719)	Abnormality of the outer ear (HP:0000356)	Abnormality of the nasal alae (HP:0000429)
Flexion contracture (HP:0001371)	Squamous cell carcinoma (HP:0002860)	Melanoma (HP:0002861)
Basal cell carcinoma (HP:0002671)		

Peeling skin syndrome (36540)

Peeling skin syndrome phenotypes (36637)

Level 3 Title	Skin fragility disorders (33346)
Level 4 Title	Peeling skin syndrome (36540)
Phenotypes	Entries ordered left to right in table

Erythema (HP:0010783)	Skin erosion (HP:0200041)	Pruritus (HP:0000989)
Abnormal blistering of the skin (HP:0008066)	Ichthyosis (HP:0008064)	Hyperkeratosis (HP:0000962)
Lichenification (HP:0100725)	Hypohidrosis (HP:0000966)	Hyperhidrosis (HP:0000975)
Leukonychia (HP:0001820)	Punctate palmoplantar hyperkeratosis (HP:0007530)	Cheilitis (HP:0100825)
Thick nail (HP:0001805)	Palmoplantar keratoderma (HP:0000982)	Atopic dermatitis (HP:0001047)
Increased IgE level (HP:0003212)	Eosinophilia (HP:0001880)	Urticaria (HP:0001025)
Failure to thrive (HP:0001508)	Recurrent bacterial skin infections (HP:0005406)	Erythroderma (HP:0001019)
Angioedema (HP:0100665)	Allergy (HP:0012393)	Hypotrichosis (HP:0001006)

Peeling skin syndrome clinical tests (36542)

Level 3 Title	Skin fragility disorders (33346)
Level 4 Title	Peeling skin syndrome (36540)
Clinical Tests	NB. Clinical test guidance General imaging diagnostics refers to medical photographs Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Erythropoietic protoporphyria, mild variant (11037)

Erythropoietic protoporphyria phenotypes (29222)

Level 3 Title	Sun-exposure related conditions (10958)
Level 4 Title	Erythropoietic protoporphyria, mild variant (11037)
Phenotypes	Entries ordered left to right in table

Increased erythrocyte protoporphyrin concentration (HP:0012187)	Cutaneous photosensitivity (HP:0000992)	Palmoplantar keratoderma (HP:0000982)
Edema (HP:0000969)	Microcytic anemia (HP:0001935)	Biliary tract abnormality (HP:0001080)
Cholelithiasis (HP:0001081)	Hepatic failure (HP:0001399)	Cirrhosis (HP:0001394)
Cholestasis (HP:0001396)	Elevated hepatic transaminases (HP:0002910)	

Erythropoietic protoporphyria, mild variant clinical tests (30859)

Level 3 Title	Sun-exposure related conditions (10958)
Level 4 Title	Erythropoietic protoporphyria, mild variant (11037)
Clinical Tests	Entries ordered left to right in table

Porphyria investigations (33164.1)

Hydroa vacciniforme (15083)

Hydroa Vacciniforme phenotypes (29223)

Level 3 Title	Sun-exposure related conditions (10958)
Level 4 Title	Hydroa vacciniforme (15083)
Phenotypes	Entries ordered left to right in table

Cutaneous photosensitivity (HP:0000992)	Lymphadenopathy (HP:0002716)	Lymphoma (HP:0002665)
Photophobia (HP:0000613)	Chemosis (HP:0012375)	Keratoconjunctivitis (HP:0001096)
Corneal ulceration (HP:0012804)	Iritis (HP:0001101)	Uveitis (HP:0000554)
Episcleritis (HP:0100534)	Recurrent aphthous stomatitis (HP:0011107)	Fever (HP:0001945)
Weight loss (HP:0001824)	Hepatomegaly (HP:0002240)	Splenomegaly (HP:0001744)
Elevated hepatic transaminases (HP:0002910)	Leukopenia (HP:0001882)	Thrombocytopenia (HP:0001873)

Hydroa vacciniforme clinical tests (30862)

Level 3 Title	Sun-exposure related conditions (10958)
Level 4 Title	Hydroa vacciniforme (15083)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to skin biopsy result Non-imaging diagnostics refers to Phototest response to UVA Virology refers to EBV viral load in blood Entries ordered left to right in table

Virology (33171.1)	General Biopsy (33614.1)	General Non-imaging Diagnostics (34838.1)

Dysmorphic and congenital abnormality syndromes (10959)

Kabuki (28664)

Kabuki syndrome (10960)

Kabuki phenotypes (27785)

Level 3 Title	Kabuki (28664)
Level 4 Title	Kabuki syndrome (10960)
Phenotypes	Entries ordered left to right in table

Short stature (HP:0004322)	Failure to thrive in infancy (HP:0001531)	Microcephaly (HP:0000252)
Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)	Seizures (HP:0001250)
Protruding ear (HP:0000411)	Long palpebral fissure (HP:0000637)	Eversion of lateral third of lower eyelids (HP:0007655)
Highly arched eyebrow (HP:0002553)	Sparse lateral eyebrow (HP:0005338)	Ptosis (HP:0000508)
Short columella (HP:0002000)	Depressed nasal tip (HP:0000437)	Cleft palate (HP:0000175)
Cleft upper lip (HP:0000204)	Hypodontia (HP:0000668)	Sensorineural hearing impairment (HP:0000407)
Conductive hearing impairment (HP:0000405)	Prominent fingertip pads (HP:0001212)	Abnormal heart morphology (HP:0001627)
Ventricular septal defect (HP:0001629)	Atrial septal defect (HP:0001631)	Coarctation of aorta (HP:0001680)
Congenital hypothyroidism (HP:0000851)	Neonatal hypoglycemia (HP:0001998)	Premature thelarche (HP:0010314)
Cryptorchidism (HP:0000028)	Abnormal renal morphology (HP:0012210)	Abnormality of the anus (HP:0004378)
Vertebral segmentation defect (HP:0003422)	Scoliosis (HP:0002650)	Immunodeficiency (HP:0002721)

Kabuki clinical tests (30864)

Level 3 Title	Kabuki (28664)
Level 4 Title	Kabuki syndrome (10960)
Clinical Tests	NB. Clinical test guidance: Imaging Diagnostics refers to facial and other relevant medical photographs Additional body measurements refers to parental heights Entries ordered left to right in table

Additional body measurements (30247.2)	General Imaging Diagnostics (33633.1)

RASopathies (10961)

Noonan syndrome (11039)

RASopathy phenotypes (28593)

Level 3 Title	RASopathies (10961)
Level 4 Title	Noonan syndrome (11039)
Phenotypes	Entries ordered left to right in table

Increased nuchal translucency (HP:0010880)	Nonimmune hydrops fetalis (HP:0001790)	Polyhydramnios (HP:0001561)
Failure to thrive in infancy (HP:0001531)	Feeding difficulties (HP:0011968)	Pulmonic stenosis (HP:0001642)
Hypertrophic cardiomyopathy (HP:0001639)	Abnormality of the atrioventricular valves (HP:0006705)	Atrial septal defect (HP:0001631)
Ventricular septal defect (HP:0001629)	Arrhythmia (HP:0011675)	Lymphedema (HP:0001004)
Pleural effusion (HP:0002202)	Intellectual disability (HP:0001249)	Specific learning disability (HP:0001328)
Delayed gross motor development (HP:0002194)	Delayed speech and language development (HP:0000750)	Abnormality of the kidney (HP:0000077)
Delayed puberty (HP:0000823)	Cryptorchidism (HP:0000028)	Decreased fertility (HP:0000144)
Abnormality of hair texture (HP:0010719)	Follicular hyperkeratosis (HP:0007502)	Cafe-au-lait spot (HP:0000957)
Multiple lentigines (HP:0001003)	Hemangioma (HP:0001028)	Papilloma (HP:0012740)
Pectus carinatum (HP:0000768)	Pectus excavatum (HP:0000767)	Wide intermamillary distance (HP:0006610)
Webbed neck (HP:0000465)	Abnormality of the coagulation cascade (HP:0003256)	Abnormality of thrombocytes (HP:0001872)
Neoplasm (HP:0002664)	Immunodeficiency (HP:0002721)	Autoimmunity (HP:0002960)
Abnormality of brain morphology (HP:0012443)	Seizures (HP:0001250)	Autism (HP:0000717)
Depression (HP:0000716)	Schizophrenia (HP:0100753)	Ptosis (HP:0000508)
Abnormality of refraction (HP:0000539)	Strabismus (HP:0000486)	Hearing abnormality (HP:0000364)

RASopathy clinical tests (30865)

<table>
<tr><td>Level 3 Title</td><td>RASopathies (10961)</td></tr>
<tr><td>Level 4 Title</td><td>Noonan syndrome (11039)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging Diagnostics refers to facial and other relevant medical photographs
Additional body measurements refers to parental heights
Entries ordered left to right in table

<table><tr><td>Intelligence Quotient Assessment (30224.1)</td><td>Additional body measurements (30247.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr></table></td></tr>
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Noonan syndrome plus other features (11040)

RASopathy phenotypes (28593)

Level 3 Title	RASopathies (10961)
Level 4 Title	Noonan syndrome plus other features (11040)

Phenotypes

Entries ordered left to right in table

Increased nuchal translucency (HP:0010880)	Nonimmune hydrops fetalis (HP:0001790)	Polyhydramnios (HP:0001561)
Failure to thrive in infancy (HP:0001531)	Feeding difficulties (HP:0011968)	Pulmonic stenosis (HP:0001642)
Hypertrophic cardiomyopathy (HP:0001639)	Abnormality of the atrioventricular valves (HP:0006705)	Atrial septal defect (HP:0001631)
Ventricular septal defect (HP:0001629)	Arrhythmia (HP:0011675)	Lymphedema (HP:0001004)
Pleural effusion (HP:0002202)	Intellectual disability (HP:0001249)	Specific learning disability (HP:0001328)
Delayed gross motor development (HP:0002194)	Delayed speech and language development (HP:0000750)	Abnormality of the kidney (HP:0000077)
Delayed puberty (HP:0000823)	Cryptorchidism (HP:0000028)	Decreased fertility (HP:0000144)
Abnormality of hair texture (HP:0010719)	Follicular hyperkeratosis (HP:0007502)	Cafe-au-lait spot (HP:0000957)
Multiple lentigines (HP:0001003)	Hemangioma (HP:0001028)	Papilloma (HP:0012740)
Pectus carinatum (HP:0000768)	Pectus excavatum (HP:0000767)	Wide intermamillary distance (HP:0006610)
Webbed neck (HP:0000465)	Abnormality of the coagulation cascade (HP:0003256)	Abnormality of thrombocytes (HP:0001872)
Neoplasm (HP:0002664)	Immunodeficiency (HP:0002721)	Autoimmunity (HP:0002960)
Abnormality of brain morphology (HP:0012443)	Seizures (HP:0001250)	Autism (HP:0000717)
Depression (HP:0000716)	Schizophrenia (HP:0100753)	Ptosis (HP:0000508)
Abnormality of refraction (HP:0000539)	Strabismus (HP:0000486)	Hearing abnormality (HP:0000364)

RASopathy clinical tests (30865)

<table>
<tr><td>Level 3 Title</td><td>RASopathies (10961)</td></tr>
<tr><td>Level 4 Title</td><td>Noonan syndrome plus other features (11040)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging Diagnostics refers to facial and other relevant medical photographs
Additional body measurements refers to parental heights
Entries ordered left to right in table

<table><tr><td>Intelligence Quotient Assessment (30224.1)</td><td>Additional body measurements (30247.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr></table></td></tr>
</table>

Cardio-facio-cutaneous syndrome (11041)

RASopathy phenotypes (28593)

Level 3 Title	RASopathies (10961)
Level 4 Title	Cardio-facio-cutaneous syndrome (11041)
Phenotypes	Entries ordered left to right in table

Increased nuchal translucency (HP:0010880)	Nonimmune hydrops fetalis (HP:0001790)	Polyhydramnios (HP:0001561)
Failure to thrive in infancy (HP:0001531)	Feeding difficulties (HP:0011968)	Pulmonic stenosis (HP:0001642)
Hypertrophic cardiomyopathy (HP:0001639)	Abnormality of the atrioventricular valves (HP:0006705)	Atrial septal defect (HP:0001631)
Ventricular septal defect (HP:0001629)	Arrhythmia (HP:0011675)	Lymphedema (HP:0001004)
Pleural effusion (HP:0002202)	Intellectual disability (HP:0001249)	Specific learning disability (HP:0001328)
Delayed gross motor development (HP:0002194)	Delayed speech and language development (HP:0000750)	Abnormality of the kidney (HP:0000077)
Delayed puberty (HP:0000823)	Cryptorchidism (HP:0000028)	Decreased fertility (HP:0000144)
Abnormality of hair texture (HP:0010719)	Follicular hyperkeratosis (HP:0007502)	Cafe-au-lait spot (HP:0000957)
Multiple lentigines (HP:0001003)	Hemangioma (HP:0001028)	Papilloma (HP:0012740)
Pectus carinatum (HP:0000768)	Pectus excavatum (HP:0000767)	Wide intermamillary distance (HP:0006610)
Webbed neck (HP:0000465)	Abnormality of the coagulation cascade (HP:0003256)	Abnormality of thrombocytes (HP:0001872)
Neoplasm (HP:0002664)	Immunodeficiency (HP:0002721)	Autoimmunity (HP:0002960)
Abnormality of brain morphology (HP:0012443)	Seizures (HP:0001250)	Autism (HP:0000717)
Depression (HP:0000716)	Schizophrenia (HP:0100753)	Ptosis (HP:0000508)
Abnormality of refraction (HP:0000539)	Strabismus (HP:0000486)	Hearing abnormality (HP:0000364)

RASopathy clinical tests (30865)

Level 3 Title	RASopathies (10961)
Level 4 Title	Cardio-facio-cutaneous syndrome (11041)
Clinical Tests	NB. Clinical test guidance: Imaging Diagnostics refers to facial and other relevant medical photographs Additional body measurements refers to parental heights Entries ordered left to right in table

Intelligence Quotient Assessment (30224.1)	Additional body measurements (30247.2)	General Imaging Diagnostics (33633.1)

LEOPARD syndrome (11042)

RASopathy phenotypes (28593)

Level 3 Title	RASopathies (10961)
Level 4 Title	LEOPARD syndrome (11042)

Phenotypes

Entries ordered left to right in table

Increased nuchal translucency (HP:0010880)	Nonimmune hydrops fetalis (HP:0001790)	Polyhydramnios (HP:0001561)
Failure to thrive in infancy (HP:0001531)	Feeding difficulties (HP:0011968)	Pulmonic stenosis (HP:0001642)
Hypertrophic cardiomyopathy (HP:0001639)	Abnormality of the atrioventricular valves (HP:0006705)	Atrial septal defect (HP:0001631)
Ventricular septal defect (HP:0001629)	Arrhythmia (HP:0011675)	Lymphedema (HP:0001004)
Pleural effusion (HP:0002202)	Intellectual disability (HP:0001249)	Specific learning disability (HP:0001328)
Delayed gross motor development (HP:0002194)	Delayed speech and language development (HP:0000750)	Abnormality of the kidney (HP:0000077)
Delayed puberty (HP:0000823)	Cryptorchidism (HP:0000028)	Decreased fertility (HP:0000144)
Abnormality of hair texture (HP:0010719)	Follicular hyperkeratosis (HP:0007502)	Cafe-au-lait spot (HP:0000957)
Multiple lentigines (HP:0001003)	Hemangioma (HP:0001028)	Papilloma (HP:0012740)
Pectus carinatum (HP:0000768)	Pectus excavatum (HP:0000767)	Wide intermamillary distance (HP:0006610)
Webbed neck (HP:0000465)	Abnormality of the coagulation cascade (HP:0003256)	Abnormality of thrombocytes (HP:0001872)
Neoplasm (HP:0002664)	Immunodeficiency (HP:0002721)	Autoimmunity (HP:0002960)
Abnormality of brain morphology (HP:0012443)	Seizures (HP:0001250)	Autism (HP:0000717)
Depression (HP:0000716)	Schizophrenia (HP:0100753)	Ptosis (HP:0000508)
Abnormality of refraction (HP:0000539)	Strabismus (HP:0000486)	Hearing abnormality (HP:0000364)

RASopathy clinical tests (30865)

Level 3 Title	RASopathies (10961)
Level 4 Title	LEOPARD syndrome (11042)
Clinical Tests	NB. Clinical test guidance: Imaging Diagnostics refers to facial and other relevant medical photographs Additional body measurements refers to parental heights Entries ordered left to right in table

Intelligence Quotient Assessment (30224.1)	Additional body measurements (30247.2)	General Imaging Diagnostics (33633.1)

Costello syndrome (11043)

RASopathy phenotypes (28593)

Level 3 Title	RASopathies (10961)
Level 4 Title	Costello syndrome (11043)

Phenotypes

Entries ordered left to right in table

Increased nuchal translucency (HP:0010880)	Nonimmune hydrops fetalis (HP:0001790)	Polyhydramnios (HP:0001561)
Failure to thrive in infancy (HP:0001531)	Feeding difficulties (HP:0011968)	Pulmonic stenosis (HP:0001642)
Hypertrophic cardiomyopathy (HP:0001639)	Abnormality of the atrioventricular valves (HP:0006705)	Atrial septal defect (HP:0001631)
Ventricular septal defect (HP:0001629)	Arrhythmia (HP:0011675)	Lymphedema (HP:0001004)
Pleural effusion (HP:0002202)	Intellectual disability (HP:0001249)	Specific learning disability (HP:0001328)
Delayed gross motor development (HP:0002194)	Delayed speech and language development (HP:0000750)	Abnormality of the kidney (HP:0000077)
Delayed puberty (HP:0000823)	Cryptorchidism (HP:0000028)	Decreased fertility (HP:0000144)
Abnormality of hair texture (HP:0010719)	Follicular hyperkeratosis (HP:0007502)	Cafe-au-lait spot (HP:0000957)
Multiple lentigines (HP:0001003)	Hemangioma (HP:0001028)	Papilloma (HP:0012740)
Pectus carinatum (HP:0000768)	Pectus excavatum (HP:0000767)	Wide intermamillary distance (HP:0006610)
Webbed neck (HP:0000465)	Abnormality of the coagulation cascade (HP:0003256)	Abnormality of thrombocytes (HP:0001872)
Neoplasm (HP:0002664)	Immunodeficiency (HP:0002721)	Autoimmunity (HP:0002960)
Abnormality of brain morphology (HP:0012443)	Seizures (HP:0001250)	Autism (HP:0000717)
Depression (HP:0000716)	Schizophrenia (HP:0100753)	Ptosis (HP:0000508)
Abnormality of refraction (HP:0000539)	Strabismus (HP:0000486)	Hearing abnormality (HP:0000364)

RASopathy clinical tests (30865)

Level 3 Title	RASopathies (10961)
Level 4 Title	Costello syndrome (11043)
Clinical Tests	NB. Clinical test guidance: Imaging Diagnostics refers to facial and other relevant medical photographs Additional body measurements refers to parental heights Entries ordered left to right in table

Intelligence Quotient Assessment (30224.1)	Additional body measurements (30247.2)	General Imaging Diagnostics (33633.1)

Legius syndrome (11044)

RASopathy phenotypes (28593)

Level 3 Title	RASopathies (10961)
Level 4 Title	Legius syndrome (11044)

Phenotypes

Entries ordered left to right in table

Increased nuchal translucency (HP:0010880)	Nonimmune hydrops fetalis (HP:0001790)	Polyhydramnios (HP:0001561)
Failure to thrive in infancy (HP:0001531)	Feeding difficulties (HP:0011968)	Pulmonic stenosis (HP:0001642)
Hypertrophic cardiomyopathy (HP:0001639)	Abnormality of the atrioventricular valves (HP:0006705)	Atrial septal defect (HP:0001631)
Ventricular septal defect (HP:0001629)	Arrhythmia (HP:0011675)	Lymphedema (HP:0001004)
Pleural effusion (HP:0002202)	Intellectual disability (HP:0001249)	Specific learning disability (HP:0001328)
Delayed gross motor development (HP:0002194)	Delayed speech and language development (HP:0000750)	Abnormality of the kidney (HP:0000077)
Delayed puberty (HP:0000823)	Cryptorchidism (HP:0000028)	Decreased fertility (HP:0000144)
Abnormality of hair texture (HP:0010719)	Follicular hyperkeratosis (HP:0007502)	Cafe-au-lait spot (HP:0000957)
Multiple lentigines (HP:0001003)	Hemangioma (HP:0001028)	Papilloma (HP:0012740)
Pectus carinatum (HP:0000768)	Pectus excavatum (HP:0000767)	Wide intermamillary distance (HP:0006610)
Webbed neck (HP:0000465)	Abnormality of the coagulation cascade (HP:0003256)	Abnormality of thrombocytes (HP:0001872)
Neoplasm (HP:0002664)	Immunodeficiency (HP:0002721)	Autoimmunity (HP:0002960)
Abnormality of brain morphology (HP:0012443)	Seizures (HP:0001250)	Autism (HP:0000717)
Depression (HP:0000716)	Schizophrenia (HP:0100753)	Ptosis (HP:0000508)
Abnormality of refraction (HP:0000539)	Strabismus (HP:0000486)	Hearing abnormality (HP:0000364)

RASopathy clinical tests (30865)

Level 3 Title	RASopathies (10961)
Level 4 Title	Legius syndrome (11044)
Clinical Tests	NB. Clinical test guidance: Imaging Diagnostics refers to facial and other relevant medical photographs Additional body measurements refers to parental heights Entries ordered left to right in table

Intelligence Quotient Assessment (30224.1)	Additional body measurements (30247.2)	General Imaging Diagnostics (33633.1)

Balanced translocations (10962)

Balanced translocations with an unusual phenotype (11045)

General Dysmorphology phenotypes (28662)

Level 3 Title	Balanced translocations (10962)
Level 4 Title	Balanced translocations with an unusual phenotype (11045)
Phenotypes	Entries ordered left to right in table

Small for gestational age (HP:0001518)	Large for gestational age (HP:0001520)	Failure to thrive (HP:0001508)
Short stature (HP:0004322)	Tall stature (HP:0000098)	Microcephaly (HP:0000252)
Macrocephaly (HP:0000256)	Abnormal facial shape (HP:0001999)	Intellectual disability (HP:0001249)
Global developmental delay (HP:0001263)	Hyperpigmented streaks (HP:0007572)	Hypermelanotic macule (HP:0001034)
Macular hypopigmented whorls, streaks, and patches (HP:0005593)	Hypopigmentation of the skin (HP:0001010)	Cutaneous photosensitivity (HP:0000992)
Abnormality of the cardiovascular system (HP:0001626)	Abnormality of the musculature (HP:0003011)	Abnormality of the gastrointestinal tract (HP:0011024)
Abnormality of the liver (HP:0001392)	Abnormality of the respiratory system (HP:0002086)	Abnormality of the endocrine system (HP:0000818)
Abnormality of metabolism/homeostasis (HP:0001939)	Abnormality of blood and blood-forming tissues (HP:0001871)	Abnormality of the immune system (HP:0002715)
Abnormality of the skeletal system (HP:0000924)	Abnormality of the genitourinary system (HP:0000119)	Abnormality of prenatal development or birth (HP:0001197)

Balanced translocations with an unusual phenotype clinical tests (30866)

<table>
<tr><td>Level 3 Title</td><td>Balanced translocations (10962)</td></tr>
<tr><td>Level 4 Title</td><td>Balanced translocations with an unusual phenotype (11045)</td></tr>
<tr><td>Clinical Tests</td><td>N.B. Please ensure IGCN nomenclature karyotype entered and report uploaded in ‘genetic investigations’
Clinical test guidance:
General Imaging Diagnostics refers to facial and other relevant medical photographs
Entries ordered left to right in table

<table><tr><td>APGAR score (30207.1)</td><td>Facial features most in keeping with an OMIM disease (29826.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr></table></td></tr>
</table>

Limb disorders (15087)

VACTERL-like phenotypes (10964)

VACTERL phenotypes (28595)

Level 3 Title	Limb disorders (15087)
Level 4 Title	VACTERL-like phenotypes (10964)
Phenotypes	Entries ordered left to right in table

Abnormality of the skeletal system (HP:0000924)	Radioulnar synostosis (HP:0002974)	Absent radius (HP:0003974)
Aplasia/Hypoplasia of the radius (HP:0006501)	Hypoplasia of the radius (HP:0002984)	Absent thumb (HP:0009777)
Short thumb (HP:0009778)	Scoliosis (HP:0002650)	Hemivertebrae (HP:0002937)
Vertebral wedging (HP:0008422)	Vertebral fusion (HP:0002948)	Supernumerary vertebrae (HP:0002946)
Abnormality of the ribs (HP:0000772)	Abnormality of the sternum (HP:0000766)	Abnormality of abdomen morphology (HP:0001438)
Anal atresia (HP:0002023)	Abnormality of the cardiovascular system (HP:0001626)	Abnormality of cardiovascular system morphology (HP:0030680)
Abnormality of the cardiac septa (HP:0001671)	Atrial septal defect (HP:0001631)	Ventricular septal defect (HP:0001629)
Complete atrioventricular canal defect (HP:0001674)	Tetralogy of Fallot (HP:0001636)	Transposition of the great arteries (HP:0001669)
Coarctation of aorta (HP:0001680)	Single umbilical artery (HP:0001195)	Abnormality of the respiratory system (HP:0002086)
Tracheoesophageal fistula (HP:0002575)	Esophageal atresia (HP:0002032)	Laryngeal stenosis (HP:0001602)
Laryngomalacia (HP:0001601)	Tracheal stenosis (HP:0002777)	Abnormality of the genitourinary system (HP:0000119)
Multicystic kidney dysplasia (HP:0000003)	Horseshoe kidney (HP:0000085)	Abnormal localization of kidney (HP:0100542)
Renal agenesis (HP:0000104)	Renal hypoplasia (HP:0000089)	Ectopic kidney (HP:0000086)
Renal dysplasia (HP:0000110)	Renal hypoplasia/aplasia (HP:0008678)	

VACTERL-like clinical tests (33365)

Level 3 Title	Limb disorders (15087)
Level 4 Title	VACTERL-like phenotypes (10964)
Clinical Tests	Entries ordered left to right in table General Imaging Diagnostics (33633.1)

DNA repair disorders (10965)

Cockayne syndrome (36497)

Cockayne syndrome phenotypes (36641)

Level 3 Title	DNA repair disorders (10965)
Level 4 Title	Cockayne syndrome (36497)
Phenotypes	Entries ordered left to right in table

Microcephaly (HP:0000252)	Short stature (HP:0004322)	Severe short stature (HP:0003510)
Global developmental delay (HP:0001263)	Cutaneous photosensitivity (HP:0000992)	Abnormality of skin pigmentation (HP:0001000)
Abnormality of dental enamel (HP:0000682)	Hearing impairment (HP:0000365)	Conductive hearing impairment (HP:0000405)
Sensorineural hearing impairment (HP:0000407)	Deeply set eye (HP:0000490)	Cataract (HP:0000518)
Congenital cataract (HP:0000519)	Pigmentary retinal degeneration (HP:0001146)	Pigmentary retinopathy (HP:0000580)
Retinal degeneration (HP:0000546)	Photophobia (HP:0000613)	Lipoatrophy (HP:0100578)
Lipodystrophy (HP:0009125)	Tremor (HP:0001337)	Seizures (HP:0001250)
Gait disturbance (HP:0001288)	Peripheral neuropathy (HP:0009830)	Dysmyelinating leukodystrophy (HP:0006978)
Cerebral calcification (HP:0002514)	Cerebellar hypoplasia (HP:0001321)	Muscle weakness (HP:0001324)
Contractures of the joints of the lower limbs (HP:0005750)	Contractures of the joints of the upper limbs (HP:0100360)	Scoliosis (HP:0002650)
Kyphosis (HP:0002808)	Kyphoscoliosis (HP:0002751)	Elevated hepatic transaminases (HP:0002910)
Hypertension (HP:0000822)		

Cockayne syndrome clinical tests (36499)

Level 3 Title	DNA repair disorders (10965)
Level 4 Title	Cockayne syndrome (36497)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to medical photographs of the face. DNA repair test results should be uploaded using the genetic investigation form. Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	Blood pressure (30245.1)	Liver biochemistry (30328.2)

Non-Fanconi anaemia (11050)

Non-Fanconi phenotypes (29225)

Level 3 Title	DNA repair disorders (10965)
Level 4 Title	Non-Fanconi anaemia (11050)
Phenotypes	Entries ordered left to right in table

Cafe-au-lait spot (HP:0000957)	Abnormality of skin pigmentation (HP:0001000)	Leukemia (HP:0001909)
Acute myeloid leukemia (HP:0004808)	Myelodysplasia (HP:0002863)	Pancytopenia (HP:0001876)
Anemia (HP:0001903)	Neutropenia (HP:0001875)	Reticulocytopenia (HP:0001896)
Thrombocytopenia (HP:0001873)	Bruising susceptibility (HP:0000978)	Increased mean corpuscular volume (HP:0005518)
Short thumb (HP:0009778)	Abnormality of cardiovascular system morphology (HP:0030680)	Horseshoe kidney (HP:0000085)
Absent radius (HP:0003974)	Hypoplasia of the radius (HP:0002984)	Aplasia/Hypoplasia of the thumb (HP:0009601)
Absent thumb (HP:0009777)	Ectopic kidney (HP:0000086)	Complete duplication of thumb phalanx (HP:0009943)
Renal agenesis (HP:0000104)	Duplicated collecting system (HP:0000081)	Short stature (HP:0004322)
Small for gestational age (HP:0001518)	Microcephaly (HP:0000252)	Cryptorchidism (HP:0000028)
Hypogonadism (HP:0000135)	Microphthalmia (HP:0000568)	Strabismus (HP:0000486)
Hearing impairment (HP:0000365)	Intellectual disability (HP:0001249)	Chromosomal breakage induced by crosslinking agents (HP:0003221)
Deficient excision of UV-induced pyrimidine dimers in DNA (HP:0003213)	Prolonged G2 phase of cell cycle (HP:0003214)	

Non-Fanconi clinical tests (33366)

Level 3 Title	DNA repair disorders (10965)
Level 4 Title	Non-Fanconi anaemia (11050)
Clinical Tests	NB. Clinical test guidance: Please ensure chromosome breakage result entered and report uploaded in 'genetic investigations' Entries ordered left to right in table

Xeroderma Pigmentosum-like disorders (15089)

Xeroderma Pigmentosum-like phenotypes (29226)

Level 3 Title	DNA repair disorders (10965)
Level 4 Title	Xeroderma Pigmentosum-like disorders (15089)
Phenotypes	Entries ordered left to right in table

Telangiectasia (HP:0001009)	Dermal atrophy (HP:0004334)	Hyperpigmentation of the skin (HP:0000953)
Numerous pigmented freckles (HP:0007587)	Freckling (HP:0001480)	Hypopigmentation of the skin (HP:0001010)
Poikiloderma (HP:0001029)	Cutaneous photosensitivity (HP:0000992)	Neoplasm of the skin (HP:0008069)
Squamous cell carcinoma of the skin (HP:0006739)	Melanoma (HP:0002861)	Cutaneous melanoma (HP:0012056)
Basal cell carcinoma (HP:0002671)	Ectropion (HP:0000656)	Entropion (HP:0000621)
Conjunctivitis (HP:0000509)	Keratitis (HP:0000491)	Cataract (HP:0000518)
Deeply set eye (HP:0000490)	Microphthalmia (HP:0000568)	Optic atrophy (HP:0000648)
Nystagmus (HP:0000639)	Astigmatism (HP:0000483)	Photophobia (HP:0000613)
Spasticity (HP:0001257)	Microcephaly (HP:0000252)	Intellectual disability (HP:0001249)
Dementia (HP:0000726)	Mental deterioration (HP:0001268)	Tremor (HP:0001337)
Basal ganglia calcification (HP:0002135)	Ataxia (HP:0001251)	Hyporeflexia (HP:0001265)
Hyperreflexia (HP:0001347)	Flexion contracture (HP:0001371)	Scoliosis (HP:0002650)
Brain atrophy (HP:0012444)	Abnormal CNS myelination (HP:0011400)	Ventriculomegaly (HP:0002119)
Cerebellar atrophy (HP:0001272)	Peripheral neuropathy (HP:0009830)	Decreased nerve conduction velocity (HP:0000762)
Hypogonadism (HP:0000135)	Hearing impairment (HP:0000365)	Sensorineural hearing impairment (HP:0000407)
Short stature (HP:0004322)	Decreased body weight (HP:0004325)	Progeroid facial appearance (HP:0005328)
Neoplasm (HP:0002664)	Increased cellular sensitivity to UV light (HP:0003224)	Defective DNA repair after ultraviolet radiation damage (HP:0003079)

Xeroderma Pigmentosum-like clinical tests (33367)

Level 3 Title	DNA repair disorders (10965)
Level 4 Title	Xeroderma Pigmentosum-like disorders (15089)
Clinical Tests	NB. Clinical test guidance: Please ensure DNA repair result entered and report uploaded in 'genetic investigations' Entries ordered left to right in table

Primary Microcephaly - Microcephalic Dwarfism Spectrum (36505)

Primary Microcephaly - Microcephalic Dwarfism Spectrum phenotypes (36642)

Level 3 Title	DNA repair disorders (10965)
Level 4 Title	Primary Microcephaly - Microcephalic Dwarfism Spectrum (36505)
Phenotypes	Entries ordered left to right in table

Intellectual disability (HP:0001249)	Intellectual disability, mild (HP:0001256)	Intellectual disability, moderate (HP:0002342)
Intellectual disability, severe (HP:0010864)	Aplasia/Hypoplasia of the patella (HP:0006498)	Patellar hypoplasia (HP:0003065)
Patellar aplasia (HP:0006443)	Microtia (HP:0008551)	Seizures (HP:0001250)
Microcephaly (HP:0000252)	Postnatal microcephaly (HP:0005484)	Progressive microcephaly (HP:0000253)
Congenital microcephaly (HP:0011451)	Short stature (HP:0004322)	Proportionate short stature (HP:0003508)
Small for gestational age (HP:0001518)	Intrauterine growth retardation (HP:0001511)	Maternal teratogenic exposure (HP:0011438)
Diabetes mellitus (HP:0000819)	Abnormality of the hair (HP:0001595)	Hearing impairment (HP:0000365)
Pancytopenia (HP:0001876)	Recurrent infections (HP:0002719)	

Primary Microcephaly - Microcephalic Dwarfism Spectrum clinical tests (36507)

Level 3 Title	DNA repair disorders (10965)
Level 4 Title	Primary Microcephaly - Microcephalic Dwarfism Spectrum (36505)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to medical photographs of face including ears, MRI brain and skeletal survey. Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Autophagy disorders (10966)

Vici Syndrome and other autophagy disorders (11051)

Vici phenotypes (28596)

Level 3 Title	Autophagy disorders (10966)
Level 4 Title	Vici Syndrome and other autophagy disorders (11051)
Phenotypes	Entries ordered left to right in table

Decreased fetal movement (HP:0001558)	Increased fetal movement (HP:0010519)	Polyhydramnios (HP:0001561)
Oligohydramnios (HP:0001562)	Feeding difficulties in infancy (HP:0008872)	Nasogastric tube feeding in infancy (HP:0011470)
Gastrostomy tube feeding in infancy (HP:0011471)	Failure to thrive (HP:0001508)	Hypoglycemia (HP:0001943)
Global developmental delay (HP:0001263)	Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)
Delayed speech and language development (HP:0000750)	Intellectual disability (HP:0001249)	Hearing impairment (HP:0000365)
Sensorineural hearing impairment (HP:0000407)	Conductive hearing impairment (HP:0000405)	Microcephaly (HP:0000252)
Visual impairment (HP:0000505)	Febrile seizures (HP:0002373)	Focal seizures (HP:0007359)
Absence seizures (HP:0002121)	Generalized seizures (HP:0002197)	Status epilepticus (HP:0002133)
Abnormality of neuronal migration (HP:0002269)	Agenesis of corpus callosum (HP:0001274)	Cerebellar atrophy (HP:0001272)
Olivopontocerebellar hypoplasia (HP:0006955)	Facial palsy (HP:0010628)	Infantile muscular hypotonia (HP:0008947)
Generalized muscle weakness (HP:0003324)	Axial muscle weakness (HP:0003327)	Proximal muscle weakness (HP:0003701)
Distal muscle weakness (HP:0002460)	Weak extraocular muscles (HP:0007715)	Reduced tendon reflexes (HP:0001315)
Areflexia (HP:0001284)	Hyperactive deep tendon reflexes (HP:0006801)	EMG abnormality (HP:0003457)

Decreased nerve conduction velocity (HP:0000762)	Generalized hyperpigmentation (HP:0007440)	Hypopigmentation of the skin (HP:0001010)
Vitiligo (HP:0001045)	Recurrent infections (HP:0002719)	Leukopenia (HP:0001882)
Abnormality of B cells (HP:0002846)	Abnormality of T cells (HP:0002843)	Abnormality of blood and blood-forming tissues (HP:0001871)
Thrombocytopenia (HP:0001873)	Anemia (HP:0001903)	Abnormality of the thymus (HP:0000777)
Aplasia/Hypoplasia of the thymus (HP:0010515)	Abnormality of the eye (HP:0000478)	Cataract (HP:0000518)
Optic atrophy (HP:0000648)	Abnormality of the retina (HP:0000479)	Abnormality of visual evoked potentials (HP:0000649)
Abnormal electroretinogram (HP:0000512)	Abnormality of the cardiovascular system (HP:0001626)	Cardiomyopathy (HP:0001638)
Dilated cardiomyopathy (HP:0001644)	Hypertrophic cardiomyopathy (HP:0001639)	Respiratory insufficiency (HP:0002093)
Respiratory failure requiring assisted ventilation (HP:0004887)	Abnormality of lung morphology (HP:0002088)	Pulmonary hypoplasia (HP:0002089)
Abnormality of the thyroid gland (HP:0000820)	Thyroid agenesis (HP:0008191)	Hyperthyroidism (HP:0000836)
Hypothyroidism (HP:0000821)	Hepatomegaly (HP:0002240)	Abnormality of the kidney (HP:0000077)
Hydronephrosis (HP:0000126)	Renal tubular acidosis (HP:0001947)	Abnormality of the liver (HP:0001392)
Elevated hepatic transaminases (HP:0002910)	Ichthyosis (HP:0008064)	Abnormality of ion homeostasis (HP:0003111)
Abnormal muscle glycogen content (HP:0012269)	Central core regions in muscle fibers (HP:0030230)	Increased variability in muscle fiber diameter (HP:0003557)
Type 1 muscle fiber predominance (HP:0003803)		

Vici clinical tests (30867)

Level 3 Title	Autophagy disorders (10966)
Level 4 Title	Vici Syndrome and other autophagy disorders (11051)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to muscle and/or nerve biopsy General Imaging diagnostics refers to MRI brain General Non-imaging diagnostics refers to EMG/nerve conduction Entries ordered left to right in table

General Biopsy (33614.1)	General Imaging Diagnostics (33633.1)	General Non-imaging Diagnostics (34838.1)

Dysmorphic disorders (36595)

Coarse facial features including Coffin-Siris-like disorders (36596)

Coarse facial features including Coffin-Siris-like disorders phenotypes (36644)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	Coarse facial features including Coffin-Siris-like disorders (36596)
Phenotypes	Entries ordered left to right in table

Coarse facial features (HP:0000280)	Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)
Delayed speech and language development (HP:0000750)	Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)
Feeding difficulties in infancy (HP:0008872)	Muscular hypotonia (HP:0001252)	Seizures (HP:0001250)
Hearing impairment (HP:0000365)	Visual impairment (HP:0000505)	Hirsutism (HP:0001007)
Sparse hair (HP:0008070)	Low anterior hairline (HP:0000294)	Synophrys (HP:0000664)
Thick eyebrow (HP:0000574)	Prominent eyelashes (HP:0011231)	Gingival overgrowth (HP:0000212)
Abnormality of the pinna (HP:0000377)	Wide intermamillary distance (HP:0006610)	Pectus excavatum (HP:0000767)
Pectus carinatum (HP:0000768)	Cryptorchidism (HP:0000028)	Umbilical hernia (HP:0001537)
Short 5th finger (HP:0009237)	Aplasia of the distal phalanx of the 5th finger (HP:0009246)	Prominent interphalangeal joints (HP:0006237)
Deep palmar crease (HP:0006191)	Deep plantar creases (HP:0001869)	Hypoplastic fifth fingernail (HP:0008398)
Hypoplastic fifth toenail (HP:0011937)	Abnormality of cardiovascular system morphology (HP:0030680)	Abnormality of the kidney (HP:0000077)
Hepatomegaly (HP:0002240)	Splenomegaly (HP:0001744)	Hypoplasia of the corpus callosum (HP:0002079)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	Coarse facial features including Coffin-Siris-like disorders (36596)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to facial and other relevant clinical photographs Entries ordered left to right in table

General Non-imaging Diagnostics (34838.1)	General Imaging Diagnostics (33633.1)

Familial non-syndromic cleft lip and or familial cleft palate (37565)

Familial Non-syndromic cleft lip and or familial cleft palate phenotypes (37740)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	Familial non-syndromic cleft lip and or familial cleft palate (37565)
Phenotypes	Entries ordered left to right in table

Oral cleft (HP:0000202)	Cleft upper lip (HP:0000204)	Median cleft lip (HP:0000161)
Non-midline cleft lip (HP:0100335)	Bilateral cleft lip (HP:0100336)	Unilateral cleft lip (HP:0100333)
Incomplete cleft of the upper lip (HP:0011340)	Alveolar ridge cleft (HP:0010289)	Cleft lower lip (HP:0010281)
Cleft palate (HP:0000175)	Cleft hard palate (HP:0410005)	Cleft soft palate (HP:0000185)
Submucous cleft soft palate (HP:0011819)	Bifid uvula (HP:0000193)	Velopharyngeal insufficiency (HP:0000220)
Lower lip pit (HP:0000196)	Micrognathia (HP:0000347)	Hypodontia (HP:0000668)
Pierre-Robin sequence (HP:0000201)	Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)
Abnormal heart morphology (HP:0001627)		

Familial Non-syndromic cleft lip and or familial cleft palate clinical tests (37567)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	Familial non-syndromic cleft lip and or familial cleft palate (37565)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to relevant medical photographs Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Syndromic cleft lip and or cleft palate (37573)

Syndromic cleft lip and or cleft palate phenotypes (37745)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	Syndromic cleft lip and or cleft palate (37573)
Phenotypes	Entries ordered left to right in table

Oral cleft (HP:0000202)	Cleft upper lip (HP:0000204)	Median cleft lip (HP:0000161)
Non-midline cleft lip (HP:0100335)	Bilateral cleft lip (HP:0100336)	Unilateral cleft lip (HP:0100333)
Incomplete cleft of the upper lip (HP:0011340)	Alveolar ridge cleft (HP:0010289)	Cleft lower lip (HP:0010281)
Cleft palate (HP:0000175)	Cleft hard palate (HP:0410005)	Cleft soft palate (HP:0000185)
Submucous cleft soft palate (HP:0011819)	Bifid uvula (HP:0000193)	Velopharyngeal insufficiency (HP:0000220)
Lower lip pit (HP:0000196)	Micrognathia (HP:0000347)	Hypodontia (HP:0000668)
Pierre-Robin sequence (HP:0000201)	Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)
Abnormal heart morphology (HP:0001627)	Facial cleft (HP:0002006)	Abnormal facial shape (HP:0001999)

Syndromic cleft lip and or cleft palate clinical tests (37575)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	Syndromic cleft lip and or cleft palate (37573)
Clinical Tests	NB. Phenotype models for Congenital heart disease and RASopathy should also be completed if necessary Clinical test guidance: General imaging diagnostics refers to relevant medical photographs Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

PHACE(S) syndrome (37578)

PHACE(S) syndrome phenotypes (37900)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	PHACE(S) syndrome (37578)
Phenotypes	Entries ordered left to right in table

Facial hemangioma (HP:0000329)	Abnormality of cerebral artery (HP:0009145)	Dandy-Walker malformation (HP:0001305)
Cerebellar hypoplasia (HP:0001321)	Cerebellar dysplasia (HP:0007033)	Coarctation of aorta (HP:0001680)
Thoracic aortic aneurysm (HP:0012727)	Vascular ring (HP:0010775)	Abnormality of the posterior segment of the globe (HP:0004329)
Remnants of the hyaloid vascular system (HP:0007968)	Abnormality of the retinal vasculature (HP:0008046)	Optic nerve hypoplasia (HP:0000609)
Coloboma (HP:0000589)	Abnormality of the anterior segment of the globe (HP:0004328)	Cataract (HP:0000518)
Sclerocornea (HP:0000647)	Abnormality of the sternum (HP:0000766)	Bifid sternum (HP:0010309)
Hypopituitarism (HP:0040075)	Ectopic thyroid (HP:0100028)	

PHACE(S) syndrome clinical tests (37580)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	PHACE(S) syndrome (37578)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to MR imaging of cerebral and vascular defects and medical photography of relevant dysmorphic and dermatological features Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Radial dysplasia (37636)

Radial dysplasia phenotypes (37749)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	Radial dysplasia (37636)
Phenotypes	Entries ordered left to right in table

Abnormality of the radius (HP:0002818)	Absent radius (HP:0003974)	Hypoplasia of the radius (HP:0002984)
Radioulnar synostosis (HP:0002974)	Radial deviation of the hand (HP:0009486)	Radial club hand (HP:0004059)
Small thenar eminence (HP:0001245)	Absent thumb (HP:0009777)	Short thumb (HP:0009778)
Triphalangeal thumb (HP:0001199)	Microcephaly (HP:0000252)	Malar flattening (HP:0000272)
Micrognathia (HP:0000347)	Cleft palate (HP:0000175)	Duane anomaly (HP:0009921)
Atrial septal defect (HP:0001631)	Arrhythmia (HP:0011675)	Hemivertebrae (HP:0002937)
Tracheoesophageal fistula (HP:0002575)	Esophageal atresia (HP:0002032)	Anal atresia (HP:0002023)
Renal hypoplasia/aplasia (HP:0008678)	Abnormal localization of kidney (HP:0100542)	Horseshoe kidney (HP:0000085)
Cow milk allergy (HP:0100327)	Cafe-au-lait spot (HP:0000957)	Hypopigmented skin patches (HP:0001053)
Anemia (HP:0001903)	Pancytopenia (HP:0001876)	Thrombocytopenia (HP:0001873)

Radial dysplasia clinical tests (37638)

Level 3 Title	Dysmorphic disorders (36595)
Level 4 Title	Radial dysplasia (37636)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to medical photography of the hands and other relevant dysmorphology, skeletal radiographs of the hands if performed Entries ordered left to right in table

ECG diagnostics (30183.2)	Echocardiogram (29800.2)	General Imaging Diagnostics (33633.1)

Fetal disorders (38586)

Fetal hydrops (37586)

Fetal hydrops phenotypes (37746)

Level 3 Title	Fetal disorders (38586)
Level 4 Title	Fetal hydrops (37586)
Phenotypes	Entries ordered left to right in table

Hydrops fetalis (HP:0001789)	Nonimmune hydrops fetalis (HP:0001790)	Fetal cystic hygroma (HP:0010878)
Fetal ascites (HP:0001791)	Pleural effusion (HP:0002202)	Generalized edema (HP:0007430)
Peripheral edema (HP:0012398)	Edema of the dorsum of feet (HP:0012098)	Abnormality of ductus venosus blood flow (HP:0010947)
Arthrogryposis multiplex congenita (HP:0002804)	Fetal akinesia sequence (HP:0001989)	Polyhydramnios (HP:0001561)
Oligohydramnios (HP:0001562)	Hepatosplenomegaly (HP:0001433)	Hepatomegaly (HP:0002240)
Splenomegaly (HP:0001744)	Short fetal femur length (HP:0011428)	Short fetal humerus length (HP:0011429)
Microcephaly (HP:0000252)	Macrocephaly (HP:0000256)	Hydronephrosis (HP:0000126)
Abnormalities of placenta or umbilical cord (HP:0001194)	Abnormal placental size (HP:0012767)	Hydropic placenta (HP:0011414)
Ventriculomegaly (HP:0002119)	Progressive ventriculomegaly (HP:0007100)	Mild fetal ventriculomegaly (HP:0010952)

Fetal hydrops clinical tests (37588)

Level 3 Title	Fetal disorders (38586)
Level 4 Title	Fetal hydrops (37586)
Clinical Tests	Entries ordered left to right in table

Unexplained monogenic fetal disorders (38665)

Ultra-rare undescribed monogenic disorders phenotypes (30790)

Level 3 Title	Fetal disorders (38586)
Level 4 Title	Unexplained monogenic fetal disorders (38665)
Phenotypes	Entries ordered left to right in table

Abnormality of the cardiovascular system (HP:0001626)	Abnormality of the nervous system (HP:0000707)	Abnormality of the musculature (HP:0003011)
Abnormality of the gastrointestinal tract (HP:0011024)	Abnormality of the liver (HP:0001392)	Abnormality of the respiratory system (HP:0002086)
Abnormality of the endocrine system (HP:0000818)	Abnormality of metabolism/homeostasis (HP:0001939)	Abnormality of blood and blood-forming tissues (HP:0001871)
Abnormality of the immune system (HP:0002715)	Abnormality of the skeletal system (HP:0000924)	Abnormality of the integument (HP:0001574)
Abnormality of the genitourinary system (HP:0000119)	Abnormality of prenatal development or birth (HP:0001197)	Growth abnormality (HP:0001507)
Abnormality of the ear (HP:0000598)	Abnormality of the eye (HP:0000478)	

Unexplained monogenic fetal disorders clinical tests (38666)

Level 3 Title	Fetal disorders (38586)
Level 4 Title	Unexplained monogenic fetal disorders (38665)
Clinical Tests	Entries ordered left to right in table

Endocrine disorders (10967)

Adrenal disorders (10969)

Congenital adrenal hypoplasia (11053)

Congenital adrenal hypoplasia phenotypes (29228)

Level 3 Title	Adrenal disorders (10969)
Level 4 Title	Congenital adrenal hypoplasia (11053)
Phenotypes	Entries ordered left to right in table

Congenital adrenal hypoplasia (HP:0008244)	Abnormality of the adrenal glands (HP:0000834)	Abnormality of adrenal physiology (HP:0011733)
Primary adrenal insufficiency (HP:0008207)	Decreased circulating aldosterone level (HP:0004319)	Hypocortisolemia (HP:0008220)
Abnormality of adrenal morphology (HP:0011732)	Adrenal gland dysgenesis (HP:0008216)	Adrenal gland agenesis (HP:0011743)

Congenital adrenal hypoplasia clinical tests (30871)

Level 3 Title	Adrenal disorders (10969)
Level 4 Title	Congenital adrenal hypoplasia (11053)
Clinical Tests	NB. Clinical test guidance: Renal biochemistry refers to lowest sodium measurement General Imaging diagnostics refers to adrenal imaging of any modality Entries ordered left to right in table

Renal biochemistry (30355.2)	Cortisol (33148.1)	Renin and aldosterone (33166.1)
Glucose (33153.1)	General Imaging Diagnostics (33633.1)	

Familial or syndromic hypoparathyroidism (11054)

Hypoparathyroidism phenotypes (29229)

Level 3 Title	Disorders of calcium homeostasis (10970)
Level 4 Title	Familial or syndromic hypoparathyroidism (11054)
Phenotypes	Entries ordered left to right in table

Hypoparathyroidism (HP:0000829)	Congenital hypoparathyroidism (HP:0008198)	Ectopic parathyroid (HP:0011769)
Parathyroid hypoplasia (HP:0000860)	Parathyroid agenesis (HP:0008211)	Autoimmune hypoparathyroidism (HP:0011771)
Abnormal concentration of calcium in blood (HP:0040077)	Abnormality of calcium homeostasis (HP:0004363)	Hypocalcemia (HP:0002901)
Abnormality of calcium-phosphate metabolism (HP:0100530)	Bilateral intracranial calcifications (HP:0005671)	

Familial or syndromic hypoparathyroidism clinical tests (30872)

Level 3 Title	Disorders of calcium homeostasis (10970)
Level 4 Title	Familial or syndromic hypoparathyroidism (11054)
Clinical Tests	Entries ordered left to right in table

Extended renal biochemistry (33152.1)	Bone profile (30317.2)	Urine electrolytes (33187.2)

Gonadal and sex development disorders (36923)

Disorders of sex development (36852)

Disorders of sex development phenotypes (36908)

Level 3 Title	Gonadal and sex development disorders (36923)
Level 4 Title	Disorders of sex development (36852)
Phenotypes	Entries ordered left to right in table

Abnormality of male external genitalia (HP:0000032)	Ambiguous genitalia, male (HP:0000033)	Male pseudohermaphroditism (HP:0000037)
Abnormality of male internal genitalia (HP:0000022)	Micropenis (HP:0000054)	Glandular hypospadias (HP:0000807)
Coronal hypospadias (HP:0008743)	Epispadias (HP:0000039)	Penoscrotal hypospadias (HP:0000808)
Perineal hypospadias (HP:0000051)	Chordee (HP:0000041)	Congenital posterior urethral valve (HP:0010957)
Cryptorchidism (HP:0000028)	Hydrocele testis (HP:0000034)	Aplasia/Hypoplasia of the testes (HP:0010468)
Gonadal dysgenesis, male (HP:0008668)	Scrotal hypoplasia (HP:0000046)	Bifid scrotum (HP:0000048)
Shawl scrotum (HP:0000049)	Penoscrotal transposition (HP:0100600)	Urogenital sinus anomaly (HP:0100779)
Abnormality of female external genitalia (HP:0000055)	Abnormality of female internal genitalia (HP:0000008)	Clitoral hypertrophy (HP:0008665)
Displacement of the external urethral meatus (HP:0100627)	Urethral stenosis (HP:0008661)	Hypoplastic labia majora (HP:0000059)
Fused labia minora (HP:0000063)	Labial hypertrophy (HP:0000065)	Vaginal atresia (HP:0000148)
Abnormality of the uterus (HP:0000130)	Menorrhagia (HP:0000132)	Gonadal dysgenesis (HP:0000133)
Hypogonadism (HP:0000135)	Bifid uterus (HP:0000136)	Abnormality of the ovary (HP:0000137)
Ovarian cyst (HP:0000138)	Abnormality of the menstrual cycle (HP:0000140)	Aplasia/Hypoplasia of the vagina (HP:0011026)

	Longitudinal vaginal septum (HP:0008740)	Rectovaginal fistula (HP:0000143)	Bicornuate uterus (HP:0000813)
	Uterus didelphys (HP:0003762)	Hypoplasia of the uterus (HP:0000013)	Aplasia of the uterus (HP:0000151)
	Aplasia/Hypoplasia of the fallopian tube (HP:0008655)	Streak ovary (HP:0010464)	Aplasia of the ovary (HP:0010463)
	Primary amenorrhea (HP:0000786)	Premature ovarian failure (HP:0008209)	Polycystic ovaries (HP:0000147)
	Hypergonadotropic hypogonadism (HP:0000815)	Hypogonadotrophic hypogonadism (HP:0000044)	Absent gonadal tissue (HP:0008633)
	Overgrowth of external genitalia (HP:0003247)	Gonadal neoplasm (HP:0010785)	Teratoma (HP:0009792)

Disorders of sex development clinical tests (36883)

Level 3 Title	Gonadal and sex development disorders (36923)
Level 4 Title	Disorders of sex development (36852)
Clinical Tests	Entries ordered left to right in table Sex hormones (33167.2)

Early onset familial premature ovarian insufficiency (36851)

Early onset familial premature ovarian insufficiency phenotypes (36906)

Level 3 Title	Gonadal and sex development disorders (36923)
Level 4 Title	Early onset familial premature ovarian insufficiency (36851)
Phenotypes	Entries ordered left to right in table

Premature ovarian failure (HP:0008209)	Primary ovarian failure (HP:0001587)	Primary amenorrhea (HP:0000786)
Secondary amenorrhea (HP:0000869)	Increased circulating gonadotropin level (HP:0000837)	Elevated follicle stimulating hormone (HP:0008232)
Hypergonadotropic hypogonadism (HP:0000815)	Female hypogonadism (HP:0000134)	Primary gonadal insufficiency (HP:0008193)
Gonadal dysgenesis (HP:0000133)	Hypoplasia of the ovary (HP:0008724)	Aplasia of the ovary (HP:0010463)
Streak ovary (HP:0010464)	Delayed puberty (HP:0000823)	Osteopenia (HP:0000938)
Osteoporosis (HP:0000939)	Generalized osteoporosis (HP:0040160)	Hyperlipidemia (HP:0003077)
Dementia (HP:0000726)	Headache (HP:0002315)	Memory impairment (HP:0002354)
Female infertility (HP:0008222)	Decreased fertility in females (HP:0000868)	Depression (HP:0000716)
Irritability (HP:0000737)	Anxiety (HP:0000739)	Sleep disturbance (HP:0002360)
Female sexual dysfunction (HP:0030014)	Decreased female libido (HP:0030018)	Dyspareunia (HP:0030016)
Night sweats (HP:0030166)	Palpitations (HP:0001962)	Arthralgia (HP:0002829)
Myalgia (HP:0003326)	Fatigue (HP:0012378)	Heat intolerance (HP:0002046)

Early onset familial premature ovarian insufficiency clinical tests (36878)

<table>
<tr><td>Level 3 Title</td><td>Gonadal and sex development disorders (36923)</td></tr>
<tr><td>Level 4 Title</td><td>Early onset familial premature ovarian insufficiency (36851)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
General imaging diagnostics refers to pelvic ultrasound scan and DEXA scan
Entries ordered left to right in table
<table><tr><td>Autoantibodies (30112.2)</td><td>Bone profile (30317.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr><tr><td>Sex hormones (33167.2)</td><td></td><td></td></tr></table></td></tr>
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Growth hormone disorders (10971)

IUGR and IGF abnormalities (11057)

IUGR phenotypes (29230)

Level 3 Title	Growth hormone disorders (10971)
Level 4 Title	IUGR and IGF abnormalities (11057)
Phenotypes	Entries ordered left to right in table

Intrauterine growth retardation (HP:0001511)	Failure to thrive (HP:0001508)	Short stature (HP:0004322)
Cryptorchidism (HP:0000028)	Hypospadias (HP:0000047)	Micropenis (HP:0000054)
Delayed puberty (HP:0000823)	Precocious puberty (HP:0000826)	Premature adrenarche (HP:0012412)
Abnormality of blood and blood-forming tissues (HP:0001871)	Abnormality of B cells (HP:0002846)	Abnormality of T cells (HP:0002843)
Recurrent bacterial infections (HP:0002718)	Recurrent viral infections (HP:0004429)	Recurrent fungal infections (HP:0002841)
Recurrent respiratory infections (HP:0002205)	Recurrent otitis media (HP:0000403)	Recurrent urinary tract infections (HP:0000010)
Anosmia (HP:0000458)	Hearing impairment (HP:0000365)	Visual impairment (HP:0000505)
Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)	Delayed speech and language development (HP:0000750)
Autistic behavior (HP:0000729)	Specific learning disability (HP:0001328)	Attention deficit hyperactivity disorder (HP:0007018)
Hemiplegia (HP:0002301)	Seizures (HP:0001250)	

IUGR clinical tests (30873)

Level 3 Title	Growth hormone disorders (10971)
Level 4 Title	IUGR and IGF abnormalities (11057)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain / pituitary and/or skeletal survey Renal and urine biochemistry results should include paired plasma and urine osmolallities Additional body measurements refers to height at presentation, final height and parental heights Entries ordered left to right in table

Renal biochemistry (30355.2)	Additional body measurements (30247.2)	Liver biochemistry (30328.2)
Full Blood Count (30318.2)	Coeliac antibodies (33145.1)	Growth hormones (33154.1)
Thyroid function testing (33168.1)	Bone profile (30317.2)	Hormones (other) (33155.1)
Sex hormones (33167.2)	Urine electrolytes (33187.2)	General Imaging Diagnostics (33633.1)
Weight (30256.2)		

Idiopathic hypogonadotropic hypogonadism (41827)

Idiopathic hypogonadotropic hypogonadism phenotypes (42105)

Level 3 Title	Hypothalamic and pituitary disorders (42204)
Level 4 Title	Idiopathic hypogonadotropic hypogonadism (41827)
Phenotypes	Entries ordered left to right in table

Anosmia (HP:0000458)	Delayed puberty (HP:0000823)	Primary amenorrhea (HP:0000786)
Decreased testosterone in males (HP:0008230)	Micropenis (HP:0000054)	Cryptorchidism (HP:0000028)
Azoospermia (HP:0000027)	Oligospermia (HP:0000798)	Cleft upper lip (HP:0000204)
Cleft hard palate (HP:0410005)	Cleft soft palate (HP:0000185)	Bifid uvula (HP:0000193)
Sensorineural hearing impairment (HP:0000407)	Agenesis of corpus callosum (HP:0001274)	

Idiopathic hypogonadotropic hypogonadism clinical tests (41830)

Level 3 Title	Hypothalamic and pituitary disorders (42204)
Level 4 Title	Idiopathic hypogonadotropic hypogonadism (41827)
Clinical Tests	Clinical test guidance: General imaging diagnostics refers to cerebral imaging and any other relevant imaging performed Entries ordered left to right in table

Hormones (other) (33155.1)	Sex hormones (33167.2)	General Imaging Diagnostics (33633.1)

Obesity syndromes (10973)

Significant early-onset obesity with or without other endocrine features and short stature (11060)

Obesity phenotypes (27789)

Level 3 Title	Obesity syndromes (10973)
Level 4 Title	Significant early-onset obesity with or without other endocrine features and short stature (11060)
Phenotypes	Entries ordered left to right in table

Obesity (HP:0001513)	Truncal obesity (HP:0001956)	Polyphagia (HP:0002591)
Abnormal eating behavior (HP:0100738)	Diabetes mellitus (HP:0000819)	Delayed menarche (HP:0012569)
Delayed puberty (HP:0000823)	Precocious puberty (HP:0000826)	Acanthosis nigricans (HP:0000956)
Abnormal facial shape (HP:0001999)	Delayed ossification of carpal bones (HP:0001216)	Advanced ossification of carpal bones (HP:0004233)
Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)	Macrocephaly (HP:0000256)
Microcephaly (HP:0000252)	Short stature (HP:0004322)	Tall stature (HP:0000098)
Abnormality of the eye (HP:0000478)	Retinopathy (HP:0000488)	Polydactyly (HP:0010442)
Seizures (HP:0001250)	Hearing impairment (HP:0000365)	Failure to thrive in infancy (HP:0001531)
Immunodeficiency (HP:0002721)	Abnormality of the skeletal system (HP:0000924)	Abnormality of the endocrine system (HP:0000818)
Abnormal heart morphology (HP:0001627)	Abnormality of the cerebrum (HP:0002060)	Abnormal renal morphology (HP:0012210)

Obesity clinical tests (30883)

<table>
<tr><td>Level 3 Title</td><td>Obesity syndromes (10973)</td></tr>
<tr><td>Level 4 Title</td><td>Significant early-onset obesity with or without other endocrine features and short stature (11060)</td></tr>
<tr><td>Clinical Tests</td><td>Entries ordered left to right in table
<table><tr><td>Renal biochemistry (30355.2)</td><td>Glucose (33153.1)</td><td>Lipids (33158.1)</td></tr><tr><td>Insulin and C-peptide (33157.1)</td><td>Thyroid function testing (33168.1)</td><td>Cortisol (33148.1)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td></td><td></td></tr></table></td></tr>
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Rare subtypes of diabetes (15099)

Familial young-onset non-insulin-dependent diabetes (15103)

Diabetes phenotypes (30556)

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Familial young-onset non-insulin-dependent diabetes (15103)
Phenotypes	Entries ordered left to right in table

Diabetes mellitus (HP:0000819)	Neonatal insulin-dependent diabetes mellitus (HP:0000857)	Neonatal hypoglycemia (HP:0001998)
Obesity (HP:0001513)	Loss of subcutaneous adipose tissue in limbs (HP:0003635)	Generalized lipodystrophy (HP:0009064)
Acanthosis nigricans (HP:0000956)	Abnormal renal morphology (HP:0012210)	Multiple renal cysts (HP:0005562)
Abnormality of the genitourinary system (HP:0000119)	Optic atrophy (HP:0000648)	Abnormality of the retina (HP:0000479)
Abnormality of the extraocular muscles (HP:0008049)	Sensorineural hearing impairment (HP:0000407)	Cardiomyopathy (HP:0001638)
Abnormality of cardiovascular system morphology (HP:0030680)	Abnormality of the cerebrum (HP:0002060)	Seizures (HP:0001250)
Myopathy (HP:0003198)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Autoimmune antibody positivity (HP:0030057)	Hypothyroidism (HP:0000821)	Primary adrenal insufficiency (HP:0008207)
Vitiligo (HP:0001045)	Malabsorption of Vitamin B12 (HP:0200118)	

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Familial young-onset non-insulin-dependent diabetes (15103)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to abdominal imaging Entries ordered left to right in table

Pancreatic autoantibodies (30337.2)	Liver biochemistry (30328.2)	Glucose (33153.1)
Insulin and C-peptide (33157.1)	Lipids (33158.1)	Fecal test (33308.1)
General Imaging Diagnostics (33633.1)		

Hyperinsulinism (15105)

Hyperinsulinism phenotypes (27782)

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Hyperinsulinism (15105)
Phenotypes	Entries ordered left to right in table

Hyperinsulinemia (HP:0000842)	Hyperinsulinemic hypoglycemia (HP:0000825)	Large for gestational age (HP:0001520)
Hyperammonemia (HP:0001987)	Abnormality of metabolism/homeostasis (HP:0001939)	Overgrowth (HP:0001548)

Hyperinsulinism clinical tests (30875)

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Hyperinsulinism (15105)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to abdominal imaging Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	Insulin and C-peptide (33157.1)	Glucose (33153.1)

Neonatal diabetes (diagnosed less than 6 months) (30553)

Diabetes phenotypes (30556)

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Neonatal diabetes (diagnosed less than 6 months) (30553)
Phenotypes	Entries ordered left to right in table

Diabetes mellitus (HP:0000819)	Neonatal insulin-dependent diabetes mellitus (HP:0000857)	Neonatal hypoglycemia (HP:0001998)
Obesity (HP:0001513)	Loss of subcutaneous adipose tissue in limbs (HP:0003635)	Generalized lipodystrophy (HP:0009064)
Acanthosis nigricans (HP:0000956)	Abnormal renal morphology (HP:0012210)	Multiple renal cysts (HP:0005562)
Abnormality of the genitourinary system (HP:0000119)	Optic atrophy (HP:0000648)	Abnormality of the retina (HP:0000479)
Abnormality of the extraocular muscles (HP:0008049)	Sensorineural hearing impairment (HP:0000407)	Cardiomyopathy (HP:0001638)
Abnormality of cardiovascular system morphology (HP:0030680)	Abnormality of the cerebrum (HP:0002060)	Seizures (HP:0001250)
Myopathy (HP:0003198)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Autoimmune antibody positivity (HP:0030057)	Hypothyroidism (HP:0000821)	Primary adrenal insufficiency (HP:0008207)
Vitiligo (HP:0001045)	Malabsorption of Vitamin B12 (HP:0200118)	

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Neonatal diabetes (diagnosed less than 6 months) (30553)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to abdominal imaging Entries ordered left to right in table

Pancreatic autoantibodies (30337.2)	Liver biochemistry (30328.2)	Glucose (33153.1)
Insulin and C-peptide (33157.1)	Lipids (33158.1)	Fecal test (33308.1)
General Imaging Diagnostics (33633.1)		

Diabetes with additional phenotypes suggestive of a monogenic aetiology (30559)

Diabetes phenotypes (30556)

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Diabetes with additional phenotypes suggestive of a monogenic aetiology (30559)
Phenotypes	Entries ordered left to right in table

Diabetes mellitus (HP:0000819)	Neonatal insulin-dependent diabetes mellitus (HP:0000857)	Neonatal hypoglycemia (HP:0001998)
Obesity (HP:0001513)	Loss of subcutaneous adipose tissue in limbs (HP:0003635)	Generalized lipodystrophy (HP:0009064)
Acanthosis nigricans (HP:0000956)	Abnormal renal morphology (HP:0012210)	Multiple renal cysts (HP:0005562)
Abnormality of the genitourinary system (HP:0000119)	Optic atrophy (HP:0000648)	Abnormality of the retina (HP:0000479)
Abnormality of the extraocular muscles (HP:0008049)	Sensorineural hearing impairment (HP:0000407)	Cardiomyopathy (HP:0001638)
Abnormality of cardiovascular system morphology (HP:0030680)	Abnormality of the cerebrum (HP:0002060)	Seizures (HP:0001250)
Myopathy (HP:0003198)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Autoimmune antibody positivity (HP:0030057)	Hypothyroidism (HP:0000821)	Primary adrenal insufficiency (HP:0008207)
Vitiligo (HP:0001045)	Malabsorption of Vitamin B12 (HP:0200118)	

Diabetes clinical tests (30877)

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Diabetes with additional phenotypes suggestive of a monogenic aetiology (30559)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to abdominal imaging Entries ordered left to right in table

Pancreatic autoantibodies (30337.2)	Liver biochemistry (30328.2)	Glucose (33153.1)
Insulin and C-peptide (33157.1)	Lipids (33158.1)	Fecal test (33308.1)
General Imaging Diagnostics (33633.1)		

Insulin resistance (including lipodystrophy) (30561)

Diabetes phenotypes (30556)

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Insulin resistance (including lipodystrophy) (30561)
Phenotypes	Entries ordered left to right in table

Diabetes mellitus (HP:0000819)	Neonatal insulin-dependent diabetes mellitus (HP:0000857)	Neonatal hypoglycemia (HP:0001998)
Obesity (HP:0001513)	Loss of subcutaneous adipose tissue in limbs (HP:0003635)	Generalized lipodystrophy (HP:0009064)
Acanthosis nigricans (HP:0000956)	Abnormal renal morphology (HP:0012210)	Multiple renal cysts (HP:0005562)
Abnormality of the genitourinary system (HP:0000119)	Optic atrophy (HP:0000648)	Abnormality of the retina (HP:0000479)
Abnormality of the extraocular muscles (HP:0008049)	Sensorineural hearing impairment (HP:0000407)	Cardiomyopathy (HP:0001638)
Abnormality of cardiovascular system morphology (HP:0030680)	Abnormality of the cerebrum (HP:0002060)	Seizures (HP:0001250)
Myopathy (HP:0003198)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Autoimmune antibody positivity (HP:0030057)	Hypothyroidism (HP:0000821)	Primary adrenal insufficiency (HP:0008207)
Vitiligo (HP:0001045)	Malabsorption of Vitamin B12 (HP:0200118)	

<table>
<tr><td>Level 3 Title</td><td>Rare subtypes of diabetes (15099)</td></tr>
<tr><td>Level 4 Title</td><td>Insulin resistance (including lipodystrophy) (30561)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to abdominal imaging
Entries ordered left to right in table
<table><tr><td>Pancreatic autoantibodies (30337.2)</td><td>Liver biochemistry (30328.2)</td><td>Glucose (33153.1)</td></tr><tr><td>Insulin and C-peptide (33157.1)</td><td>Lipids (33158.1)</td><td>Fecal test (33308.1)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td></td><td></td></tr></table></td></tr>
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Multi-organ autoimmune diabetes (30563)

Diabetes phenotypes (30556)

Level 3 Title	Rare subtypes of diabetes (15099)
Level 4 Title	Multi-organ autoimmune diabetes (30563)
Phenotypes	Entries ordered left to right in table

Diabetes mellitus (HP:0000819)	Neonatal insulin-dependent diabetes mellitus (HP:0000857)	Neonatal hypoglycemia (HP:0001998)
Obesity (HP:0001513)	Loss of subcutaneous adipose tissue in limbs (HP:0003635)	Generalized lipodystrophy (HP:0009064)
Acanthosis nigricans (HP:0000956)	Abnormal renal morphology (HP:0012210)	Multiple renal cysts (HP:0005562)
Abnormality of the genitourinary system (HP:0000119)	Optic atrophy (HP:0000648)	Abnormality of the retina (HP:0000479)
Abnormality of the extraocular muscles (HP:0008049)	Sensorineural hearing impairment (HP:0000407)	Cardiomyopathy (HP:0001638)
Abnormality of cardiovascular system morphology (HP:0030680)	Abnormality of the cerebrum (HP:0002060)	Seizures (HP:0001250)
Myopathy (HP:0003198)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Autoimmune antibody positivity (HP:0030057)	Hypothyroidism (HP:0000821)	Primary adrenal insufficiency (HP:0008207)
Vitiligo (HP:0001045)	Malabsorption of Vitamin B12 (HP:0200118)	

<table>
<tr><td>Level 3 Title</td><td>Rare subtypes of diabetes (15099)</td></tr>
<tr><td>Level 4 Title</td><td>Multi-organ autoimmune diabetes (30563)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to abdominal imaging
Entries ordered left to right in table
<table><tr><td>Pancreatic autoantibodies (30337.2)</td><td>Liver biochemistry (30328.2)</td><td>Glucose (33153.1)</td></tr><tr><td>Insulin and C-peptide (33157.1)</td><td>Lipids (33158.1)</td><td>Fecal test (33308.1)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td></td><td></td></tr></table></td></tr>
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Congenital hypothyroidism (41908)

Congenital hypothyroidism phenotypes (42116)

Level 3 Title	Thyroid disorders (42208)
Level 4 Title	Congenital hypothyroidism (41908)
Phenotypes	Entries ordered left to right in table

Primary hypothyroidism (HP:0000832)	Central hypothyroidism (HP:0011787)	Hypothalamic hypothyroidism (HP:0008237)
Pituitary hypothyroidism (HP:0008245)	Thyroid agenesis (HP:0008191)	Thyroid hypoplasia (HP:0005990)
Ectopic thyroid (HP:0100028)	Goiter (HP:0000853)	Thyroid defect in oxidation and organification of iodide (HP:0008263)
Thyroid-stimulating hormone excess (HP:0002925)	Decreased T3/T4 ratio (HP:0012560)	Increased T3/T4 ratio (HP:0012559)
Increased serum free triiodothyronine (HP:0011788)	Thyroid hormone receptor defect (HP:0002930)	

Congenital hypothyroidism clinical tests (41910)

Level 3 Title	Thyroid disorders (42208)
Level 4 Title	Congenital hypothyroidism (41908)
Clinical Tests	Clinical test guidance: General Imaging Diagnostics refers to Thyroid ultrasound Entries ordered left to right in table

Thyroid function testing (33168.1)	General Imaging Diagnostics (33633.1)

Resistance to thyroid hormone (41916)

Resistance to thyroid hormone phenotypes (42226)

Level 3 Title	Thyroid disorders (42208)
Level 4 Title	Resistance to thyroid hormone (41916)
Phenotypes	Entries ordered left to right in table

Primary hypothyroidism (HP:0000832)	Central hypothyroidism (HP:0011787)	Hypothalamic hypothyroidism (HP:0008237)
Pituitary hypothyroidism (HP:0008245)	Thyroid agenesis (HP:0008191)	Thyroid hypoplasia (HP:0005990)
Ectopic thyroid (HP:0100028)	Goiter (HP:0000853)	Thyroid defect in oxidation and organification of iodide (HP:0008263)
Thyroid-stimulating hormone excess (HP:0002925)	Decreased T3/T4 ratio (HP:0012560)	Increased T3/T4 ratio (HP:0012559)
Increased serum free triiodothyronine (HP:0011788)	Thyroid hormone receptor defect (HP:0002930)	

Resistance to thyroid hormone clinical tests (41918)

Level 3 Title	Thyroid disorders (42208)
Level 4 Title	Resistance to thyroid hormone (41916)
Clinical Tests	Clinical test guidance: General Imaging Diagnostics refers to Thyroid ultrasound Entries ordered left to right in table

Thyroid function testing (33168.1)	General Imaging Diagnostics (33633.1)

Gastroenterological disorders (38581)

Gastrointestinal disorders (38582)

Infantile enterocolitis and monogenic inflammatory bowel disease (37490)

Infantile enterocolitis and monogenic inflammatory bowel disease phenotypes (37496)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Infantile enterocolitis and monogenic inflammatory bowel disease (37490)
Phenotypes	Entries ordered left to right in table

Crohn's disease (HP:0100280)	Non-caseating epithelioid cell granulomatosis (HP:0012220)	Ulcerative colitis (HP:0100279)
Perianal abscess (HP:0009789)	Intestinal fistula (HP:0100819)	Gastrointestinal obstruction (HP:0004796)
Intestinal obstruction (HP:0005214)	Neonatal intestinal obstruction (HP:0005234)	Localized skin lesion (HP:0011355)
Recurrent infections (HP:0002719)	Fever (HP:0001945)	Splenomegaly (HP:0001744)
Lymphadenopathy (HP:0002716)	Hemophagocytosis (HP:0012156)	Neutropenia (HP:0001875)
Absence of bactericidal oxidative 'respiratory burst' in phagocytes (HP:0002723)	Lymphopenia (HP:0001888)	Abnormality of natural killer cells (HP:0012176)
Decreased antibody level in blood (HP:0004313)	Increased IgM level (HP:0003496)	Increased IgE level (HP:0003212)
Abnormal bleeding (HP:0001892)	Neoplasm (HP:0002664)	

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Infantile enterocolitis and monogenic inflammatory bowel disease (37490)
Clinical Tests	NB. Clinical test guidance: General biopsy refers to any relevant gastrointestingal biopsy results Entries ordered left to right in table

Autoantibodies (30112.2)	General Biopsy (33614.1)

Gastrointestinal epithelial barrier disorders (37772)

Gastrointestinal epithelial barrier disorders phenotypes (40234)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Gastrointestinal epithelial barrier disorders (37772)
Phenotypes	Entries ordered left to right in table

Failure to thrive (HP:0001508)	Growth delay (HP:0001510)	Polyhydramnios (HP:0001561)
Intractable diarrhea (HP:0002041)	Abdominal distention (HP:0003270)	Protein-losing enteropathy (HP:0002243)
Inflammation of the large intestine (HP:0002037)	Enterocolitis (HP:0004387)	Abdominal pain (HP:0002027)
Intestinal atresia (HP:0011100)	Malnutrition (HP:0004395)	Small for gestational age (HP:0001518)
Villous atrophy (HP:0011473)	Autoimmunity (HP:0002960)	Immunodeficiency (HP:0002721)
Increased inflammatory response (HP:0012649)	Elevated erythrocyte sedimentation rate (HP:0003565)	Negative nitroblue tetrazolium reduction test (HP:0003203)

Gastrointestinal epithelial barrier disorders clinical tests (37774)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Gastrointestinal epithelial barrier disorders (37772)
Clinical Tests	NB. Clinical test guidance: General biopsy refers to biopsy of abnormal intestinal features Inflammatory markers refers to C reactive protein General Imaging Diagnostics refers to Endoscopy Entries ordered left to right in table

General Biopsy (33614.1)	Primary immunodeficiency investigations (33165.1)	General Imaging Diagnostics (33633.1)
Renal biochemistry (30355.2)	Full Blood Count (30318.2)	Liver biochemistry (30328.2)
Bone profile (30317.2)	Inflammatory markers (33156.1)	Serum immunoglobulins (30338.2)
Fecal test (33308.1)		

Non-syndromic familial congenital anorectal malformations (41868)

Non-syndromic familial congenital anorectal malformations phenotypes (42110)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Non-syndromic familial congenital anorectal malformations (41868)
Phenotypes	Entries ordered left to right in table

Perineal fistula (HP:0004871)	Rectoperineal fistula (HP:0004792)	Urogenital fistula (HP:0100589)
Anal atresia (HP:0002023)	Rectal atresia (HP:0025023)	Rectovestibular fistula (HP:0025025)
Persistent cloaca (HP:0012621)	Rectovaginal fistula (HP:0000143)	Meconium ileus (HP:0004401)
Chronic constipation (HP:0012450)	Bowel incontinence (HP:0002607)	Urinary incontinence (HP:0000020)
Megarectum (HP:0025024)	Lipoma (HP:0012032)	Tethered cord (HP:0002144)
Myelomeningocele (HP:0002475)	Syringomyelia (HP:0003396)	Aplasia/Hypoplasia of the sacrum (HP:0008517)
Premature birth (HP:0001622)	Small for gestational age (HP:0001518)	Abnormal facial shape (HP:0001999)

Non-syndromic familial congenital anorectal malformations clinical tests (41870)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Non-syndromic familial congenital anorectal malformations (41868)
Clinical Tests	Clinical test guidance: General Imaging Diagnostics refers to abdomen, genitourinary system, dynamic bladder and endoanal ultrasound ; pelvic and spinal MRI; and lumbar sacral, pelvic and other skeletal radiographs Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Early onset or familial intestinal pseudo obstruction (41876)

Early onset or familial intestinal pseudo obstruction phenotypes (42111)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Early onset or familial intestinal pseudo obstruction (41876)
Phenotypes	Entries ordered left to right in table

Intestinal pseudo-obstruction (HP:0004389)	Functional intestinal obstruction (HP:0005249)	Gastrointestinal dysmotility (HP:0002579)
Myopathy (HP:0003198)	Inflammatory myopathy (HP:0009071)	Mitochondrial myopathy (HP:0003737)
Infantile muscular hypotonia (HP:0008947)	Peripheral neuropathy (HP:0009830)	External ophthalmoplegia (HP:0000544)
Pyloric stenosis (HP:0002021)	Intestinal malrotation (HP:0002566)	Hypoplasia of the small intestine (HP:0004790)
Aganglionosis of the small intestine (HP:0011464)	Aganglionic megacolon (HP:0002251)	Enteric neuronal degeneration (HP:0025030)
Enteric intraneuronal nuclear inclusion bodies (HP:0030938)	Atrophic muscularis propria (HP:0025149)	Abnormal layering of muscularis propria (HP:0030936)
Nasogastric tube feeding in infancy (HP:0011470)	Dependency on intravenous nutrition (HP:0025156)	Feeding difficulties (HP:0011968)
Failure to thrive (HP:0001508)	Nausea (HP:0002018)	Vomiting (HP:0002013)
Abdominal distention (HP:0003270)	Abdominal pain (HP:0002027)	Meconium ileus (HP:0004401)
Constipation (HP:0002019)	Diarrhea (HP:0002014)	Fetal megacystis (HP:0010956)
Megacystis (HP:0000021)	Hydronephrosis (HP:0000126)	Congenital megaureter (HP:0008676)
Recurrent urinary tract infections (HP:0000010)	Intellectual disability (HP:0001249)	Spasticity (HP:0001257)
Seizures (HP:0001250)	Polymicrogyria (HP:0002126)	Periventricular gray matter heterotopia (HP:0007165)
Cerebellar hypoplasia (HP:0001321)		

Early onset or familial intestinal pseudo obstruction clinical tests (41878)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Early onset or familial intestinal pseudo obstruction (41876)
Clinical Tests	Clinical test guidance: General Imaging Diagnostics refers to plain x-ray, MRI brain, ultrasound of urinary tract and facial imaging General biopsy refers to skeletal muscle biopsy Entries ordered left to right in table

General Biopsy (33614.1)	General Imaging Diagnostics (33633.1)

Familial Hirschsprung Disease (55463)

Familial Hirschsprung Disease phenotypes (68113)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Familial Hirschsprung Disease (55463)
Phenotypes	Entries ordered left to right in table

Abnormality of enteric ganglion morphology (HP:0004362)	Total intestinal aganglionosis (HP:0005241)	Total colonic aganglionosis (HP:0011286)
Short-segment aganglionic megacolon (HP:0011284)	Long-segment aganglionic megacolon (HP:0011285)	Intestinal atresia (HP:0011100)
Sensorineural hearing impairment (HP:0000407)	Hypopigmented skin patches (HP:0001053)	Intellectual disability (HP:0001249)
Microcephaly (HP:0000252)	Aplasia/Hypoplasia of the corpus callosum (HP:0007370)	Abnormal heart morphology (HP:0001627)
Cleft palate (HP:0000175)	Central hypoventilation (HP:0007110)	Dysautonomia (HP:0002459)
Medullary thyroid carcinoma (HP:0002865)	Aqueductal stenosis (HP:0002410)	

Familial Hirschsprung Disease clinical tests (55464)

Level 3 Title	Gastrointestinal disorders (38582)
Level 4 Title	Familial Hirschsprung Disease (55463)
Clinical Tests	Entries ordered left to right in table

Liver disease (55663)

Ductal plate malformation (55469)

Ductal plate malformation phenotypes (68114)

Level 3 Title	Liver disease (55663)
Level 4 Title	Ductal plate malformation (55469)
Phenotypes	Entries ordered left to right in table

Malformation of the hepatic ductal plate (HP:0006563)	Hepatomegaly (HP:0002240)	Splenomegaly (HP:0001744)
Ascites (HP:0001541)	Portal hypertension (HP:0001409)	Esophageal varix (HP:0002040)
Intrahepatic cholestasis (HP:0001406)	Hepatic encephalopathy (HP:0002480)	Polycystic liver disease (HP:0006557)
Polycystic kidney dysplasia (HP:0000113)	Renal cyst (HP:0000107)	Cyst of the ductus choledochus (HP:0100890)
Congenital hepatic fibrosis (HP:0002612)	Macronodular cirrhosis (HP:0006577)	Biliary hyperplasia (HP:0006560)
Intrahepatic bile duct cysts (HP:0005209)		

Ductal plate malformation clinical tests (55470)

Level 3 Title	Liver disease (55663)
Level 4 Title	Ductal plate malformation (55469)
Clinical Tests	NB Clinical testing guidance: General Imaging Diagnostics refers to MRI Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Neonatal cholestasis (71744)

Neonatal cholestasis phenotypes (71769)

Level 3 Title	Liver disease (55663)
Level 4 Title	Neonatal cholestasis (71744)
Phenotypes	Entries ordered left to right in table

Intrahepatic cholestasis (HP:0001406)	Conjugated hyperbilirubinemia (HP:0002908)	Cirrhosis (HP:0001394)
Hepatic failure (HP:0001399)	Hepatomegaly (HP:0002240)	Abnormality of coagulation (HP:0001928)
Diabetes mellitus (HP:0000819)	Diarrhea (HP:0002014)	Abnormality of metabolism/homeostasis (HP:0001939)
Abnormal facial shape (HP:0001999)	Exocrine pancreatic insufficiency (HP:0001738)	Abnormality of pancreas morphology (HP:0012090)
Intestinal atresia (HP:0011100)	Polysplenia (HP:0001748)	Asplenia (HP:0001746)
Splenomegaly (HP:0001744)	Failure to thrive (HP:0001508)	Abnormal heart morphology (HP:0001627)
Abnormal eye morphology (HP:0012372)	Abnormal renal morphology (HP:0012210)	Abnormality of the genital system (HP:0000078)
Abnormality of the vertebrae (HP:0003468)	Cleft palate (HP:0000175)	Situs inversus totalis (HP:0001696)
Hypoglycemia (HP:0001943)		

Neonatal cholestasis clinical tests (71747)

Level 3 Title	Liver disease (55663)
Level 4 Title	Neonatal cholestasis (71744)
Clinical Tests	NB. Clinical Test Guidance: Liver biochemistry tests refers to Gamma-glutamyl transferase (GGT) result Entries ordered left to right in table

Liver biochemistry (30328.2)

Growth disorders (10974)

Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)

Classical Beckwith-Wiedemann syndrome (11063)

Overgrowth phenotypes (27790)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Classical Beckwith-Wiedemann syndrome (11063)
Phenotypes	Entries ordered left to right in table

Overgrowth (HP:0001548)	Preeclampsia (HP:0100602)	Polyhydramnios (HP:0001561)
Large placenta (HP:0006267)	Neonatal hypoglycemia (HP:0001998)	Feeding difficulties in infancy (HP:0008872)
Apnea (HP:0002104)	Apneic episodes in infancy (HP:0005949)	Cyanosis (HP:0000961)
Accelerated skeletal maturation (HP:0005616)	Asymmetric growth (HP:0100555)	Hemihypertrophy (HP:0001528)
Facial asymmetry (HP:0000324)	Hemihypertrophy of upper limb (HP:0100554)	Asymmetry of the thorax (HP:0001555)
Hemihypertrophy of lower limb (HP:0100553)	Nevus flammeus (HP:0001052)	Macroglossia (HP:0000158)
Mandibular prognathia (HP:0000303)	Prominent occiput (HP:0000269)	Aplasia/Hypoplasia of the maxilla (HP:0009117)
Anterior creases of earlobe (HP:0009908)	Posterior helix pit (HP:0008523)	Cleft palate (HP:0000175)
Polycythemia (HP:0001901)	Ventricular septal defect (HP:0001629)	Atrial septal defect (HP:0001631)
Tetralogy of Fallot (HP:0001636)	Coarctation of aorta (HP:0001680)	Cardiomyopathy (HP:0001638)
Abdominal wall defect (HP:0010866)	Omphalocele (HP:0001539)	Umbilical hernia (HP:0001537)
Malrotation of small bowel (HP:0004794)	Enlarged kidney (HP:0000105)	Splenomegaly (HP:0001744)
Hepatomegaly (HP:0002240)	Hypospadias (HP:0000047)	Cryptorchidism (HP:0000028)

	Inguinal hernia (HP:0000023)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
	Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)
	Specific learning disability (HP:0001328)	Autistic behavior (HP:0000729)	Attention deficit hyperactivity disorder (HP:0007018)
	Abnormal social behavior (HP:0012433)	Abnormal emotion/affect behavior (HP:0100851)	Visual impairment (HP:0000505)
	Hearing impairment (HP:0000365)	Seizures (HP:0001250)	Nephroblastoma (HP:0002667)
	Neuroblastoma (HP:0003006)	Hepatoblastoma (HP:0002884)	Neoplasm of the adrenal cortex (HP:0100641)
	Rhabdomyosarcoma (HP:0002859)		

Beckwith-Wiedemann syndrome clinical tests (30884)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Classical Beckwith-Wiedemann syndrome (11063)
Clinical Tests	NB. Clinical test guidance: Additional body measurements refers to parental heights and head circumferences Entries ordered left to right in table Additional body measurements (30247.2)

Atypical Beckwith-Wiedemann syndrome (11064)

Overgrowth phenotypes (27790)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Atypical Beckwith-Wiedemann syndrome (11064)
Phenotypes	Entries ordered left to right in table

Overgrowth (HP:0001548)	Preeclampsia (HP:0100602)	Polyhydramnios (HP:0001561)
Large placenta (HP:0006267)	Neonatal hypoglycemia (HP:0001998)	Feeding difficulties in infancy (HP:0008872)
Apnea (HP:0002104)	Apneic episodes in infancy (HP:0005949)	Cyanosis (HP:0000961)
Accelerated skeletal maturation (HP:0005616)	Asymmetric growth (HP:0100555)	Hemihypertrophy (HP:0001528)
Facial asymmetry (HP:0000324)	Hemihypertrophy of upper limb (HP:0100554)	Asymmetry of the thorax (HP:0001555)
Hemihypertrophy of lower limb (HP:0100553)	Nevus flammeus (HP:0001052)	Macroglossia (HP:0000158)
Mandibular prognathia (HP:0000303)	Prominent occiput (HP:0000269)	Aplasia/Hypoplasia of the maxilla (HP:0009117)
Anterior creases of earlobe (HP:0009908)	Posterior helix pit (HP:0008523)	Cleft palate (HP:0000175)
Polycythemia (HP:0001901)	Ventricular septal defect (HP:0001629)	Atrial septal defect (HP:0001631)
Tetralogy of Fallot (HP:0001636)	Coarctation of aorta (HP:0001680)	Cardiomyopathy (HP:0001638)
Abdominal wall defect (HP:0010866)	Omphalocele (HP:0001539)	Umbilical hernia (HP:0001537)
Malrotation of small bowel (HP:0004794)	Enlarged kidney (HP:0000105)	Splenomegaly (HP:0001744)
Hepatomegaly (HP:0002240)	Hypospadias (HP:0000047)	Cryptorchidism (HP:0000028)
Inguinal hernia (HP:0000023)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)

	Specific learning disability (HP:0001328)	Autistic behavior (HP:0000729)	Attention deficit hyperactivity disorder (HP:0007018)
	Abnormal social behavior (HP:0012433)	Abnormal emotion/affect behavior (HP:0100851)	Visual impairment (HP:0000505)
	Hearing impairment (HP:0000365)	Seizures (HP:0001250)	Nephroblastoma (HP:0002667)
	Neuroblastoma (HP:0003006)	Hepatoblastoma (HP:0002884)	Neoplasm of the adrenal cortex (HP:0100641)
	Rhabdomyosarcoma (HP:0002859)		

Beckwith-Wiedemann syndrome clinical tests (30884)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Atypical Beckwith-Wiedemann syndrome (11064)
Clinical Tests	NB. Clinical test guidance: Additional body measurements refers to parental heights and head circumferences Entries ordered left to right in table Additional body measurements (30247.2)

Simpson-Golabi-Behmel syndrome (11065)

Overgrowth phenotypes (27790)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Simpson-Golabi-Behmel syndrome (11065)
Phenotypes	Entries ordered left to right in table

Overgrowth (HP:0001548)	Preeclampsia (HP:0100602)	Polyhydramnios (HP:0001561)
Large placenta (HP:0006267)	Neonatal hypoglycemia (HP:0001998)	Feeding difficulties in infancy (HP:0008872)
Apnea (HP:0002104)	Apneic episodes in infancy (HP:0005949)	Cyanosis (HP:0000961)
Accelerated skeletal maturation (HP:0005616)	Asymmetric growth (HP:0100555)	Hemihypertrophy (HP:0001528)
Facial asymmetry (HP:0000324)	Hemihypertrophy of upper limb (HP:0100554)	Asymmetry of the thorax (HP:0001555)
Hemihypertrophy of lower limb (HP:0100553)	Nevus flammeus (HP:0001052)	Macroglossia (HP:0000158)
Mandibular prognathia (HP:0000303)	Prominent occiput (HP:0000269)	Aplasia/Hypoplasia of the maxilla (HP:0009117)
Anterior creases of earlobe (HP:0009908)	Posterior helix pit (HP:0008523)	Cleft palate (HP:0000175)
Polycythemia (HP:0001901)	Ventricular septal defect (HP:0001629)	Atrial septal defect (HP:0001631)
Tetralogy of Fallot (HP:0001636)	Coarctation of aorta (HP:0001680)	Cardiomyopathy (HP:0001638)
Abdominal wall defect (HP:0010866)	Omphalocele (HP:0001539)	Umbilical hernia (HP:0001537)
Malrotation of small bowel (HP:0004794)	Enlarged kidney (HP:0000105)	Splenomegaly (HP:0001744)
Hepatomegaly (HP:0002240)	Hypospadias (HP:0000047)	Cryptorchidism (HP:0000028)
Inguinal hernia (HP:0000023)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)

	Specific learning disability (HP:0001328)	Autistic behavior (HP:0000729)	Attention deficit hyperactivity disorder (HP:0007018)
	Abnormal social behavior (HP:0012433)	Abnormal emotion/affect behavior (HP:0100851)	Visual impairment (HP:0000505)
	Hearing impairment (HP:0000365)	Seizures (HP:0001250)	Nephroblastoma (HP:0002667)
	Neuroblastoma (HP:0003006)	Hepatoblastoma (HP:0002884)	Neoplasm of the adrenal cortex (HP:0100641)
	Rhabdomyosarcoma (HP:0002859)		

Beckwith-Wiedemann syndrome clinical tests (30884)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Simpson-Golabi-Behmel syndrome (11065)
Clinical Tests	NB. Clinical test guidance: Additional body measurements refers to parental heights and head circumferences Entries ordered left to right in table Additional body measurements (30247.2)

Sotos syndrome (11066)

Overgrowth phenotypes (27790)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Sotos syndrome (11066)

Phenotypes

Entries ordered left to right in table

Overgrowth (HP:0001548)	Preeclampsia (HP:0100602)	Polyhydramnios (HP:0001561)
Large placenta (HP:0006267)	Neonatal hypoglycemia (HP:0001998)	Feeding difficulties in infancy (HP:0008872)
Apnea (HP:0002104)	Apneic episodes in infancy (HP:0005949)	Cyanosis (HP:0000961)
Accelerated skeletal maturation (HP:0005616)	Asymmetric growth (HP:0100555)	Hemihypertrophy (HP:0001528)
Facial asymmetry (HP:0000324)	Hemihypertrophy of upper limb (HP:0100554)	Asymmetry of the thorax (HP:0001555)
Hemihypertrophy of lower limb (HP:0100553)	Nevus flammeus (HP:0001052)	Macroglossia (HP:0000158)
Mandibular prognathia (HP:0000303)	Prominent occiput (HP:0000269)	Aplasia/Hypoplasia of the maxilla (HP:0009117)
Anterior creases of earlobe (HP:0009908)	Posterior helix pit (HP:0008523)	Cleft palate (HP:0000175)
Polycythemia (HP:0001901)	Ventricular septal defect (HP:0001629)	Atrial septal defect (HP:0001631)
Tetralogy of Fallot (HP:0001636)	Coarctation of aorta (HP:0001680)	Cardiomyopathy (HP:0001638)
Abdominal wall defect (HP:0010866)	Omphalocele (HP:0001539)	Umbilical hernia (HP:0001537)
Malrotation of small bowel (HP:0004794)	Enlarged kidney (HP:0000105)	Splenomegaly (HP:0001744)
Hepatomegaly (HP:0002240)	Hypospadias (HP:0000047)	Cryptorchidism (HP:0000028)
Inguinal hernia (HP:0000023)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)

	Specific learning disability (HP:0001328)	Autistic behavior (HP:0000729)	Attention deficit hyperactivity disorder (HP:0007018)
	Abnormal social behavior (HP:0012433)	Abnormal emotion/affect behavior (HP:0100851)	Visual impairment (HP:0000505)
	Hearing impairment (HP:0000365)	Seizures (HP:0001250)	Nephroblastoma (HP:0002667)
	Neuroblastoma (HP:0003006)	Hepatoblastoma (HP:0002884)	Neoplasm of the adrenal cortex (HP:0100641)
	Rhabdomyosarcoma (HP:0002859)		

Beckwith-Wiedemann syndrome clinical tests (30884)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Sotos syndrome (11066)
Clinical Tests	NB. Clinical test guidance: Additional body measurements refers to parental heights and head circumferences Entries ordered left to right in table Additional body measurements (30247.2)

Weaver syndrome (11067)

Overgrowth phenotypes (27790)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Weaver syndrome (11067)

Phenotypes

Entries ordered left to right in table

Overgrowth (HP:0001548)	Preeclampsia (HP:0100602)	Polyhydramnios (HP:0001561)
Large placenta (HP:0006267)	Neonatal hypoglycemia (HP:0001998)	Feeding difficulties in infancy (HP:0008872)
Apnea (HP:0002104)	Apneic episodes in infancy (HP:0005949)	Cyanosis (HP:0000961)
Accelerated skeletal maturation (HP:0005616)	Asymmetric growth (HP:0100555)	Hemihypertrophy (HP:0001528)
Facial asymmetry (HP:0000324)	Hemihypertrophy of upper limb (HP:0100554)	Asymmetry of the thorax (HP:0001555)
Hemihypertrophy of lower limb (HP:0100553)	Nevus flammeus (HP:0001052)	Macroglossia (HP:0000158)
Mandibular prognathia (HP:0000303)	Prominent occiput (HP:0000269)	Aplasia/Hypoplasia of the maxilla (HP:0009117)
Anterior creases of earlobe (HP:0009908)	Posterior helix pit (HP:0008523)	Cleft palate (HP:0000175)
Polycythemia (HP:0001901)	Ventricular septal defect (HP:0001629)	Atrial septal defect (HP:0001631)
Tetralogy of Fallot (HP:0001636)	Coarctation of aorta (HP:0001680)	Cardiomyopathy (HP:0001638)
Abdominal wall defect (HP:0010866)	Omphalocele (HP:0001539)	Umbilical hernia (HP:0001537)
Malrotation of small bowel (HP:0004794)	Enlarged kidney (HP:0000105)	Splenomegaly (HP:0001744)
Hepatomegaly (HP:0002240)	Hypospadias (HP:0000047)	Cryptorchidism (HP:0000028)
Inguinal hernia (HP:0000023)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)

	Specific learning disability (HP:0001328)	Autistic behavior (HP:0000729)	Attention deficit hyperactivity disorder (HP:0007018)
	Abnormal social behavior (HP:0012433)	Abnormal emotion/affect behavior (HP:0100851)	Visual impairment (HP:0000505)
	Hearing impairment (HP:0000365)	Seizures (HP:0001250)	Nephroblastoma (HP:0002667)
	Neuroblastoma (HP:0003006)	Hepatoblastoma (HP:0002884)	Neoplasm of the adrenal cortex (HP:0100641)
	Rhabdomyosarcoma (HP:0002859)		

Beckwith-Wiedemann syndrome clinical tests (30884)

Level 3 Title	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)
Level 4 Title	Weaver syndrome (11067)
Clinical Tests	NB. Clinical test guidance: Additional body measurements refers to parental heights and head circumferences Entries ordered left to right in table Additional body measurements (30247.2)

Growth restriction (38585)

Silver Russell syndrome (37553)

Silver Russell syndrome phenotypes (37738)

Level 3 Title	Growth restriction (38585)
Level 4 Title	Silver Russell syndrome (37553)
Phenotypes	Entries ordered left to right in table

Intrauterine growth retardation (HP:0001511)	Oligohydramnios (HP:0001562)	Small placenta (HP:0006266)
Small for gestational age (HP:0001518)	Failure to thrive (HP:0001508)	Feeding difficulties (HP:0011968)
Neonatal hypoglycemia (HP:0001998)	Fasting hypoglycemia (HP:0003162)	Gastroesophageal reflux (HP:0002020)
Relative macrocephaly (HP:0004482)	Prominent forehead (HP:0011220)	Cleft palate (HP:0000175)
Micrognathia (HP:0000347)	Short stature (HP:0004322)	Facial asymmetry (HP:0000324)
Asymmetry of the thorax (HP:0001555)	Upper limb asymmetry (HP:0100560)	Lower limb asymmetry (HP:0100559)
Clinodactyly of the 5th finger (HP:0004209)	Brachydactyly syndrome (HP:0001156)	Camptodactyly of finger (HP:0100490)
Hydronephrosis (HP:0000126)	Micropenis (HP:0000054)	Hypospadias (HP:0000047)
Cryptorchidism (HP:0000028)	Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)
Hydrocephalus (HP:0000238)	Cafe-au-lait spot (HP:0000957)	

Silver Russell syndrome clinical tests (37555)

Level 3 Title	Growth restriction (38585)
Level 4 Title	Silver Russell syndrome (37553)
Clinical Tests	NB. Clinical test guidance: General Imaging Diagnostics refers to hand, facial and other relevant medical photographs Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	Growth hormones (33154.1)

Haematological and immunological disorders (10977)

Anaemias and red cell disorders (10979)

Congenital anaemias (11075)

Anemia phenotypes (29231)

Level 3 Title	Anaemias and red cell disorders (10979)
Level 4 Title	Congenital anaemias (11075)
Phenotypes	Entries ordered left to right in table

Anemia (HP:0001903)	Pancytopenia (HP:0001876)	Sideroblastic anemia (HP:0001924)
Macrocytic dyserythropoietic anemia (HP:0005532)	Thrombocytopenia (HP:0001873)	Leukopenia (HP:0001882)
Prolonged neonatal jaundice (HP:0006579)	Recurrent infections (HP:0002719)	Immunodeficiency (HP:0002721)
Failure to thrive (HP:0001508)	Intellectual disability (HP:0001249)	

Anaemia clinical tests (30889)

Level 3 Title	Anaemias and red cell disorders (10979)
Level 4 Title	Congenital anaemias (11075)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to bone marrow aspirate histology Imaging diagnostics refers to ultrasound of the liver and T2* heart and liver Entries ordered left to right in table

Heart/liver Iron measurement (30137.2)	Full Blood Count (30318.2)	General Biopsy (33614.1)
General Imaging Diagnostics (33633.1)	Extended haematology investigations (33151.1)	Liver biochemistry (30328.2)
Metabolic biochemistry (33160.2)	Vitamin B12 (33172.1)	

Hereditary erythrocytosis (55505)

Hereditary erythrocytosis phenotypes (68121)

Level 3 Title	Anaemias and red cell disorders (10979)
Level 4 Title	Hereditary erythrocytosis (55505)
Phenotypes	Entries ordered left to right in table

Polycythemia (HP:0001901)	Increased hemoglobin (HP:0001900)	Increased hematocrit (HP:0001899)
Increased red blood cell mass (HP:0001898)	Plethora (HP:0001050)	Exertional dyspnea (HP:0002875)
Peripheral thrombosis (HP:0002641)	Abnormal thrombosis (HP:0001977)	Stroke (HP:0001297)
Thromboembolism (HP:0001907)	Hemangioma (HP:0001028)	Superficial thrombophlebitis (HP:0002638)
Cholelithiasis (HP:0001081)	Paraganglioma (HP:0002668)	Pheochromocytoma (HP:0002666)
Erythroid hyperplasia (HP:0012132)	Pulmonary arterial hypertension (HP:0002092)	

Hereditary erythrocytosis clinical tests (55506)

Level 3 Title	Anaemias and red cell disorders (10979)
Level 4 Title	Hereditary erythrocytosis (55505)
Clinical Tests	Entries ordered left to right in table

Primary immunodeficiency disorders (10978)

Primary immunodeficiency (55674)

Primary immunodeficiency phenotypes (68125)

Level 3 Title	Primary immunodeficiency disorders (10978)
Level 4 Title	Primary immunodeficiency (55674)
Phenotypes	Entries ordered left to right in table

Fever (HP:0001945)	Sepsis (HP:0100806)	Recurrent bacterial infections (HP:0002718)
Recurrent fungal infections (HP:0002841)	Recurrent viral infections (HP:0004429)	Severe viral infections (HP:0005364)
Recurrent viral skin infections (HP:0011371)	Recurrent mycobacterial infections (HP:0011274)	Recurrent opportunistic infections (HP:0005390)
Lymphadenopathy (HP:0002716)	Splenomegaly (HP:0001744)	Hepatomegaly (HP:0002240)
Abnormal bleeding (HP:0001892)	Lymphoma (HP:0002665)	Autoimmunity (HP:0002960)
Systemic lupus erythematosus (HP:0002725)	Hypothyroidism (HP:0000821)	Abnormality of the intestine (HP:0002242)
Inflammatory abnormality of the skin (HP:0011123)	Vasculitis (HP:0002633)	Cerebral calcification (HP:0002514)
Abnormal facial shape (HP:0001999)	Microcephaly (HP:0000252)	Intellectual disability (HP:0001249)
Decrease in T cell count (HP:0005403)	B lymphocytopenia (HP:0010976)	Anemia (HP:0001903)
Thrombocytopenia (HP:0001873)	Neutropenia (HP:0001875)	Agammaglobulinemia (HP:0004432)
Abnormality of the coagulation cascade (HP:0003256)	Decreased antibody level in blood (HP:0004313)	Congenital neutropenia (HP:0005549)
Monocytopenia (HP:0012312)	Hemophagocytosis (HP:0012156)	Reduced natural killer cell activity (HP:0012178)
Increased serum ferritin (HP:0003281)	Hypofibrinogenemia (HP:0011900)	Hypertriglyceridemia (HP:0002155)
Urticaria (HP:0001025)	Episodic fever (HP:0001954)	Recurrent aphthous stomatitis (HP:0011107)

	Arthralgia/arthritis (HP:0005059)	Uveitis (HP:0000554)	Amyloidosis (HP:0011034)
	Elevated erythrocyte sedimentation rate (HP:0003565)	Triggered by vaccination (HP:0025219)	Respiratory failure requiring assisted ventilation (HP:0004887)
	Encephalitis (HP:0002383)	Fulminant hepatitis (HP:0004787)	Susceptibility to chickenpox (HP:0005360)
	Severe recurrent varicella (HP:0005428)	Susceptibility to herpesvirus (HP:0005353)	Herpes simplex encephalitis (HP:0012302)
	Kaposi's sarcoma (HP:0100726)	Recurrent enteroviral infections (HP:0002743)	Complement deficiency (HP:0004431)
	Hemolytic-uremic syndrome (HP:0005575)		

Primary immunodeficiency clinical tests (55676)

Level 3 Title	Primary immunodeficiency disorders (10978)
Level 4 Title	Primary immunodeficiency (55674)
Clinical Tests	Entries ordered left to right in table

Haemostasis disorders (55664)

Inherited bleeding and or platelet disorders (55475)

Inherited bleeding and or platelet disorders phenotypes (68116)

Level 3 Title	Haemostasis disorders (55664)
Level 4 Title	Inherited bleeding and or platelet disorders (55475)
Phenotypes	Entries ordered left to right in table

Thrombocytopenia (HP:0001873)	Abnormal platelet volume (HP:0011876)	Abnormal platelet morphology (HP:0011875)
Impaired platelet aggregation (HP:0003540)	Abnormal platelet granule secretion (HP:0030396)	Abnormal platelet membrane protein expression (HP:0011878)
Abnormality of prothrombin (HP:0012200)	Abnormality of the coagulation cascade (HP:0003256)	Abnormality of von Willebrand factor (HP:0012146)
Abnormality of the intrinsic pathway (HP:0010989)	Abnormality of the extrinsic pathway (HP:0010988)	Abnormality of connective tissue (HP:0003549)
Menorrhagia (HP:0000132)	Epistaxis (HP:0000421)	Subcutaneous hemorrhage (HP:0001933)
Post-partum hemorrhage (HP:0011891)	Intracranial hemorrhage (HP:0002170)	Joint hemorrhage (HP:0005261)
Gastrointestinal hemorrhage (HP:0002239)	Abnormal bleeding (HP:0001892)	

Inherited bleeding and or platelet disorders clinical tests (55476)

Level 3 Title	Haemostasis disorders (55664)
Level 4 Title	Inherited bleeding and or platelet disorders (55475)
Clinical Tests	Entries ordered left to right in table

Monogenic venous thrombosis (55523)

Monogenic venous thrombosis phenotypes (68123)

Level 3 Title	Haemostasis disorders (55664)
Level 4 Title	Monogenic venous thrombosis (55523)
Phenotypes	Entries ordered left to right in table

Deep venous thrombosis (HP:0002625)	Cerebral venous thrombosis (HP:0005305)	Splanchnic vein thrombosis (HP:0030247)
Pulmonary embolism (HP:0002204)	Reduced antithrombin III activity (HP:0001976)	Abnormality of the protein C anticoagulant pathway (HP:0030780)
Portal vein thrombosis (HP:0030242)	Hepatic vein thrombosis (HP:0030243)	Mesenteric venous thrombosis (HP:0030248)
Recurrent deep vein thrombosis (HP:0004850)		

Monogenic venous thrombosis clinical tests (55524)

Level 3 Title	Haemostasis disorders (55664)
Level 4 Title	Monogenic venous thrombosis (55523)
Clinical Tests	Entries ordered left to right in table

Myeloid and marrow failure disorders (71739)

Cytopenia and pancytopenia (71752)

Cytopenia and pancytopenia phenotypes (71771)

Level 3 Title	Myeloid and marrow failure disorders (71739)
Level 4 Title	Cytopenia and pancytopenia (71752)
Phenotypes	Entries ordered left to right in table

Pancytopenia (HP:0001876)	Anemia (HP:0001903)	Congenital hypoplastic anemia (HP:0004810)
Aplastic anemia (HP:0001915)	Megaloblastic anemia (HP:0001889)	Reticulocytopenia (HP:0001896)
Neutropenia (HP:0001875)	Lymphopenia (HP:0001888)	Abnormality of cells of the granulocytic lineage (HP:0012135)
Bilineage myelodysplasia (HP:0012149)	Thrombocytopenia (HP:0001873)	Increased serum ferritin (HP:0003281)
Failure to thrive (HP:0001508)	Exocrine pancreatic insufficiency (HP:0001738)	Cafe-au-lait spot (HP:0000957)
Hepatomegaly (HP:0002240)	Splenomegaly (HP:0001744)	Recurrent infections (HP:0002719)
Skeletal dysplasia (HP:0002652)	Aplasia/Hypoplasia of the thumb (HP:0009601)	Microcephaly (HP:0000252)
Micrognathia (HP:0000347)	Cleft palate (HP:0000175)	Global developmental delay (HP:0001263)

Cytopenia and pancytopenia clinical tests (71755)

Level 3 Title	Myeloid and marrow failure disorders (71739)
Level 4 Title	Cytopenia and pancytopenia (71752)
Clinical Tests	Entries ordered left to right in table

Hearing and ear disorders (10980)

Non-syndromic hearing loss (10981)

Congenital hearing impairment (11076)

Autosomal dominant deafness phenotypes (28663)

Level 3 Title	Non-syndromic hearing loss (10981)
Level 4 Title	Congenital hearing impairment (11076)
Phenotypes	Entries ordered left to right in table

Bilateral sensorineural hearing impairment (HP:0008619)	Mid-frequency hearing loss (HP:0012781)	Progressive hearing impairment (HP:0001730)
High-frequency hearing impairment (HP:0005101)	Mild hearing impairment (HP:0012712)	Moderate hearing impairment (HP:0012713)
Aplasia of the vestibular nerve. (HP:0011393)	Unilateral deafness (HP:0009900)	Vestibular hypofunction (HP:0001756)
Morphological abnormality of the semicircular canal (HP:0011380)	Abnormality of the middle ear ossicles (HP:0004452)	Low-frequency hearing loss (HP:0008542)
Adult onset sensorineural hearing impairment (HP:0008615)	Childhood onset sensorineural hearing impairment (HP:0011474)	Severe sensorineural hearing impairment (HP:0008625)
Vertigo (HP:0002321)	Congenital sensorineural hearing impairment (HP:0008527)	Sensorineural hearing impairment (HP:0000407)
Enlarged vestibular aqueduct (HP:0011387)	Abnormality of the cochlear nerve (HP:0011396)	Hypoplasia of the vestibular nerve (HP:0011394)
Morphological abnormality of the vestibule of the inner ear (HP:0011376)	Abnormality of cochlea (HP:0000375)	Profound sensorineural hearing impairment (HP:0011476)

Level 3 Title	Non-syndromic hearing loss (10981)
Level 4 Title	Congenital hearing impairment (11076)
Clinical Tests	NB. Clinical test guidance: General Imaging Diagnostics refers to imaging of the inner ear (MRI/CT head) General Non-Imaging Diagnostics refers to Pure tone audiometry Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Auditory Neuropathy Spectrum Disorder (30607)

Auditory Neuropathy Spectrum Disorder phenotypes (30606)

Level 3 Title	Non-syndromic hearing loss (10981)
Level 4 Title	Auditory Neuropathy Spectrum Disorder (30607)
Phenotypes	Entries ordered left to right in table

Abnormality of the vestibulocochlear nerve (HP:0009591)	Sensorineural hearing impairment (HP:0000407)	High-frequency hearing impairment (HP:0005101)
Mid-frequency hearing loss (HP:0012781)	Low-frequency hearing loss (HP:0008542)	Conductive hearing impairment (HP:0000405)
Abnormality of the auditory canal (HP:0000372)	Abnormality of the internal auditory canal (HP:0011384)	Abnormality of the middle ear (HP:0000370)
Abnormality of the middle ear ossicles (HP:0004452)	Aplasia/Hypoplasia of the middle ear (HP:0008773)	Morphological abnormality of the vestibule of the inner ear (HP:0011376)
Abnormality of cochlea (HP:0000375)	Abnormality of the cochlear nerve (HP:0011396)	Hypoplasia of the vestibular nerve (HP:0011394)
Aplasia of the vestibular nerve. (HP:0011393)	Morphological abnormality of the semicircular canal (HP:0011380)	Persistent stapedial artery (HP:0011475)
Vestibular dysfunction (HP:0001751)	Abnormal speech discrimination (HP:0001963)	Tinnitus (HP:0000360)
Hyperacusis (HP:0010780)	Otitis media (HP:0000388)	Cholesteatoma (HP:0009797)
Neoplasm of the middle ear (HP:0100799)	Abnormality of the outer ear (HP:0000356)	Abnormal location of ears (HP:0000357)
Abnormality of periauricular region (HP:0000383)	Abnormality of earlobe (HP:0000363)	Abnormality of the antihelix (HP:0009738)
Abnormality of the helix (HP:0011039)	Crumpled ear (HP:0009901)	Cryptotia (HP:0011252)
Cupped ear (HP:0000378)	Macrotia (HP:0000400)	Microtia (HP:0008551)
Anotia (HP:0009892)	Polyotia (HP:0100687)	Protruding ear (HP:0000411)
Calcification of the auricular cartilage (HP:0005103)	Cystic lesions of the pinnae (HP:0010723)	Telangiectasia of the ear (HP:0009893)
Asymmetry of the ears (HP:0010722)		

Level 3 Title	Non-syndromic hearing loss (10981)
Level 4 Title	Auditory Neuropathy Spectrum Disorder (30607)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI of the internal auditory apparatus and/or medical photography of the external ear. Entries ordered left to right in table

Auditory Brainstem Response (32992.1)	Otoacoustic Emissions (32993.1)	General Imaging Diagnostics (33633.1)

Autosomal dominant deafness (36848)

Autosomal dominant deafness phenotypes (28663)

Level 3 Title	Non-syndromic hearing loss (10981)
Level 4 Title	Autosomal dominant deafness (36848)
Phenotypes	Entries ordered left to right in table

Bilateral sensorineural hearing impairment (HP:0008619)	Mid-frequency hearing loss (HP:0012781)	Progressive hearing impairment (HP:0001730)
High-frequency hearing impairment (HP:0005101)	Mild hearing impairment (HP:0012712)	Moderate hearing impairment (HP:0012713)
Aplasia of the vestibular nerve. (HP:0011393)	Unilateral deafness (HP:0009900)	Vestibular hypofunction (HP:0001756)
Morphological abnormality of the semicircular canal (HP:0011380)	Abnormality of the middle ear ossicles (HP:0004452)	Low-frequency hearing loss (HP:0008542)
Adult onset sensorineural hearing impairment (HP:0008615)	Childhood onset sensorineural hearing impairment (HP:0011474)	Severe sensorineural hearing impairment (HP:0008625)
Vertigo (HP:0002321)	Congenital sensorineural hearing impairment (HP:0008527)	Sensorineural hearing impairment (HP:0000407)
Enlarged vestibular aqueduct (HP:0011387)	Abnormality of the cochlear nerve (HP:0011396)	Hypoplasia of the vestibular nerve (HP:0011394)
Morphological abnormality of the vestibule of the inner ear (HP:0011376)	Abnormality of cochlea (HP:0000375)	Profound sensorineural hearing impairment (HP:0011476)

Autosomal dominant deafness clinical tests (30890)

Level 3 Title	Non-syndromic hearing loss (10981)
Level 4 Title	Autosomal dominant deafness (36848)
Clinical Tests	NB. Clinical test guidance: General Imaging Diagnostics refers to imaging of the inner ear (MRI/CT head) General Non-Imaging Diagnostics refers to Pure tone audiometry Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Deafness and congenital structural abnormalities (10982)

Bilateral microtia (11077)

Microtia phenotypes (40260)

Level 3 Title	Deafness and congenital structural abnormalities (10982)
Level 4 Title	Bilateral microtia (11077)
Phenotypes	Entries ordered left to right in table

Asymmetry of the ears (HP:0010722)	Abnormal location of ears (HP:0000357)	Abnormality of brain morphology (HP:0012443)
Epibulbar dermoid (HP:0001140)	Abnormality of the eye (HP:0000478)	Abnormality of the vertebrae (HP:0003468)
Branchial sinus (HP:0100272)	Crumpled ear (HP:0009901)	Microtia (HP:0008551)
Cryptotia (HP:0011252)	Abnormality of the genitourinary system (HP:0000119)	Anotia (HP:0009892)
Micrognathia (HP:0000347)	Cupped ear (HP:0000378)	Sensorineural hearing impairment (HP:0000407)
Abnormality of the cochlear nerve (HP:0011396)	Conductive hearing impairment (HP:0000405)	Abnormality of cochlea (HP:0000375)
Morphological abnormality of the vestibule of the inner ear (HP:0011376)	Preauricular pit (HP:0004467)	Macrotia (HP:0000400)
Facial asymmetry (HP:0000324)	Microtia, first degree (HP:0011266)	Microtia, third degree (HP:0011267)
Microtia, second degree (HP:0008569)	Abnormality of the middle ear ossicles (HP:0004452)	Morphological abnormality of the semicircular canal (HP:0011380)
Polyotia (HP:0100687)	Preauricular skin tag (HP:0000384)	Abnormality of the kidney (HP:0000077)
Abnormal heart morphology (HP:0001627)	Abnormality of the helix (HP:0011039)	Abnormality of earlobe (HP:0000363)

Microtia clinical tests (40261)

Level 3 Title	Deafness and congenital structural abnormalities (10982)
Level 4 Title	Bilateral microtia (11077)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI of the internal auditory apparatus and/or medical photography of the external ear. Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Familial hemifacial microsomia (37649)

Microtia phenotypes (40260)

Level 3 Title	Deafness and congenital structural abnormalities (10982)
Level 4 Title	Familial hemifacial microsomia (37649)
Phenotypes	Entries ordered left to right in table

Asymmetry of the ears (HP:0010722)	Abnormal location of ears (HP:0000357)	Abnormality of brain morphology (HP:0012443)
Epibulbar dermoid (HP:0001140)	Abnormality of the eye (HP:0000478)	Abnormality of the vertebrae (HP:0003468)
Branchial sinus (HP:0100272)	Crumpled ear (HP:0009901)	Microtia (HP:0008551)
Cryptotia (HP:0011252)	Abnormality of the genitourinary system (HP:0000119)	Anotia (HP:0009892)
Micrognathia (HP:0000347)	Cupped ear (HP:0000378)	Sensorineural hearing impairment (HP:0000407)
Abnormality of the cochlear nerve (HP:0011396)	Conductive hearing impairment (HP:0000405)	Abnormality of cochlea (HP:0000375)
Morphological abnormality of the vestibule of the inner ear (HP:0011376)	Preauricular pit (HP:0004467)	Macrotia (HP:0000400)
Facial asymmetry (HP:0000324)	Microtia, first degree (HP:0011266)	Microtia, third degree (HP:0011267)
Microtia, second degree (HP:0008569)	Abnormality of the middle ear ossicles (HP:0004452)	Morphological abnormality of the semicircular canal (HP:0011380)
Polyotia (HP:0100687)	Preauricular skin tag (HP:0000384)	Abnormality of the kidney (HP:0000077)
Abnormal heart morphology (HP:0001627)	Abnormality of the helix (HP:0011039)	Abnormality of earlobe (HP:0000363)

Microtia clinical tests (40261)

Level 3 Title	Deafness and congenital structural abnormalities (10982)
Level 4 Title	Familial hemifacial microsomia (37649)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI of the internal auditory apparatus and/or medical photography of the external ear. Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Ear malformations with hearing impairment (37657)

Ear malformations with hearing impairment phenotypes (37831)

Level 3 Title	Deafness and congenital structural abnormalities (10982)
Level 4 Title	Ear malformations with hearing impairment (37657)

Phenotypes

Entries ordered left to right in table

Cochlear aplasia (HP:0011375)	Hypoplasia of the cochlea (HP:0008586)	Incomplete partition of the cochlea type I (HP:0011374)
Incomplete partition of the cochlea type II (HP:0000376)	Abnormality of the internal auditory canal (HP:0011384)	Absent internal auditory canal (HP:0011385)
Dilatated internal auditory canal (HP:0004458)	Narrow internal auditory canal (HP:0011386)	Abnormality of the cochlear nerve (HP:0011396)
Abnormality of the vestibular nerve (HP:0011392)	Aplasia of the semicircular canal (HP:0011381)	Hypoplasia of the semicircular canal (HP:0011382)
Enlarged semicircular canal (HP:0011383)	Morphological abnormality of the anterior semicircular canal (HP:0040108)	Morphological abnormality of the lateral semicircular canal (HP:0040106)
Morphological abnormality of the posterior semicircular canal (HP:0040107)	Aplasia of the vestibular nerve. (HP:0011393)	Aplasia of the vestibule (HP:0011377)
Dilated vestibule of the inner ear (HP:0011379)	Hypoplasia of the vestibule of the inner ear (HP:0011378)	Enlarged vestibular aqueduct (HP:0011387)
Abnormality of the middle ear ossicles (HP:0004452)	Abnormality of the incus (HP:0011453)	Abnormality of the malleus (HP:0011454)
Absent malleus (HP:0011455)	Abnormality of the stapes (HP:0008628)	Absent stapes (HP:0011456)
Absent stapes head (HP:0200111)	Otosclerosis (HP:0000362)	Stapes ankylosis (HP:0000381)
Congenital stapes ankylosis (HP:0007943)	Aplasia of the middle ear ossicles (HP:0009910)	Fusion of middle ear ossicles (HP:0005473)
Conductive hearing impairment (HP:0000405)	Sensorineural hearing impairment (HP:0000407)	

Ear malformations with hearing impairment clinical tests (37659)

Level 3 Title	Deafness and congenital structural abnormalities (10982)
Level 4 Title	Ear malformations with hearing impairment (37657)
Clinical Tests	Entries ordered left to right in table

Other hearing and ear disorders (71738)

Familial Meniere Disease (71748)

Familial Meniere Disease phenotypes (71770)

Level 3 Title	Other hearing and ear disorders (71738)
Level 4 Title	Familial Meniere Disease (71748)
Phenotypes	Entries ordered left to right in table

Vertigo (HP:0002321)	Tinnitus (HP:0000360)	Hearing impairment (HP:0000365)
Nausea (HP:0002018)	Vestibular dysfunction (HP:0001751)	Paroxysmal vertigo (HP:0010532)
Headache (HP:0002315)	Migraine (HP:0002076)	Low-frequency hearing loss (HP:0008542)
Mid-frequency hearing loss (HP:0012781)	Transient hearing impairment (HP:0012779)	Progressive hearing impairment (HP:0001730)
Unilateral deafness (HP:0009900)		

Familial Meniere Disease clinical tests (71751)

Level 3 Title	Other hearing and ear disorders (71738)
Level 4 Title	Familial Meniere Disease (71748)
Clinical Tests	Entries ordered left to right in table

Infectious diseases (42209)

Bacterial disorders (42210)

Disseminated non-tuberculous mycobacterial infection (41932)

Disseminated non-tuberculous mycobacterial infection phenotypes (41933)

Level 3 Title	Bacterial disorders (42210)
Level 4 Title	Disseminated non-tuberculous mycobacterial infection (41932)
Phenotypes	Entries ordered left to right in table

Recurrent mycobacterial infections (HP:0011274)	Recurrent gram-negative bacterial infections (HP:0005420)	Recurrent staphylococcal infections (HP:0007499)
Recurrent fungal infections (HP:0002841)	Recurrent viral infections (HP:0004429)	Fever (HP:0001945)
Weight loss (HP:0001824)	Hepatomegaly (HP:0002240)	Splenomegaly (HP:0001744)

Disseminated non-tuberculous mycobacterial infection clinical tests (41934)

Level 3 Title	Bacterial disorders (42210)
Level 4 Title	Disseminated non-tuberculous mycobacterial infection (41932)
Clinical Tests	Entries ordered left to right in table

General Biopsy (33614.1)	Culture (28285.2)

Sepsis (55671)

GAinS study (55665)

GAinS study phenotypes (68124)

Level 3 Title	Sepsis (55671)
Level 4 Title	GAinS study (55665)
Phenotypes	Entries ordered left to right in table

Pneumonia (HP:0002090)	Sepsis (HP:0100806)	Hypotension (HP:0002615)
Respiratory distress (HP:0002098)	Acute kidney injury (HP:0001919)	

GAinS study clinical tests (55667)

Level 3 Title	Sepsis (55671)
Level 4 Title	GAinS study (55665)
Clinical Tests	Entries ordered left to right in table

Full Blood Count (30318.2)	Culture (28285.2)	General Imaging Diagnostics (33633.1)

Metabolic disorders (10983)

Specific metabolic abnormalities (10984)

Ketotic hypoglycaemia (11080)

Metabolic phenotypes (29171)

Level 3 Title	Specific metabolic abnormalities (10984)
Level 4 Title	Ketotic hypoglycaemia (11080)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Growth delay (HP:0001510)	Cataract (HP:0000518)	Corneal opacity (HP:0007957)
Cherry red spot of the macula (HP:0010729)	Abnormality of the heart valves (HP:0001654)	Cardiomyopathy (HP:0001638)
Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)	Hepatomegaly (HP:0002240)
Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)	Hepatic steatosis (HP:0001397)
Episodic vomiting (HP:0002572)	Abnormality of the renal tubule (HP:0000091)	Vacuolated lymphocytes (HP:0001922)
Seizures (HP:0001250)	Ataxia (HP:0001251)	Spasticity (HP:0001257)
Abnormality of extrapyramidal motor function (HP:0002071)	Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)
Abnormality of the basal ganglia (HP:0002134)	Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Hydrops fetalis (HP:0001789)
Ichthyosis (HP:0008064)	Cutaneous photosensitivity (HP:0000992)	Angiokeratoma corporis diffusum (HP:0001071)
Woolly hair (HP:0002224)		

Metabolic clinical tests (30909)

Level 3 Title	Specific metabolic abnormalities (10984)
Level 4 Title	Ketotic hypoglycaemia (11080)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain, skeletal survey and abdominal imaging and medical photographs of the face as appropriate Biopsy refers to muscle, skin and/or liver biopsy as relevant Please enter pertinent other test results including those in decompensation episodes Entries ordered left to right in table

CSF tests (30352.2)	Arterial blood gas (30304.2)	Bone profile (30317.2)
Renal biochemistry (30355.2)	Liver biochemistry (30328.2)	Full Blood Count (30318.2)
APGAR score (30207.1)	Facial features most in keeping with an OMIM disease (29826.2)	Urine dip - sulphites (31318.2)
Glucose (33153.1)	Extended renal biochemistry (33152.1)	Inflammatory markers (33156.1)
Metabolic biochemistry (33160.2)	Biotinidase (33142.1)	Insulin and C-peptide (33157.1)
Clotting (33144.1)	General Non-imaging Diagnostics (34838.1)	Urine reducing substances (33191.2)
Urine metabolic tests (33189.1)		

Lactic acidosis (11081)

Metabolic phenotypes (29171)

Level 3 Title	Specific metabolic abnormalities (10984)
Level 4 Title	Lactic acidosis (11081)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Growth delay (HP:0001510)	Cataract (HP:0000518)	Corneal opacity (HP:0007957)
Cherry red spot of the macula (HP:0010729)	Abnormality of the heart valves (HP:0001654)	Cardiomyopathy (HP:0001638)
Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)	Hepatomegaly (HP:0002240)
Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)	Hepatic steatosis (HP:0001397)
Episodic vomiting (HP:0002572)	Abnormality of the renal tubule (HP:0000091)	Vacuolated lymphocytes (HP:0001922)
Seizures (HP:0001250)	Ataxia (HP:0001251)	Spasticity (HP:0001257)
Abnormality of extrapyramidal motor function (HP:0002071)	Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)
Abnormality of the basal ganglia (HP:0002134)	Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Hydrops fetalis (HP:0001789)
Ichthyosis (HP:0008064)	Cutaneous photosensitivity (HP:0000992)	Angiokeratoma corporis diffusum (HP:0001071)
Woolly hair (HP:0002224)		

Metabolic clinical tests (30909)

<table>
<tr><td>Level 3 Title</td><td>Specific metabolic abnormalities (10984)</td></tr>
<tr><td>Level 4 Title</td><td>Lactic acidosis (11081)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to MRI brain, skeletal survey and abdominal imaging and medical photographs of the face as appropriate
Biopsy refers to muscle, skin and/or liver biopsy as relevant
Please enter pertinent other test results including those in decompensation episodes
Entries ordered left to right in table

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<tr><td>Renal biochemistry (30355.2)</td><td>Liver biochemistry (30328.2)</td><td>Full Blood Count (30318.2)</td></tr>
<tr><td>APGAR score (30207.1)</td><td>Facial features most in keeping with an OMIM disease (29826.2)</td><td>Urine dip - sulphites (31318.2)</td></tr>
<tr><td>Glucose (33153.1)</td><td>Extended renal biochemistry (33152.1)</td><td>Inflammatory markers (33156.1)</td></tr>
<tr><td>Metabolic biochemistry (33160.2)</td><td>Biotinidase (33142.1)</td><td>Insulin and C-peptide (33157.1)</td></tr>
<tr><td>Clotting (33144.1)</td><td>General Non-imaging Diagnostics (34838.1)</td><td>Urine reducing substances (33191.2)</td></tr>
<tr><td>Urine metabolic tests (33189.1)</td><td></td><td></td></tr>
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Cerebral folate deficiency (11083)

Metabolic phenotypes (29171)

Level 3 Title	Specific metabolic abnormalities (10984)
Level 4 Title	Cerebral folate deficiency (11083)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Growth delay (HP:0001510)	Cataract (HP:0000518)	Corneal opacity (HP:0007957)
Cherry red spot of the macula (HP:0010729)	Abnormality of the heart valves (HP:0001654)	Cardiomyopathy (HP:0001638)
Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)	Hepatomegaly (HP:0002240)
Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)	Hepatic steatosis (HP:0001397)
Episodic vomiting (HP:0002572)	Abnormality of the renal tubule (HP:0000091)	Vacuolated lymphocytes (HP:0001922)
Seizures (HP:0001250)	Ataxia (HP:0001251)	Spasticity (HP:0001257)
Abnormality of extrapyramidal motor function (HP:0002071)	Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)
Abnormality of the basal ganglia (HP:0002134)	Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Hydrops fetalis (HP:0001789)
Ichthyosis (HP:0008064)	Cutaneous photosensitivity (HP:0000992)	Angiokeratoma corporis diffusum (HP:0001071)
Woolly hair (HP:0002224)		

Metabolic clinical tests (30909)

Level 3 Title	Specific metabolic abnormalities (10984)
Level 4 Title	Cerebral folate deficiency (11083)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain, skeletal survey and abdominal imaging and medical photographs of the face as appropriate Biopsy refers to muscle, skin and/or liver biopsy as relevant Please enter pertinent other test results including those in decompensation episodes Entries ordered left to right in table

CSF tests (30352.2)	Arterial blood gas (30304.2)	Bone profile (30317.2)
Renal biochemistry (30355.2)	Liver biochemistry (30328.2)	Full Blood Count (30318.2)
APGAR score (30207.1)	Facial features most in keeping with an OMIM disease (29826.2)	Urine dip - sulphites (31318.2)
Glucose (33153.1)	Extended renal biochemistry (33152.1)	Inflammatory markers (33156.1)
Metabolic biochemistry (33160.2)	Biotinidase (33142.1)	Insulin and C-peptide (33157.1)
Clotting (33144.1)	General Non-imaging Diagnostics (34838.1)	Urine reducing substances (33191.2)
Urine metabolic tests (33189.1)		

Undiagnosed metabolic disorders (37620)

Undiagnosed metabolic disorders phenotypes (37748)

Level 3 Title	Specific metabolic abnormalities (10984)
Level 4 Title	Undiagnosed metabolic disorders (37620)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Delayed gross motor development (HP:0002194)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Failure to thrive (HP:0001508)	Cataract (HP:0000518)	Abnormality of the cornea (HP:0000481)
Rod-cone dystrophy (HP:0000510)	Cherry red spot of the macula (HP:0010729)	Sensorineural hearing impairment (HP:0000407)
Cardiomyopathy (HP:0001638)	Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)
Hepatomegaly (HP:0002240)	Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)
Elevated hepatic transaminases (HP:0002910)	Increased hepatic glycogen content (HP:0006568)	Reye syndrome-like episodes (HP:0006582)
Vacuolated lymphocytes (HP:0001922)	Seizures (HP:0001250)	Chorea (HP:0002072)
Ataxia (HP:0001251)	Spasticity (HP:0001257)	Dystonia (HP:0001332)
Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)	Abnormality of the basal ganglia (HP:0002134)
Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Polyhydramnios (HP:0001561)	Hydrops fetalis (HP:0001789)

Congenital disorders of glycosylation (37628)

Undiagnosed metabolic disorders phenotypes (37748)

Level 3 Title	Specific metabolic abnormalities (10984)
Level 4 Title	Congenital disorders of glycosylation (37628)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Delayed gross motor development (HP:0002194)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Failure to thrive (HP:0001508)	Cataract (HP:0000518)	Abnormality of the cornea (HP:0000481)
Rod-cone dystrophy (HP:0000510)	Cherry red spot of the macula (HP:0010729)	Sensorineural hearing impairment (HP:0000407)
Cardiomyopathy (HP:0001638)	Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)
Hepatomegaly (HP:0002240)	Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)
Elevated hepatic transaminases (HP:0002910)	Increased hepatic glycogen content (HP:0006568)	Reye syndrome-like episodes (HP:0006582)
Vacuolated lymphocytes (HP:0001922)	Seizures (HP:0001250)	Chorea (HP:0002072)
Ataxia (HP:0001251)	Spasticity (HP:0001257)	Dystonia (HP:0001332)
Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)	Abnormality of the basal ganglia (HP:0002134)
Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Polyhydramnios (HP:0001561)	Hydrops fetalis (HP:0001789)

Urea Cycle disorders (15108)

Hyperammonaemia (11079)

Metabolic phenotypes (29171)

Level 3 Title	Urea Cycle disorders (15108)
Level 4 Title	Hyperammonaemia (11079)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Growth delay (HP:0001510)	Cataract (HP:0000518)	Corneal opacity (HP:0007957)
Cherry red spot of the macula (HP:0010729)	Abnormality of the heart valves (HP:0001654)	Cardiomyopathy (HP:0001638)
Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)	Hepatomegaly (HP:0002240)
Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)	Hepatic steatosis (HP:0001397)
Episodic vomiting (HP:0002572)	Abnormality of the renal tubule (HP:0000091)	Vacuolated lymphocytes (HP:0001922)
Seizures (HP:0001250)	Ataxia (HP:0001251)	Spasticity (HP:0001257)
Abnormality of extrapyramidal motor function (HP:0002071)	Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)
Abnormality of the basal ganglia (HP:0002134)	Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Hydrops fetalis (HP:0001789)
Ichthyosis (HP:0008064)	Cutaneous photosensitivity (HP:0000992)	Angiokeratoma corporis diffusum (HP:0001071)
Woolly hair (HP:0002224)		

Metabolic clinical tests (30909)

<table>
<tr><td>Level 3 Title</td><td>Urea Cycle disorders (15108)</td></tr>
<tr><td>Level 4 Title</td><td>Hyperammonaemia (11079)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to MRI brain, skeletal survey and abdominal imaging and medical photographs of the face as appropriate
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<tr><td>Clotting (33144.1)</td><td>General Non-imaging Diagnostics (34838.1)</td><td>Urine reducing substances (33191.2)</td></tr>
<tr><td>Urine metabolic tests (33189.1)</td><td></td><td></td></tr>
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Lysosomal storage disorders (10985)

Mucopolysaccharideosis, Gaucher, Fabry (11084)

Metabolic phenotypes (29171)

Level 3 Title	Lysosomal storage disorders (10985)
Level 4 Title	Mucopolysaccharideosis, Gaucher, Fabry (11084)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Growth delay (HP:0001510)	Cataract (HP:0000518)	Corneal opacity (HP:0007957)
Cherry red spot of the macula (HP:0010729)	Abnormality of the heart valves (HP:0001654)	Cardiomyopathy (HP:0001638)
Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)	Hepatomegaly (HP:0002240)
Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)	Hepatic steatosis (HP:0001397)
Episodic vomiting (HP:0002572)	Abnormality of the renal tubule (HP:0000091)	Vacuolated lymphocytes (HP:0001922)
Seizures (HP:0001250)	Ataxia (HP:0001251)	Spasticity (HP:0001257)
Abnormality of extrapyramidal motor function (HP:0002071)	Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)
Abnormality of the basal ganglia (HP:0002134)	Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Hydrops fetalis (HP:0001789)
Ichthyosis (HP:0008064)	Cutaneous photosensitivity (HP:0000992)	Angiokeratoma corporis diffusum (HP:0001071)
Woolly hair (HP:0002224)		

Metabolic clinical tests (30909)

<table>
<tr><td>Level 3 Title</td><td>Lysosomal storage disorders (10985)</td></tr>
<tr><td>Level 4 Title</td><td>Mucopolysaccharideosis, Gaucher, Fabry (11084)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to MRI brain, skeletal survey and abdominal imaging and medical photographs of the face as appropriate Biopsy refers to muscle, skin and/or liver biopsy as relevant
Please enter pertinent other test results including those in decompensation episodes
Entries ordered left to right in table

<table>
<tr><td>CSF tests (30352.2)</td><td>Arterial blood gas (30304.2)</td><td>Bone profile (30317.2)</td></tr>
<tr><td>Renal biochemistry (30355.2)</td><td>Liver biochemistry (30328.2)</td><td>Full Blood Count (30318.2)</td></tr>
<tr><td>APGAR score (30207.1)</td><td>Facial features most in keeping with an OMIM disease (29826.2)</td><td>Urine dip - sulphites (31318.2)</td></tr>
<tr><td>Glucose (33153.1)</td><td>Extended renal biochemistry (33152.1)</td><td>Inflammatory markers (33156.1)</td></tr>
<tr><td>Metabolic biochemistry (33160.2)</td><td>Biotinidase (33142.1)</td><td>Insulin and C-peptide (33157.1)</td></tr>
<tr><td>Clotting (33144.1)</td><td>General Non-imaging Diagnostics (34838.1)</td><td>Urine reducing substances (33191.2)</td></tr>
<tr><td>Urine metabolic tests (33189.1)</td><td></td><td></td></tr>
</table>
</td></tr>
</table>

Mitochondrial (10986)

Mitochondrial disorders (11085)

Mitochondrial phenotypes (29172)

Level 3 Title	Mitochondrial (10986)
Level 4 Title	Mitochondrial disorders (11085)
Phenotypes	Entries ordered left to right in table

Lactic acidosis (HP:0003128)	Increased CSF lactate (HP:0002490)	Abnormal mitochondrial morphology (HP:0008322)
Depletion of mitochondrial DNA in muscle tissue (HP:0009141)	Multiple mitochondrial DNA deletions (HP:0003689)	Decreased activity of mitochondrial complex I (HP:0011923)
Decreased activity of mitochondrial complex II (HP:0008314)	Decreased activity of mitochondrial complex III (HP:0011924)	Decreased activity of mitochondrial complex IV (HP:0008347)
Decreased activity of the pyruvate dehydrogenase complex (HP:0002928)	Intellectual disability (HP:0001249)	Delayed gross motor development (HP:0002194)
Developmental regression (HP:0002376)	Failure to thrive (HP:0001508)	Progressive external ophthalmoplegia (HP:0000590)
Ptosis (HP:0000508)	Rod-cone dystrophy (HP:0000510)	Optic atrophy (HP:0000648)
Sensorineural hearing impairment (HP:0000407)	Cardiomyopathy (HP:0001638)	Hepatic failure (HP:0001399)
Exocrine pancreatic insufficiency (HP:0001738)	Proximal tubulopathy (HP:0000114)	Generalized hypotonia (HP:0001290)
Myopathy (HP:0003198)	Ragged-red muscle fibers (HP:0003200)	Sensory neuropathy (HP:0000763)
Motor polyneuropathy (HP:0007178)	Dementia (HP:0000726)	Encephalopathy (HP:0001298)
Seizures (HP:0001250)	Ataxia (HP:0001251)	Dystonia (HP:0001332)
Abnormality of extrapyramidal motor function (HP:0002071)	Stroke-like episodes (HP:0002401)	Abnormality of the basal ganglia (HP:0002134)
Leukodystrophy (HP:0002415)	Abnormality of the internal capsule (HP:0012502)	Cerebellar atrophy (HP:0001272)
Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Focal white matter lesions (HP:0007042)	Sideroblastic anemia (HP:0001924)
Multiple lipomas (HP:0001012)		

Mitochondrial clinical tests (30910)

Level 3 Title	Mitochondrial (10986)
Level 4 Title	Mitochondrial disorders (11085)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain, skeletal survey and abdominal imaging and medical photographs of the face as appropriate Biopsy refers to muscle, skin and/or liver biopsy as relevant Please enter pertinent other test results including those in decompensation episodes Entries ordered left to right in table

Renal biochemistry (30355.2)	Arterial blood gas (30304.2)	Bone profile (30317.2)
Liver biochemistry (30328.2)	CSF tests (30352.2)	APGAR score (30207.1)
Full Blood Count (30318.2)	Facial features most in keeping with an OMIM disease (29826.2)	Urine dip - sulphites (31318.2)
Glucose (33153.1)	Extended renal biochemistry (33152.1)	Biotinidase (33142.1)
Metabolic biochemistry (33160.2)	Inflammatory markers (33156.1)	Insulin and C-peptide (33157.1)
Clotting (33144.1)	General Biopsy (33614.1)	General Non-imaging Diagnostics (34838.1)
General Imaging Diagnostics (33633.1)	Urine reducing substances (33191.2)	Urine metabolic tests (33189.1)

Peroxisomal disorders (10987)

Peroxisomal biogenesis disorders (11086)

Metabolic phenotypes (29171)

Level 3 Title	Peroxisomal disorders (10987)
Level 4 Title	Peroxisomal biogenesis disorders (11086)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Growth delay (HP:0001510)	Cataract (HP:0000518)	Corneal opacity (HP:0007957)
Cherry red spot of the macula (HP:0010729)	Abnormality of the heart valves (HP:0001654)	Cardiomyopathy (HP:0001638)
Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)	Hepatomegaly (HP:0002240)
Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)	Hepatic steatosis (HP:0001397)
Episodic vomiting (HP:0002572)	Abnormality of the renal tubule (HP:0000091)	Vacuolated lymphocytes (HP:0001922)
Seizures (HP:0001250)	Ataxia (HP:0001251)	Spasticity (HP:0001257)
Abnormality of extrapyramidal motor function (HP:0002071)	Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)
Abnormality of the basal ganglia (HP:0002134)	Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Hydrops fetalis (HP:0001789)
Ichthyosis (HP:0008064)	Cutaneous photosensitivity (HP:0000992)	Angiokeratoma corporis diffusum (HP:0001071)
Woolly hair (HP:0002224)		

Metabolic clinical tests (30909)

Level 3 Title	Peroxisomal disorders (10987)
Level 4 Title	Peroxisomal biogenesis disorders (11086)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain, skeletal survey and abdominal imaging and medical photographs of the face as appropriate Biopsy refers to muscle, skin and/or liver biopsy as relevant Please enter pertinent other test results including those in decompensation episodes Entries ordered left to right in table

CSF tests (30352.2)	Arterial blood gas (30304.2)	Bone profile (30317.2)
Renal biochemistry (30355.2)	Liver biochemistry (30328.2)	Full Blood Count (30318.2)
APGAR score (30207.1)	Facial features most in keeping with an OMIM disease (29826.2)	Urine dip - sulphites (31318.2)
Glucose (33153.1)	Extended renal biochemistry (33152.1)	Inflammatory markers (33156.1)
Metabolic biochemistry (33160.2)	Biotinidase (33142.1)	Insulin and C-peptide (33157.1)
Clotting (33144.1)	General Non-imaging Diagnostics (34838.1)	Urine reducing substances (33191.2)
Urine metabolic tests (33189.1)		

Other peroxisomal disorders (15109)

Metabolic phenotypes (29171)

Level 3 Title	Peroxisomal disorders (10987)
Level 4 Title	Other peroxisomal disorders (15109)
Phenotypes	Entries ordered left to right in table

Abnormality of metabolism/homeostasis (HP:0001939)	Ketotic hypoglycemia (HP:0012734)	Low CSF 5-methyltetrahydrofolate (HP:0012446)
Hyperammonemia (HP:0001987)	Abnormality of lysosomal metabolism (HP:0004356)	Very long chain fatty acid accumulation (HP:0008167)
Abnormality of glycoprotein metabolism (HP:0004367)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)
Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)	Developmental regression (HP:0002376)
Growth delay (HP:0001510)	Cataract (HP:0000518)	Corneal opacity (HP:0007957)
Cherry red spot of the macula (HP:0010729)	Abnormality of the heart valves (HP:0001654)	Cardiomyopathy (HP:0001638)
Dysostosis multiplex (HP:0000943)	Epiphyseal stippling (HP:0010655)	Hepatomegaly (HP:0002240)
Splenomegaly (HP:0001744)	Cholestasis (HP:0001396)	Hepatic steatosis (HP:0001397)
Episodic vomiting (HP:0002572)	Abnormality of the renal tubule (HP:0000091)	Vacuolated lymphocytes (HP:0001922)
Seizures (HP:0001250)	Ataxia (HP:0001251)	Spasticity (HP:0001257)
Abnormality of extrapyramidal motor function (HP:0002071)	Generalized hypotonia (HP:0001290)	Leukodystrophy (HP:0002415)
Abnormality of the basal ganglia (HP:0002134)	Aplasia/Hypoplasia of the cerebellum (HP:0007360)	Hydrops fetalis (HP:0001789)
Ichthyosis (HP:0008064)	Cutaneous photosensitivity (HP:0000992)	Angiokeratoma corporis diffusum (HP:0001071)
Woolly hair (HP:0002224)		

Level 3 Title	Peroxisomal disorders (10987)
Level 4 Title	Other peroxisomal disorders (15109)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain, skeletal survey and abdominal imaging and medical photographs of the face as appropriate Biopsy refers to muscle, skin and/or liver biopsy as relevant Please enter pertinent other test results including those in decompensation episodes Entries ordered left to right in table

CSF tests (30352.2)	Arterial blood gas (30304.2)	Bone profile (30317.2)
Renal biochemistry (30355.2)	Liver biochemistry (30328.2)	Full Blood Count (30318.2)
APGAR score (30207.1)	Facial features most in keeping with an OMIM disease (29826.2)	Urine dip - sulphites (31318.2)
Glucose (33153.1)	Extended renal biochemistry (33152.1)	Inflammatory markers (33156.1)
Metabolic biochemistry (33160.2)	Biotinidase (33142.1)	Insulin and C-peptide (33157.1)
Clotting (33144.1)	General Non-imaging Diagnostics (34838.1)	Urine reducing substances (33191.2)
Urine metabolic tests (33189.1)		

Neurology and neurodevelopmental disorders (10988)

Motor Disorders of the CNS (10989)

Cerebellar hypoplasia (36512)

Cerebellar hypoplasia phenotypes (36646)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Cerebellar hypoplasia (36512)
Phenotypes	Entries ordered left to right in table

Delayed gross motor development (HP:0002194)	Delayed fine motor development (HP:0010862)	Delayed speech and language development (HP:0000750)
Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)	Microcephaly (HP:0000252)
Macrocephaly (HP:0000256)	Generalized hypotonia (HP:0001290)	Ataxia (HP:0001251)
Cerebellar atrophy (HP:0001272)	Dandy-Walker malformation (HP:0001305)	Cerebellar hypoplasia (HP:0001321)
Cerebellar agenesis (HP:0012642)	Cerebellar vermis hypoplasia (HP:0001320)	Cerebellar hemisphere hypoplasia (HP:0100307)
Cerebellar dysplasia (HP:0007033)	Olivopontocerebellar hypoplasia (HP:0006955)	Molar tooth sign on MRI (HP:0002419)
Arnold-Chiari type I malformation (HP:0007099)	Cerebellar cyst (HP:0002350)	Small posterior fossa (HP:0040010)
Encephalocele (HP:0002084)	Enlarged cisterna magna (HP:0002280)	Hypoplasia of the pons (HP:0012110)
Abnormality of brainstem morphology (HP:0002363)	Abnormality of the cerebral cortex (HP:0002538)	Atrophy/Degeneration affecting the brainstem (HP:0007366)
Abnormality of the cerebral white matter (HP:0002500)	Abnormality of the basal ganglia (HP:0002134)	Ventriculomegaly (HP:0002119)
Arachnoid cyst (HP:0100702)	Abnormality of the corpus callosum (HP:0001273)	Abnormality of thalamus morphology (HP:0010663)
Seizures (HP:0001250)	Polymicrogyria (HP:0002126)	Nystagmus (HP:0000639)

	Peripheral neuropathy (HP:0009830)	Sensory neuropathy (HP:0000763)	Sensorimotor neuropathy (HP:0007141)
	Cerebral palsy (HP:0100021)	Oculomotor apraxia (HP:0000657)	Strabismus (HP:0000486)
	Retinal dystrophy (HP:0000556)	Sensorineural hearing impairment (HP:0000407)	Abnormal facial shape (HP:0001999)
	Abnormal renal morphology (HP:0012210)	Elevated hepatic transaminases (HP:0002910)	Abnormality of cardiovascular system morphology (HP:0030680)
	Arrhythmia (HP:0011675)		

Cerebellar hypoplasia clinical tests (36514)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Cerebellar hypoplasia (36512)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to medical photographs and MRI brain Entries ordered left to right in table General Imaging Diagnostics (33633.1)

Hereditary ataxia (11087)

Ataxia phenotypes (28525)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Hereditary ataxia (11087)
Phenotypes	Entries ordered left to right in table

Ataxia (HP:0001251)	Abnormality of eye movement (HP:0000496)	Ophthalmoparesis (HP:0000597)
Abnormality of saccadic eye movements (HP:0000570)	Dysmetric saccades (HP:0000641)	Slow saccadic eye movements (HP:0000514)
Nystagmus (HP:0000639)	Gaze-evoked nystagmus (HP:0000640)	Hyperreflexia (HP:0001347)
Areflexia (HP:0001284)	Babinski sign (HP:0003487)	Abnormal pyramidal signs (HP:0007256)
Spasticity (HP:0001257)	Muscle weakness (HP:0001324)	Proximal muscle weakness in upper limbs (HP:0008997)
Proximal muscle weakness in lower limbs (HP:0008994)	Distal upper limb muscle weakness (HP:0008959)	Distal lower limb muscle weakness (HP:0009053)
Skeletal muscle atrophy (HP:0003202)	Proximal upper limb amyotrophy (HP:0008948)	Proximal lower limb amyotrophy (HP:0008956)
Distal upper limb amyotrophy (HP:0007149)	Distal lower limb amyotrophy (HP:0008944)	Abnormality of the basal ganglia (HP:0002134)
Dystonia (HP:0001332)	Chorea (HP:0002072)	Rigidity (HP:0002063)
Resting tremor (HP:0002322)	Visual impairment (HP:0000505)	Diplopia (HP:0000651)
Dysphagia (HP:0002015)	Functional abnormality of the bladder (HP:0000009)	Cognitive impairment (HP:0100543)

Ataxia clinical tests (30912)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Hereditary ataxia (11087)
Clinical Tests	Entries ordered left to right in table

Ataxia assessment (30228.2)	General Imaging Diagnostics (33633.1)

Early onset dystonia (11088)

Early onset dystonia phenotypes (29174)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Early onset dystonia (11088)
Phenotypes	Entries ordered left to right in table

Dystonia (HP:0001332)	Blepharospasm (HP:0000643)	Torticollis (HP:0000473)
Craniofacial dystonia (HP:0012179)	Oromandibular dystonia (HP:0012048)	Laryngeal dystonia (HP:0012049)
Axial dystonia (HP:0002530)	Limb dystonia (HP:0002451)	Writer's cramp (HP:0002356)
Action tremor (HP:0002345)	Head tremor (HP:0002346)	Myoclonus (HP:0001336)
Parkinsonism (HP:0001300)	Parkinsonism with favorable response to dopaminergic medication (HP:0002548)	Abnormality of eye movement (HP:0000496)
Optic atrophy (HP:0000648)	Dysarthria (HP:0001260)	Bulbar signs (HP:0002483)
Dysphagia (HP:0002015)	Ataxia (HP:0001251)	Spasticity (HP:0001257)
Skeletal muscle atrophy (HP:0003202)	Intellectual disability (HP:0001249)	Abnormality of higher mental function (HP:0011446)
Dementia (HP:0000726)	Psychosis (HP:0000709)	Seizures (HP:0001250)
Sensory impairment (HP:0003474)	Peripheral neuropathy (HP:0009830)	Abnormality of the basal ganglia (HP:0002134)
Focal white matter lesions (HP:0007042)	Cerebral atrophy (HP:0002059)	Cerebellar atrophy (HP:0001272)
Abnormality of metabolism/homeostasis (HP:0001939)		

Hereditary spastic paraplegia (11089)

Hereditary spastic paraplegia phenotypes (27781)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Hereditary spastic paraplegia (11089)
Phenotypes	Entries ordered left to right in table

Abnormality of higher mental function (HP:0011446)	Cognitive impairment (HP:0100543)	Psychosis (HP:0000709)
Neurological speech impairment (HP:0002167)	Dysarthria (HP:0001260)	Dysphagia (HP:0002015)
Abnormality of the eye (HP:0000478)	Cataract (HP:0000518)	Congenital cataract (HP:0000519)
Optic atrophy (HP:0000648)	Abnormality of eye movement (HP:0000496)	Spasticity (HP:0001257)
Abnormal pyramidal signs (HP:0007256)	Hyperreflexia in upper limbs (HP:0007350)	Upper limb spasticity (HP:0006986)
Skeletal muscle atrophy (HP:0003202)	Fasciculations (HP:0002380)	Proximal upper limb amyotrophy (HP:0008948)
Proximal lower limb amyotrophy (HP:0008956)	Distal upper limb amyotrophy (HP:0007149)	Distal lower limb amyotrophy (HP:0008944)
Ataxia (HP:0001251)	Gait ataxia (HP:0002066)	Limb ataxia (HP:0002070)
Abnormality of the basal ganglia (HP:0002134)	Dystonia (HP:0001332)	Parkinsonism (HP:0001300)
Myoclonus (HP:0001336)	Sensory impairment (HP:0003474)	Impaired vibratory sensation (HP:0002495)
Impaired tactile sensation (HP:0010830)	Impaired proprioception (HP:0010831)	Impaired temperature sensation (HP:0010829)
Impaired pain sensation (HP:0007328)	Abnormality of central motor conduction (HP:0012079)	Motor axonal neuropathy (HP:0007002)
Demyelinating motor neuropathy (HP:0007220)	Distal peripheral sensory neuropathy (HP:0007067)	Sensory axonal neuropathy (HP:0003390)
Demyelinating sensory neuropathy (HP:0011402)	Mixed demyelinating and axonal polyneuropathy (HP:0007327)	Hypoplasia of the corpus callosum (HP:0002079)
Focal white matter lesions (HP:0007042)	Cerebral atrophy (HP:0002059)	Cerebellar atrophy (HP:0001272)

Hereditary spastic paraplegia clinical tests (30951)

<table>
<tr><td>Level 3 Title</td><td>Motor Disorders of the CNS (10989)</td></tr>
<tr><td>Level 4 Title</td><td>Hereditary spastic paraplegia (11089)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to MRI brain
Non-imaging diagnostics refers Motor nerve conduction studies, Sensory nerve conduction studies, Motor evoked potentials (MEP)
Entries ordered left to right in table
<table><tr><td>Spastic Paraplegia Assessment (30230.1)</td><td>General Imaging Diagnostics (33633.1)</td><td>General Non-imaging Diagnostics (34838.1)</td></tr></table></td></tr>
</table>

Neurotransmitter disorders (37779)

Neurotransmitter disorders phenotypes (37902)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Neurotransmitter disorders (37779)
Phenotypes	Entries ordered left to right in table

Abnormality of the cerebrospinal fluid (HP:0002921)	Prolactin deficiency (HP:0008202)	Prolactin excess (HP:0000870)
Dyskinesia (HP:0100660)	Dystonia (HP:0001332)	Gait disturbance (HP:0001288)
Hyperkinesis (HP:0002487)	Oculogyric crisis (HP:0010553)	Parkinsonism (HP:0001300)
Generalized hypotonia (HP:0001290)	Microcephaly (HP:0000252)	Intellectual disability (HP:0001249)
Abnormal autonomic nervous system physiology (HP:0012332)	Sleep disturbance (HP:0002360)	Feeding difficulties (HP:0011968)
Bulbar palsy (HP:0001283)		

Neurotransmitter disorders clinical tests (37780)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Neurotransmitter disorders (37779)
Clinical Tests	NB. Clinical test guidance: CSF tests refers to CSF neurotransmitters Entries ordered left to right in table

CSF tests (30352.2)	Hormones (other) (33155.1)

Structural basal ganglia disorders (37786)

Structural basal ganglia disorders phenotypes (37901)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Structural basal ganglia disorders (37786)
Phenotypes	Entries ordered left to right in table

Abnormality of the basal ganglia (HP:0002134)	Bilateral basal ganglia lesions (HP:0007146)	Symmetric lesions of the basal ganglia (HP:0007039)
Small basal ganglia (HP:0012697)	Basal ganglia calcification (HP:0002135)	Basal ganglia cysts (HP:0006799)
Basal ganglia gliosis (HP:0006999)	Basal ganglia necrosis (HP:0012128)	Cavitation of the basal ganglia (HP:0007007)
Abnormality of the globus pallidus (HP:0002453)	Abnormality of the striatum (HP:0010994)	Abnormality of the caudate nucleus (HP:0002339)
Abnormality of the substantia nigra (HP:0045007)	Iron accumulation in substantia nigra (HP:0012678)	Abnormality of movement (HP:0100022)
Dystonia (HP:0001332)	Microcephaly (HP:0000252)	Intellectual disability (HP:0001249)
Feeding difficulties (HP:0011968)	Bulbar palsy (HP:0001283)	

Structural basal ganglia disorders clinical tests (37787)

Level 3 Title	Motor Disorders of the CNS (10989)
Level 4 Title	Structural basal ganglia disorders (37786)
Clinical Tests	N.B. Clinical test guidance: General biopsy refers to respiratory chain enzyme analysis General imaging diagnostics refers to MRI brain Liver biochemistry refers to AST Other enzymes refers to LDH Full blood count refers to blood film Entries ordered left to right in table

General Biopsy (33614.1)	Electroretinogram (30291.2)

Genetic Epilepsies with Febrile Seizures Plus (11091)

Genetic Epilepsies with Febrile Seizures Plus phenotypes (29175)

Level 3 Title	Inherited Epilepsy Syndromes (10990)
Level 4 Title	Genetic Epilepsies with Febrile Seizures Plus (11091)
Phenotypes	Entries ordered left to right in table

Febrile seizures (HP:0002373)	Focal seizures (HP:0007359)	Focal seizures without impairment of consciousness or awareness (HP:0002349)
Focal seizures with impairment of consciousness or awareness (HP:0002384)	Auditory auras (HP:0011158)	Epigastric auras (HP:0011159)
Psychic auras (HP:0011162)	Vegetative auras (HP:0011164)	Gustatory auras (HP:0011160)
Olfactory auras (HP:0011161)	Somatosensory auras (HP:0011163)	Visual auras (HP:0011165)
Generalized seizures (HP:0002197)	Generalized tonic-clonic seizures (HP:0002069)	Generalized tonic seizures (HP:0010818)
Generalized clonic seizures (HP:0011169)	Generalized myoclonic seizures (HP:0002123)	Atonic seizures (HP:0010819)
Absence seizures (HP:0002121)	EEG abnormality (HP:0002353)	EEG with focal spikes (HP:0011193)
EEG with focal spike waves (HP:0011197)	EEG with focal sharp waves (HP:0011196)	EEG with focal sharp slow waves (HP:0011195)
EEG with spike-wave complexes (HP:0010850)	EEG with polyspike wave complexes (HP:0002392)	EEG with generalized spikes (HP:0012000)
EEG with frontal focal spike waves (HP:0012010)	EEG with frontal focal spikes (HP:0012015)	EEG with frontal sharp slow waves (HP:0011290)
EEG with frontal sharp waves (HP:0011294)	EEG with occipital focal spike waves (HP:0012011)	EEG with occipital focal spikes (HP:0012016)
EEG with occipital sharp slow waves (HP:0011287)	EEG with occipital sharp waves (HP:0011292)	EEG with temporal focal spike waves (HP:0012013)
EEG with temporal focal spikes (HP:0012018)	EEG with temporal sharp slow waves (HP:0011289)	EEG with temporal sharp waves (HP:0011296)

	EEG with parietal focal spike waves (HP:0012012)	EEG with parietal focal spikes (HP:0012017)	EEG with parietal sharp slow waves (HP:0011288)
	EEG with parietal sharp waves (HP:0011295)	EEG with centrotemporal focal spike waves (HP:0012557)	EEG with central focal spike waves (HP:0012009)
	EEG with central focal spikes (HP:0012014)	EEG with central sharp slow waves (HP:0011291)	EEG with central sharp waves (HP:0011293)
	EEG with multifocal slow activity (HP:0010844)	Abnormality of neuronal migration (HP:0002269)	

Genetic Epilepsies with Febrile Seizures Plus clinical tests (30918)

Level 3 Title	Inherited Epilepsy Syndromes (10990)
Level 4 Title	Genetic Epilepsies with Febrile Seizures Plus (11091)
Clinical Tests	N.B. Clinical test guidance: Imaging diagnostics refers to MRI brain Non-imaging diagnostics refers to EEG Entries ordered left to right in table

Intelligence Quotient Assessment (30224.1)	Performance Intelligence Quotient assessment (30226.1)	General Imaging Diagnostics (33633.1)
General Non-imaging Diagnostics (34838.1)		

Familial Genetic Generalised Epilepsies (11092)

Familial Genetic Generalised Epilepsies phenotypes (29176)

<table>
<tr><td>Level 3 Title</td><td>Inherited Epilepsy Syndromes (10990)</td></tr>
<tr><td>Level 4 Title</td><td>Familial Genetic Generalised Epilepsies (11092)</td></tr>
<tr><td>Phenotypes</td><td>Entries ordered left to right in table
<table>
<tr><td>Febrile seizures (HP:0002373)</td><td>Generalized tonic-clonic seizures (HP:0002069)</td><td>Generalized tonic-clonic seizures on awakening (HP:0007193)</td></tr>
<tr><td>Photosensitive tonic-clonic seizures (HP:0007207)</td><td>Generalized myoclonic seizures (HP:0002123)</td><td>Photomyoclonic seizures (HP:0001327)</td></tr>
<tr><td>Absence seizures (HP:0002121)</td><td>Typical absence seizures (HP:0011147)</td><td>Atypical absence seizures (HP:0007270)</td></tr>
<tr><td>Absence seizures with special features (HP:0011148)</td><td>Absence seizures with eyelid myoclonia (HP:0011149)</td><td>Myoclonic absences (HP:0011150)</td></tr>
<tr><td>Atonic seizures (HP:0010819)</td><td>Generalized tonic seizures (HP:0010818)</td><td>Generalized clonic seizures (HP:0011169)</td></tr>
<tr><td>EEG abnormality (HP:0002353)</td><td>EEG with spike-wave complexes (HP:0010850)</td><td>EEG with polyspike wave complexes (HP:0002392)</td></tr>
<tr><td>EEG with generalized spikes (HP:0012000)</td><td>EEG with generalized epileptiform discharges (HP:0011198)</td><td>EEG with hyperventilation-induced epileptiform discharges (HP:0010858)</td></tr>
<tr><td>EEG with hyperventilation-induced generalized epileptiform discharges (HP:0011184)</td><td>EEG with photoparoxysmal response (HP:0010852)</td><td></td></tr>
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Familial Genetic Generalised Epilepsies clinical tests (30925)

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<tr><td>Level 4 Title</td><td>Familial Genetic Generalised Epilepsies (11092)</td></tr>
<tr><td>Clinical Tests</td><td>N.B. Clinical test guidance:
Imaging diagnostics refers to MRI brain
Non-imaging diagnostics refers to EEG
Entries ordered left to right in table

<table><tr><td>Intelligence Quotient Assessment (30224.1)</td><td>Performance Intelligence Quotient assessment (30226.1)</td><td>General Non-imaging Diagnostics (34838.1)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td></td><td></td></tr></table></td></tr>
</table>

Familial Focal Epilepsies (11093)

Familial Focal Epilepsies phenotypes (29177)

Level 3 Title	Inherited Epilepsy Syndromes (10990)
Level 4 Title	Familial Focal Epilepsies (11093)
Phenotypes	Entries ordered left to right in table

Febrile seizures (HP:0002373)	Focal seizures without impairment of consciousness or awareness (HP:0002349)	Focal seizures with impairment of consciousness or awareness (HP:0002384)
Auditory auras (HP:0011158)	Epigastric auras (HP:0011159)	Psychic auras (HP:0011162)
Vegetative auras (HP:0011164)	Gustatory auras (HP:0011160)	Olfactory auras (HP:0011161)
Somatosensory auras (HP:0011163)	Visual auras (HP:0011165)	Generalized tonic-clonic seizures with focal onset (HP:0007334)
EEG abnormality (HP:0002353)	EEG with focal spike waves (HP:0011197)	EEG with focal sharp waves (HP:0011196)
EEG with focal sharp slow waves (HP:0011195)	EEG with focal spikes (HP:0011193)	EEG with frontal focal spike waves (HP:0012010)
EEG with frontal focal spikes (HP:0012015)	EEG with frontal sharp slow waves (HP:0011290)	EEG with frontal sharp waves (HP:0011294)
EEG with occipital focal spike waves (HP:0012011)	EEG with occipital focal spikes (HP:0012016)	EEG with occipital sharp slow waves (HP:0011287)
EEG with occipital sharp waves (HP:0011292)	EEG with temporal focal spike waves (HP:0012013)	EEG with temporal focal spikes (HP:0012018)
EEG with temporal sharp slow waves (HP:0011289)	EEG with temporal sharp waves (HP:0011296)	EEG with parietal focal spike waves (HP:0012012)
EEG with parietal focal spikes (HP:0012017)	EEG with parietal sharp slow waves (HP:0011288)	EEG with parietal sharp waves (HP:0011295)
EEG with centrotemporal focal spike waves (HP:0012557)	EEG with central focal spike waves (HP:0012009)	EEG with central focal spikes (HP:0012014)
EEG with central sharp slow waves (HP:0011291)	EEG with central sharp waves (HP:0011293)	EEG with multifocal slow activity (HP:0010844)

Familial Focal Epilepsies clinical tests (30927)

<table>
<tr><td>Level 3 Title</td><td>Inherited Epilepsy Syndromes (10990)</td></tr>
<tr><td>Level 4 Title</td><td>Familial Focal Epilepsies (11093)</td></tr>
<tr><td>Clinical Tests</td><td>N.B. Clinical test guidance:
Imaging diagnostics refers to MRI brain
Non-imaging diagnostics refers to EEG
Entries ordered left to right in table
<table><tr><td>Performance Intelligence Quotient assessment (30226.1)</td><td>Intelligence Quotient Assessment (30224.1)</td><td>General Imaging Diagnostics (33633.1)</td></tr><tr><td>General Non-imaging Diagnostics (34838.1)</td><td></td><td></td></tr></table></td></tr>
</table>

Epileptic encephalopathy (11094)

Epileptic encephalopathy phenotypes (29178)

Level 3 Title	Inherited Epilepsy Syndromes (10990)
Level 4 Title	Epileptic encephalopathy (11094)
Phenotypes	Entries ordered left to right in table

Seizures (HP:0001250)	Generalized seizures (HP:0002197)	Focal seizures (HP:0007359)
Epileptic spasms (HP:0011097)	Infantile spasms (HP:0012469)	Myoclonic spasms (HP:0003739)
Absence seizures (HP:0002121)	Febrile seizures (HP:0002373)	EEG abnormality (HP:0002353)
Hypsarrhythmia (HP:0002521)	EEG with burst suppression (HP:0010851)	EEG with generalized epileptiform discharges (HP:0011198)
EEG with focal epileptiform discharges (HP:0011185)	Multifocal epileptiform discharges (HP:0010841)	Polymorphic focal epileptiform discharges (HP:0011192)
EEG with spike-wave complexes (2.5-3.5 Hz) (HP:0010848)	Infantile encephalopathy (HP:0007105)	Intellectual disability, mild (HP:0001256)
Intellectual disability, moderate (HP:0002342)	Intellectual disability, severe (HP:0010864)	Intellectual disability, profound (HP:0002187)
Abnormality of movement (HP:0100022)	Muscular hypotonia of the trunk (HP:0008936)	Generalized hypotonia (HP:0001290)
Limb hypertonia (HP:0002509)		

Epileptic encephalopathy clinical tests (30932)

Level 3 Title	Inherited Epilepsy Syndromes (10990)
Level 4 Title	Epileptic encephalopathy (11094)
Clinical Tests	N.B. Clinical test guidance: Imaging diagnostics refers to MRI brain Non-imaging diagnostics refers to EEG Entries ordered left to right in table

General Non-imaging Diagnostics (34838.1)	General Imaging Diagnostics (33633.1)

Epilepsy plus other features (41924)

Epilepsy plus other features phenotypes (42117)

Level 3 Title	Inherited Epilepsy Syndromes (10990)
Level 4 Title	Epilepsy plus other features (41924)
Phenotypes	Entries ordered left to right in table

Seizures (HP:0001250)	Focal seizures with impairment of consciousness or awareness (HP:0002384)	Focal seizures without impairment of consciousness or awareness (HP:0002349)
Generalized tonic-clonic seizures (HP:0002069)	Atypical absence seizures (HP:0007270)	Generalized myoclonic seizures (HP:0002123)
Segmental myoclonic seizures (HP:0025191)	Generalized tonic seizures (HP:0010818)	Generalized tonic-clonic seizures with focal onset (HP:0007334)
Generalized tonic-clonic seizures without focal onset (HP:0025190)	Atonic seizures (HP:0010819)	Focal seizures (HP:0007359)
EEG abnormality (HP:0002353)	EEG with focal epileptiform discharges (HP:0011185)	Multifocal epileptiform discharges (HP:0010841)
EEG with generalized epileptiform discharges (HP:0011198)	Abnormality of the cerebral cortex (HP:0002538)	Focal white matter lesions (HP:0007042)
Diffuse white matter abnormalities (HP:0007204)	Abnormality of the cerebellum (HP:0001317)	Abnormality of the pons (HP:0007361)
Abnormality of the basal ganglia (HP:0002134)	Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)
Autism (HP:0000717)	Cognitive impairment (HP:0100543)	Dementia (HP:0000726)
Microcephaly (HP:0000252)	Macrocephaly (HP:0000256)	Abnormal facial shape (HP:0001999)
Abnormality of the skull (HP:0000929)	Abnormality of the skin (HP:0000951)	Abnormality of the ear (HP:0000598)
Abnormality of the eye (HP:0000478)	Abnormality of the skeletal system (HP:0000924)	Abnormality of the cardiovascular system (HP:0001626)
Abnormality of the respiratory system (HP:0002086)	Abnormality of the genitourinary system (HP:0000119)	Abnormality of the endocrine system (HP:0000818)
Abnormality of the gastrointestinal tract (HP:0011024)		

Epilepsy plus other features clinical tests (41926)

<table>
<tr><td>Level 3 Title</td><td>Inherited Epilepsy Syndromes (10990)</td></tr>
<tr><td>Level 4 Title</td><td>Epilepsy plus other features (41924)</td></tr>
<tr><td>Clinical Tests</td><td>Clinical test guidance:
General Imaging Diagnostics refers to CT or MRI brain
Entries ordered left to right in table
<table><tr><td>General Imaging Diagnostics (33633.1)</td><td>Intelligence Quotient Assessment (30224.1)</td><td>General Non-imaging Diagnostics (34838.1)</td></tr></table></td></tr>
</table>

Motor and Sensory Disorders of the PNS (10991)

Charcot-Marie-Tooth disease (15111)

Hereditary motor and sensory neuropathy phenotypes (28665)

Level 3 Title	Motor and Sensory Disorders of the PNS (10991)
Level 4 Title	Charcot-Marie-Tooth disease (15111)
Phenotypes	Entries ordered left to right in table

Peripheral neuropathy (HP:0009830)	Demyelinating peripheral neuropathy (HP:0007108)	Peripheral axonal neuropathy (HP:0003477)
Pes cavus (HP:0001761)	Foot dorsiflexor weakness (HP:0009027)	Hyperreflexia in upper limbs (HP:0007350)
Lower limb hyperreflexia (HP:0002395)	Hyporeflexia of lower limbs (HP:0002600)	Hyporeflexia of upper limbs (HP:0012391)
Upper limb spasticity (HP:0006986)	Lower limb spasticity (HP:0002061)	Proximal muscle weakness in upper limbs (HP:0008997)
Proximal muscle weakness in lower limbs (HP:0008994)	Distal upper limb muscle weakness (HP:0008959)	Distal lower limb muscle weakness (HP:0009053)
Axial muscle weakness (HP:0003327)	Skeletal muscle atrophy (HP:0003202)	Impaired temperature sensation (HP:0010829)
Impaired pain sensation (HP:0007328)	Impaired proprioception (HP:0010831)	Impaired vibratory sensation (HP:0002495)
Ataxia (HP:0001251)	Choreoathetosis (HP:0001266)	Seizures (HP:0001250)
Intellectual disability (HP:0001249)	Motor delay (HP:0001270)	Fasciculations (HP:0002380)
Hearing impairment (HP:0000365)	Cataract (HP:0000518)	Dementia (HP:0000726)
Abnormality of the cranial nerves (HP:0001291)	Contractures of the joints of the upper limbs (HP:0100360)	Contractures of the joints of the lower limbs (HP:0005750)
Respiratory insufficiency (HP:0002093)		

Charcot-Marie-Tooth disease clinical tests (30934)

<table>
<tr><td>Level 3 Title</td><td>Motor and Sensory Disorders of the PNS (10991)</td></tr>
<tr><td>Level 4 Title</td><td>Charcot-Marie-Tooth disease (15111)</td></tr>
<tr><td>Clinical Tests</td><td>N.B. Clinical test guidance:
Biopsy refers to muscle and/or nerve biopsy
Imaging diagnostics refers to Muscle MRI and/or plain x-rays
Non-imaging diagnostics refers to EMG, Motor nerve conduction studies and Repetitive nerve stimulation
Entries ordered left to right in table
<table><tr><td>Forced vital capacity (30180.2)</td><td>Other enzymes (33123.1)</td><td>General Biopsy (33614.1)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td>General Non-imaging Diagnostics (34838.1)</td><td></td></tr></table></td></tr>
</table>

Paediatric motor neuronopathies (11099)

Spinal Muscular Atrophy phenotypes (28594)

Level 3 Title	Motor and Sensory Disorders of the PNS (10991)
Level 4 Title	Paediatric motor neuronopathies (11099)
Phenotypes	Entries ordered left to right in table

Motor axonal neuropathy (HP:0007002)	Demyelinating motor neuropathy (HP:0007220)	Sensory axonal neuropathy (HP:0003390)
Demyelinating sensory neuropathy (HP:0011402)	Skeletal muscle atrophy (HP:0003202)	Lower limb amyotrophy (HP:0007210)
Upper limb amyotrophy (HP:0009129)	Progressive muscle weakness (HP:0003323)	Limb muscle weakness (HP:0003690)
Axial muscle weakness (HP:0003327)	Facial hypotonia (HP:0000297)	Dysphonia (HP:0001618)
Reduced tendon reflexes (HP:0001315)	Hyperactive deep tendon reflexes (HP:0006801)	Hypertonia (HP:0001276)
Muscular hypotonia (HP:0001252)	Muscle fibrillation (HP:0010546)	Dystonia (HP:0001332)
Tremor (HP:0001337)	Seizures (HP:0001250)	Ataxia (HP:0001251)
Choreoathetosis (HP:0001266)	Dysarthria (HP:0001260)	Dysphagia (HP:0002015)
Tongue fasciculations (HP:0001308)	Gastrostomy tube feeding in infancy (HP:0011471)	Sensory impairment (HP:0003474)
Dysautonomia (HP:0002459)	Dementia (HP:0000726)	Respiratory insufficiency (HP:0002093)
Personality changes (HP:0000751)	Behavioral abnormality (HP:0000708)	Intellectual disability (HP:0001249)
Motor delay (HP:0001270)	Loss of ability to walk (HP:0006957)	Intrauterine growth retardation (HP:0001511)
Decreased fetal movement (HP:0001558)	Polyhydramnios (HP:0001561)	Multiple prenatal fractures (HP:0005855)
Flexion contracture (HP:0001371)	Arrhythmia (HP:0011675)	Cardiomyopathy (HP:0001638)
Scoliosis (HP:0002650)	Hearing impairment (HP:0000365)	Abnormality of the eye (HP:0000478)
Ptosis (HP:0000508)	Glaucoma (HP:0000501)	Ophthalmoparesis (HP:0000597)
Optic atrophy (HP:0000648)	Nystagmus (HP:0000639)	Cataract (HP:0000518)
Cherry red spot of the macula (HP:0010729)		

Spinal Muscular Atrophy clinical tests (30949)

Level 3 Title	Motor and Sensory Disorders of the PNS (10991)
Level 4 Title	Paediatric motor neuronopathies (11099)
Clinical Tests	N.B. Clinical test guidance: Biopsy refers to muscle and/or nerve biopsy Imaging diagnostics refers to Muscle MRI and/or plain x-rays Non-imaging diagnostics refers to EMG, Motor nerve conduction studies and Repetitive nerve stimulation Entries ordered left to right in table

Bone profile (30317.2)	Other enzymes (33123.1)	Congenital Myaesthenia Antibodies (33147.2)
General Biopsy (33614.1)	General Non-imaging Diagnostics (34838.1)	General Imaging Diagnostics (33633.1)

Pain channelopathies (82148)

Hereditary motor and sensory neuropathy phenotypes (28665)

Level 3 Title	Motor and Sensory Disorders of the PNS (10991)
Level 4 Title	Pain channelopathies (82148)
Phenotypes	Entries ordered left to right in table

Peripheral neuropathy (HP:0009830)	Demyelinating peripheral neuropathy (HP:0007108)	Peripheral axonal neuropathy (HP:0003477)
Pes cavus (HP:0001761)	Foot dorsiflexor weakness (HP:0009027)	Hyperreflexia in upper limbs (HP:0007350)
Lower limb hyperreflexia (HP:0002395)	Hyporeflexia of lower limbs (HP:0002600)	Hyporeflexia of upper limbs (HP:0012391)
Upper limb spasticity (HP:0006986)	Lower limb spasticity (HP:0002061)	Proximal muscle weakness in upper limbs (HP:0008997)
Proximal muscle weakness in lower limbs (HP:0008994)	Distal upper limb muscle weakness (HP:0008959)	Distal lower limb muscle weakness (HP:0009053)
Axial muscle weakness (HP:0003327)	Skeletal muscle atrophy (HP:0003202)	Impaired temperature sensation (HP:0010829)
Impaired pain sensation (HP:0007328)	Impaired proprioception (HP:0010831)	Impaired vibratory sensation (HP:0002495)
Ataxia (HP:0001251)	Choreoathetosis (HP:0001266)	Seizures (HP:0001250)
Intellectual disability (HP:0001249)	Motor delay (HP:0001270)	Fasciculations (HP:0002380)
Hearing impairment (HP:0000365)	Cataract (HP:0000518)	Dementia (HP:0000726)
Abnormality of the cranial nerves (HP:0001291)	Contractures of the joints of the upper limbs (HP:0100360)	Contractures of the joints of the lower limbs (HP:0005750)
Respiratory insufficiency (HP:0002093)		

Charcot-Marie-Tooth disease clinical tests (30934)

<table>
<tr><td>Level 3 Title</td><td>Motor and Sensory Disorders of the PNS (10991)</td></tr>
<tr><td>Level 4 Title</td><td>Pain channelopathies (82148)</td></tr>
<tr><td>Clinical Tests</td><td>Entries ordered left to right in table<table>
<tr><td>Forced vital capacity (30180.2)</td><td>Other enzymes (33123.1)</td><td>General Biopsy (33614.1)</td></tr>
<tr><td>General Imaging Diagnostics (33633.1)</td><td>General Non-imaging Diagnostics (34838.1)</td><td></td></tr>
</table></td></tr>
</table>

Neurodegenerative disorders (10992)

Early onset and familial Parkinson's Disease (11100)

Parkinson phenotypes (29179)

Level 3 Title	Neurodegenerative disorders (10992)
Level 4 Title	Early onset and familial Parkinson's Disease (11100)
Phenotypes	Entries ordered left to right in table

Parkinsonism (HP:0001300)	Parkinsonism with favorable response to dopaminergic medication (HP:0002548)	Ophthalmoparesis (HP:0000597)
Vertical supranuclear gaze palsy (HP:0000511)	Abnormality of saccadic eye movements (HP:0000570)	Optic atrophy (HP:0000648)
Dysarthria (HP:0001260)	Bulbar signs (HP:0002483)	Dysphagia (HP:0002015)
Amyotrophic lateral sclerosis (HP:0007354)	Resting tremor (HP:0002322)	Bradykinesia (HP:0002067)
Cogwheel rigidity (HP:0002396)	Short stepped shuffling gait (HP:0007311)	Postural instability (HP:0002172)
Spasticity (HP:0001257)	Upper limb spasticity (HP:0006986)	Lower limb spasticity (HP:0002061)
Ataxia (HP:0001251)	Dystonia (HP:0001332)	Skeletal muscle atrophy (HP:0003202)
Constipation (HP:0002019)	Orthostatic hypotension (HP:0001278)	Anosmia (HP:0000458)
Abnormal rapid eye movement sleep (HP:0002494)	Abnormality of higher mental function (HP:0011446)	Frontotemporal dementia (HP:0002145)
Psychosis (HP:0000709)	Impaired vibratory sensation (HP:0002495)	Impaired tactile sensation (HP:0010830)
Impaired proprioception (HP:0010831)	Impaired temperature sensation (HP:0010829)	Impaired pain sensation (HP:0007328)
Motor axonal neuropathy (HP:0007002)	Demyelinating motor neuropathy (HP:0007220)	Sensory axonal neuropathy (HP:0003390)
Demyelinating sensory neuropathy (HP:0011402)	Abnormality of the basal ganglia (HP:0002134)	Focal white matter lesions (HP:0007042)
Cerebral atrophy (HP:0002059)	Cerebellar atrophy (HP:0001272)	

Complex Parkinsonism (includes pallido-pyramidal syndromes) (15112)

Parkinson phenotypes (29179)

Level 3 Title	Neurodegenerative disorders (10992)
Level 4 Title	Complex Parkinsonism (includes pallido-pyramidal syndromes) (15112)
Phenotypes	Entries ordered left to right in table

Parkinsonism (HP:0001300)	Parkinsonism with favorable response to dopaminergic medication (HP:0002548)	Ophthalmoparesis (HP:0000597)
Vertical supranuclear gaze palsy (HP:0000511)	Abnormality of saccadic eye movements (HP:0000570)	Optic atrophy (HP:0000648)
Dysarthria (HP:0001260)	Bulbar signs (HP:0002483)	Dysphagia (HP:0002015)
Amyotrophic lateral sclerosis (HP:0007354)	Resting tremor (HP:0002322)	Bradykinesia (HP:0002067)
Cogwheel rigidity (HP:0002396)	Short stepped shuffling gait (HP:0007311)	Postural instability (HP:0002172)
Spasticity (HP:0001257)	Upper limb spasticity (HP:0006986)	Lower limb spasticity (HP:0002061)
Ataxia (HP:0001251)	Dystonia (HP:0001332)	Skeletal muscle atrophy (HP:0003202)
Constipation (HP:0002019)	Orthostatic hypotension (HP:0001278)	Anosmia (HP:0000458)
Abnormal rapid eye movement sleep (HP:0002494)	Abnormality of higher mental function (HP:0011446)	Frontotemporal dementia (HP:0002145)
Psychosis (HP:0000709)	Impaired vibratory sensation (HP:0002495)	Impaired tactile sensation (HP:0010830)
Impaired proprioception (HP:0010831)	Impaired temperature sensation (HP:0010829)	Impaired pain sensation (HP:0007328)
Motor axonal neuropathy (HP:0007002)	Demyelinating motor neuropathy (HP:0007220)	Sensory axonal neuropathy (HP:0003390)
Demyelinating sensory neuropathy (HP:0011402)	Abnormality of the basal ganglia (HP:0002134)	Focal white matter lesions (HP:0007042)
Cerebral atrophy (HP:0002059)	Cerebellar atrophy (HP:0001272)	

Early onset dementia (15113)

Fronto-temporal dementia phenotypes (28658)

Level 3 Title	Neurodegenerative disorders (10992)
Level 4 Title	Early onset dementia (15113)
Phenotypes	Entries ordered left to right in table

Dementia (HP:0000726)	Frontotemporal dementia (HP:0002145)	Behavioral abnormality (HP:0000708)
Progressive language deterioration (HP:0007064)	Aphasia (HP:0002381)	Progressive extrapyramidal movement disorder (HP:0007153)
Parkinsonism (HP:0001300)	Amyotrophic lateral sclerosis (HP:0007354)	Bulbar signs (HP:0002483)
Neurological speech impairment (HP:0002167)	Dysphagia (HP:0002015)	Abnormality of the eye (HP:0000478)
Ophthalmoparesis (HP:0000597)	Abnormality of saccadic eye movements (HP:0000570)	Slow saccadic eye movements (HP:0000514)
Dysmetric saccades (HP:0000641)	EMG abnormality (HP:0003457)	EMG: neuropathic changes (HP:0003445)
Frontotemporal cerebral atrophy (HP:0006892)	Abnormality of the basal ganglia (HP:0002134)	Abnormal lower motor neuron morphology (HP:0002366)

Fronto-temporal dementia clinical tests (30950)

Level 3 Title	Neurodegenerative disorders (10992)
Level 4 Title	Early onset dementia (15113)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Amyotrophic lateral sclerosis or motor neuron disease (15114)

Hereditary spastic paraplegia phenotypes (27781)

Level 3 Title	Neurodegenerative disorders (10992)
Level 4 Title	Amyotrophic lateral sclerosis or motor neuron disease (15114)
Phenotypes	Entries ordered left to right in table

Abnormality of higher mental function (HP:0011446)	Cognitive impairment (HP:0100543)	Psychosis (HP:0000709)
Neurological speech impairment (HP:0002167)	Dysarthria (HP:0001260)	Dysphagia (HP:0002015)
Abnormality of the eye (HP:0000478)	Cataract (HP:0000518)	Congenital cataract (HP:0000519)
Optic atrophy (HP:0000648)	Abnormality of eye movement (HP:0000496)	Spasticity (HP:0001257)
Abnormal pyramidal signs (HP:0007256)	Hyperreflexia in upper limbs (HP:0007350)	Upper limb spasticity (HP:0006986)
Skeletal muscle atrophy (HP:0003202)	Fasciculations (HP:0002380)	Proximal upper limb amyotrophy (HP:0008948)
Proximal lower limb amyotrophy (HP:0008956)	Distal upper limb amyotrophy (HP:0007149)	Distal lower limb amyotrophy (HP:0008944)
Ataxia (HP:0001251)	Gait ataxia (HP:0002066)	Limb ataxia (HP:0002070)
Abnormality of the basal ganglia (HP:0002134)	Dystonia (HP:0001332)	Parkinsonism (HP:0001300)
Myoclonus (HP:0001336)	Sensory impairment (HP:0003474)	Impaired vibratory sensation (HP:0002495)
Impaired tactile sensation (HP:0010830)	Impaired proprioception (HP:0010831)	Impaired temperature sensation (HP:0010829)
Impaired pain sensation (HP:0007328)	Abnormality of central motor conduction (HP:0012079)	Motor axonal neuropathy (HP:0007002)
Demyelinating motor neuropathy (HP:0007220)	Distal peripheral sensory neuropathy (HP:0007067)	Sensory axonal neuropathy (HP:0003390)
Demyelinating sensory neuropathy (HP:0011402)	Mixed demyelinating and axonal polyneuropathy (HP:0007327)	Hypoplasia of the corpus callosum (HP:0002079)
Focal white matter lesions (HP:0007042)	Cerebral atrophy (HP:0002059)	Cerebellar atrophy (HP:0001272)

Hereditary spastic paraplegia clinical tests (30951)

<table>
<tr><td>Level 3 Title</td><td>Neurodegenerative disorders (10992)</td></tr>
<tr><td>Level 4 Title</td><td>Amyotrophic lateral sclerosis or motor neuron disease (15114)</td></tr>
<tr><td>Clinical Tests</td><td>N.B. Clinical test guidance:
Imaging diagnostics refers to MRI brain
Non-imaging diagnostics refers to Motor evoked potentials (MEP), Motor nerve conduction studies, Sensory nerve conduction studies
Entries ordered left to right in table
<table><tr><td>Spastic Paraplegia Assessment (30230.1)</td><td>General Imaging Diagnostics (33633.1)</td><td>General Non-imaging Diagnostics (34838.1)</td></tr></table></td></tr>
</table>

Neurodevelopmental disorders (10993)

Classical tuberous sclerosis (11101)

Classical tuberous sclerosis phenotypes (29180)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Classical tuberous sclerosis (11101)
Phenotypes	Entries ordered left to right in table

Cafe-au-lait spot (HP:0000957)	Hypomelanotic macule (HP:0009719)	Adenoma sebaceum (HP:0009720)
Shagreen patch (HP:0009721)	Subungual fibromas (HP:0009724)	Subcutaneous nodule (HP:0001482)
Projection of scalp hair onto lateral cheek (HP:0009554)	Gingival fibromatosis (HP:0000169)	Dental enamel pits (HP:0009722)
Intellectual disability (HP:0001249)	Specific learning disability (HP:0001328)	Autism (HP:0000717)
Attention deficit hyperactivity disorder (HP:0007018)	Seizures (HP:0001250)	Infantile spasms (HP:0012469)
Cerebral calcification (HP:0002514)	Subependymal nodules (HP:0009716)	Cortical tubers (HP:0009717)
Astrocytoma (HP:0009592)	Ependymoma (HP:0002888)	Optic glioma (HP:0009734)
Achromatic retinal patches (HP:0009727)	Retinal hamartoma (HP:0009594)	Renal angiomyolipoma (HP:0006772)
Renal hamartoma (HP:0008696)	Renal cyst (HP:0000107)	Renal cell carcinoma (HP:0005584)
Hypothyroidism (HP:0000821)	Precocious puberty (HP:0000826)	Cardiac rhabdomyoma (HP:0009729)
Wolff-Parkinson-White syndrome (HP:0001716)	Abnormality of the respiratory system (HP:0002086)	Pulmonary lymphangiomyomatosis (HP:0012798)
Chordoma (HP:0010762)	Hamartomatous polyposis (HP:0004390)	Bone cyst (HP:0012062)
Hamartoma (HP:0010566)		

Intellectual disability (11102)

Intellectual disability phenotypes (27784)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Intellectual disability (11102)

Phenotypes

Entries ordered left to right in table

Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)	Delayed fine motor development (HP:0010862)
Delayed gross motor development (HP:0002194)	Inability to walk (HP:0002540)	Delayed speech and language development (HP:0000750)
Autistic behavior (HP:0000729)	Small for gestational age (HP:0001518)	Failure to thrive (HP:0001508)
Disproportionate short stature (HP:0003498)	Proportionate short stature (HP:0003508)	Large for gestational age (HP:0001520)
Tall stature (HP:0000098)	Microcephaly (HP:0000252)	Macrocephaly (HP:0000256)
Hyperpigmentation of the skin (HP:0000953)	Hypopigmentation of the skin (HP:0001010)	Vascular skin abnormality (HP:0011276)
Abnormal facial shape (HP:0001999)	Abnormality of the eye (HP:0000478)	Abnormality of the nose (HP:0000366)
Abnormality of the outer ear (HP:0000356)	Abnormality of the palate (HP:0000174)	Abnormality of finger (HP:0001167)
Abnormality of the thumb (HP:0001172)	Abnormality of toe (HP:0001780)	Abnormality of cardiovascular system morphology (HP:0030680)
Abnormality of the gastrointestinal tract (HP:0011024)	Abnormal renal morphology (HP:0012210)	Abnormality of female external genitalia (HP:0000055)
Abnormality of male external genitalia (HP:0000032)	Seizures (HP:0001250)	Generalized hypotonia (HP:0001290)
Spasticity (HP:0001257)	Chorea (HP:0002072)	Ataxia (HP:0001251)
Dystonia (HP:0001332)	Morphological abnormality of the central nervous system (HP:0002011)	Toxemia of pregnancy (HP:0100603)
Maternal diabetes (HP:0009800)	Maternal hypertension (HP:0008071)	Abnormality of prenatal development or birth (HP:0001197)
Increased nuchal translucency (HP:0010880)	Polyhydramnios (HP:0001561)	Oligohydramnios (HP:0001562)

Intellectual disability clinical tests (30952)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Intellectual disability (11102)
Clinical Tests	Entries ordered left to right in table

APGAR score (30207.1)	General Imaging Diagnostics (33633.1)

Holoprosencephaly (36519)

Holoprosencephaly phenotypes (36648)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Holoprosencephaly (36519)
Phenotypes	Entries ordered left to right in table

Median cleft lip and palate (HP:0008501)	Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)
Holoprosencephaly (HP:0001360)	Alobar holoprosencephaly (HP:0006988)	Semilobar holoprosencephaly (HP:0002507)
Cerebellar malformation (HP:0002438)	Fusion of the cerebellar hemispheres (HP:0006899)	Abnormality of the cerebellar peduncle (HP:0011931)
Abnormality of the dentate nucleus (HP:0100321)	Microcephaly (HP:0000252)	Ptosis (HP:0000508)
Hypotelorism (HP:0000601)	Agenesis of corpus callosum (HP:0001274)	Partial agenesis of the corpus callosum (HP:0001338)
Single median maxillary incisor (HP:0006315)	Anterior pituitary hypoplasia (HP:0010627)	Panhypopituitarism (HP:0000871)
Anterior pituitary agenesis (HP:0010626)	Growth hormone deficiency (HP:0000824)	Optic nerve hypoplasia (HP:0000609)
Choanal stenosis (HP:0000452)	Choanal atresia (HP:0000453)	Maternal diabetes (HP:0009800)
Maternal teratogenic exposure (HP:0011438)	Patchy alopecia (HP:0002232)	

Holoprosencephaly clinical tests (36521)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Holoprosencephaly (36519)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostsics refers to MRI brain and/or medical photographs of facial features and other physical features as appropriate Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Rhomboencephalosynapsis (36603)

Holoprosencephaly phenotypes (36648)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Rhomboencephalosynapsis (36603)
Phenotypes	Entries ordered left to right in table

Median cleft lip and palate (HP:0008501)	Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)
Holoprosencephaly (HP:0001360)	Alobar holoprosencephaly (HP:0006988)	Semilobar holoprosencephaly (HP:0002507)
Cerebellar malformation (HP:0002438)	Fusion of the cerebellar hemispheres (HP:0006899)	Abnormality of the cerebellar peduncle (HP:0011931)
Abnormality of the dentate nucleus (HP:0100321)	Microcephaly (HP:0000252)	Ptosis (HP:0000508)
Hypotelorism (HP:0000601)	Agenesis of corpus callosum (HP:0001274)	Partial agenesis of the corpus callosum (HP:0001338)
Single median maxillary incisor (HP:0006315)	Anterior pituitary hypoplasia (HP:0010627)	Panhypopituitarism (HP:0000871)
Anterior pituitary agenesis (HP:0010626)	Growth hormone deficiency (HP:0000824)	Optic nerve hypoplasia (HP:0000609)
Choanal stenosis (HP:0000452)	Choanal atresia (HP:0000453)	Maternal diabetes (HP:0009800)
Maternal teratogenic exposure (HP:0011438)	Patchy alopecia (HP:0002232)	

Holoprosencephaly clinical tests (36521)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Rhomboencephalosynapsis (36603)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain and/or medical photographs of facial features and other physical features as appropriate Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Malformations of cortical development (36526)

Malformations of cortical development phenotypes (36651)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Malformations of cortical development (36526)

Phenotypes

Entries ordered left to right in table

Lissencephaly (HP:0001339)	Type I lissencephaly (HP:0006818)	Pachygyria (HP:0001302)
Heterotopia (HP:0002282)	Polymicrogyria (HP:0002126)	Frontoparietal polymicrogyria (HP:0007095)
Perisylvian polymicrogyria (HP:0012650)	Unilateral polymicrogyria (HP:0006927)	Schizencephaly (HP:0010636)
Hemimegalencephaly (HP:0007206)	Gray matter heterotopias (HP:0002281)	Periventricular gray matter heterotopia (HP:0007165)
Cortical dysplasia (HP:0002539)	Frontoparietal cortical dysplasia (HP:0006930)	Cortical gyral simplification (HP:0009879)
Hydranencephaly (HP:0002324)	Microcephaly (HP:0000252)	Congenital microcephaly (HP:0011451)
Postnatal microcephaly (HP:0005484)	Progressive microcephaly (HP:0000253)	Macrocephaly (HP:0000256)
Macrocephaly at birth (HP:0004488)	Postnatal macrocephaly (HP:0005490)	Progressive macrocephaly (HP:0004481)
Hydrocephalus (HP:0000238)	Aqueductal stenosis (HP:0002410)	Communicating hydrocephalus (HP:0001334)
Noncommunicating hydrocephalus (HP:0010953)	Agenesis of corpus callosum (HP:0001274)	Hypoplasia of the corpus callosum (HP:0002079)
Abnormality of the basal ganglia (HP:0002134)	Abnormality of the internal capsule (HP:0012502)	Dilation of lateral ventricles (HP:0006956)
Dilated third ventricle (HP:0007082)	Dilated fourth ventricle (HP:0002198)	Colpocephaly (HP:0030048)
Cerebellar agenesis (HP:0012642)	Cerebellar hypoplasia (HP:0001321)	Cerebellar atrophy (HP:0001272)
Cerebellar dysplasia (HP:0007033)	Cerebellar cyst (HP:0002350)	Agenesis of cerebellar vermis (HP:0002335)

	Cerebellar vermis hypoplasia (HP:0001320)	Partial absence of cerebellar vermis (HP:0002951)	Dysgenesis of the cerebellar vermis (HP:0002195)
	Hypoplasia of the brainstem (HP:0002365)	Cerebral atrophy (HP:0002059)	Diffuse white matter abnormalities (HP:0007204)
	Focal white matter lesions (HP:0007042)	Leukoencephalopathy (HP:0002352)	Subcortical white matter calcifications (HP:0007346)
	Basal ganglia calcification (HP:0002135)	Enlarged hippocampus (HP:0100961)	Septo-optic dysplasia (HP:0100842)

Malformations of cortical development clinical tests (36528)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Malformations of cortical development (36526)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to medical photographs and MRI brain. Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	Urine electrolytes (33187.2)

Fetal structural CNS abnormalities (36850)

Fetal structural CNS abnormalities phenotypes (36912)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Fetal structural CNS abnormalities (36850)
Phenotypes	Entries ordered left to right in table

Abnormality of neuronal migration (HP:0002269)	Lissencephaly (HP:0001339)	Polymicrogyria (HP:0002126)
Pachygyria (HP:0001302)	Cortical dysplasia (HP:0002539)	White matter neuronal heterotopia (HP:0007314)
Periventricular gray matter heterotopia (HP:0007165)	Cerebral white matter hypoplasia (HP:0012430)	Schizencephaly (HP:0010636)
Aplasia/Hypoplasia of the corpus callosum (HP:0007370)	Absent septum pellucidum (HP:0001331)	Abnormality of the cerebrum (HP:0002060)
Small cerebral cortex (HP:0002472)	Colpocephaly (HP:0030048)	Abnormality of the cerebellum (HP:0001317)
Aplasia/Hypoplasia of the cerebellar vermis (HP:0006817)	Cerebellar hypoplasia (HP:0001321)	Enlarged cerebellum (HP:0012081)
Olivopontocerebellar hypoplasia (HP:0006955)	Cerebellar vermis hypoplasia (HP:0001320)	Inferior vermis hypoplasia (HP:0007068)
Enlarged fetal cisterna magna (HP:0011427)	Dandy-Walker malformation (HP:0001305)	Alobar holoprosencephaly (HP:0006988)
Lobar holoprosencephaly (HP:0006870)	Semilobar holoprosencephaly (HP:0002507)	Hydranencephaly (HP:0002324)
Molar tooth sign on MRI (HP:0002419)	Fusion of the left and right thalami (HP:0010664)	Intracranial cystic lesion (HP:0010576)
Ventriculomegaly (HP:0002119)	Aqueductal stenosis (HP:0002410)	Dilated fourth ventricle (HP:0002198)
Dilation of lateral ventricles (HP:0006956)	Macrocephaly (HP:0000256)	Microcephaly (HP:0000252)
Megalencephaly (HP:0001355)	Nonimmune hydrops fetalis (HP:0001790)	Increased nuchal translucency (HP:0010880)
Fetal ascites (HP:0001791)	Fetal cystic hygroma (HP:0010878)	Polyhydramnios (HP:0001561)
Decreased fetal movement (HP:0001558)	Contractures of the joints of the upper limbs (HP:0100360)	Contractures of the joints of the lower limbs (HP:0005750)

Fetal structural CNS abnormalities clinical tests (36873)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Fetal structural CNS abnormalities (36850)
Clinical Tests	NB. Clinical test guidance: General Imaging Diagnostics refers to in-/ex-utero MRI scan where relevant Entries ordered left to right in table

TORCH screen (30122.2)	Metabolic biochemistry (33160.2)

Pontine tegmental cap dysplasia (55493)

Pontine tegmental cap dysplasia phenotypes (68119)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Pontine tegmental cap dysplasia (55493)
Phenotypes	Entries ordered left to right in table

Hypoplasia of the ventral pons (HP:0006850)	Abnormality of the cerebellar peduncle (HP:0011931)	Cerebellar vermis hypoplasia (HP:0001320)
Cerebellar hypoplasia (HP:0001321)	Abnormality of brainstem morphology (HP:0002363)	Delayed fine motor development (HP:0010862)
Delayed gross motor development (HP:0002194)	Delayed speech and language development (HP:0000750)	Abnormality of the cornea (HP:0000481)
Facial palsy (HP:0010628)	Abnormality of the vestibulocochlear nerve (HP:0009591)	Sensorineural hearing impairment (HP:0000407)
Feeding difficulties (HP:0011968)	Abnormality of the ribs (HP:0000772)	Abnormality of the vertebrae (HP:0003468)
Morphological abnormality of the gastrointestinal tract (HP:0012718)	Abnormality of the internal auditory canal (HP:0011384)	Syndactyly (HP:0001159)
Abnormal heart morphology (HP:0001627)		

Pontine tegmental cap dysplasia clinical tests (55494)

Level 3 Title	Neurodevelopmental disorders (10993)
Level 4 Title	Pontine tegmental cap dysplasia (55493)
Clinical Tests	NB Clinical testing guidance: General Imaging Diagnostics refers to USS – antenatal and Neuroradiology General Non-imaging Diagnostics refers to EEG Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	General Non-imaging Diagnostics (34838.1)

Neuromuscular disorders (10994)

Congenital muscular dystrophy (15135)

Congenital muscular dystrophy phenotypes (28647)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Congenital muscular dystrophy (15135)
Phenotypes	Entries ordered left to right in table

Myopathy (HP:0003198)	Abnormality of muscle morphology (HP:0011805)	Skeletal muscle atrophy (HP:0003202)
Skeletal muscle hypertrophy (HP:0003712)	Limb muscle weakness (HP:0003690)	Axial muscle weakness (HP:0003327)
Decreased fetal movement (HP:0001558)	Flexion contracture (HP:0001371)	Congenital hip dislocation (HP:0001374)
Joint laxity (HP:0001388)	Neonatal hypotonia (HP:0001319)	Motor delay (HP:0001270)
Loss of ability to walk (HP:0006957)	Inability to walk (HP:0002540)	Intellectual disability (HP:0001249)
Seizures (HP:0001250)	Abnormality of the cerebrum (HP:0002060)	Abnormality of the cerebellum (HP:0001317)
Abnormal cortical gyration (HP:0002536)	Abnormality of the cerebral white matter (HP:0002500)	Abnormality of the eye (HP:0000478)
Ptosis (HP:0000508)	Cataract (HP:0000518)	Ophthalmoparesis (HP:0000597)
Arrhythmia (HP:0011675)	Cardiomyopathy (HP:0001638)	Dysphonia (HP:0001618)
Dysphagia (HP:0002015)	Dysarthria (HP:0001260)	Respiratory insufficiency (HP:0002093)
Scoliosis (HP:0002650)	Spinal rigidity (HP:0003306)	Atypical scarring of skin (HP:0000987)
Follicular hyperkeratosis (HP:0007502)	Abnormality of muscle fibers (HP:0004303)	Elevated serum creatine phosphokinase (HP:0003236)
Abnormal muscle fiber alpha dystroglycan (HP:0030112)	Abnormal muscle fiber merosin expression (HP:0030090)	Reduced muscle collagen VI (HP:0030095)

Congenital muscular dystrophy clinical tests (30953)

<table>
<tr><td>Level 3 Title</td><td>Neuromuscular disorders (10994)</td></tr>
<tr><td>Level 4 Title</td><td>Congenital muscular dystrophy (15135)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Biopsy refers to muscle biopsy Imaging diagnostics refers to Brain, Spine and/or Muscle MRI and Muscle Ultrasound
Non-imaging diagnostics refers to EMG, Motor nerve conduction studies, Sensory nerve conduction studies
Other enzymes refers to CK
Entries ordered left to right in table
<table><tr><td>Forced vital capacity (30180.2)</td><td>Other enzymes (33123.1)</td><td>General Biopsy (33614.1)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td>General Non-imaging Diagnostics (34838.1)</td><td></td></tr></table></td></tr>
</table>

Congenital myopathy (11103)

Congenital myopathy phenotypes (40400)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Congenital myopathy (11103)
Phenotypes	Entries ordered left to right in table

Myopathy (HP:0003198)	Abnormality of muscle morphology (HP:0011805)	Skeletal muscle atrophy (HP:0003202)
Limb muscle weakness (HP:0003690)	Axial muscle weakness (HP:0003327)	Facial hypotonia (HP:0000297)
Decreased fetal movement (HP:0001558)	Bulbar signs (HP:0002483)	Flexion contracture (HP:0001371)
Congenital hip dislocation (HP:0001374)	Joint laxity (HP:0001388)	Neonatal hypotonia (HP:0001319)
Motor delay (HP:0001270)	Loss of ability to walk (HP:0006957)	Inability to walk (HP:0002540)
Intellectual disability (HP:0001249)	Ptosis (HP:0000508)	Ophthalmoparesis (HP:0000597)
Dysphonia (HP:0001618)	Dysphagia (HP:0002015)	Dysarthria (HP:0001260)
Cardiomyopathy (HP:0001638)	Malignant hyperthermia (HP:0002047)	Scoliosis (HP:0002650)
Spinal rigidity (HP:0003306)	Large for gestational age (HP:0001520)	High, narrow palate (HP:0002705)
Skeletal dysplasia (HP:0002652)	Elevated serum creatine phosphokinase (HP:0003236)	Centrally nucleated skeletal muscle fibers (HP:0003687)
Abnormality of muscle fibers (HP:0004303)	Minicore myopathy (HP:0003789)	Nemaline bodies (HP:0003798)
Muscle fibrillation (HP:0010546)	Type 1 muscle fiber predominance (HP:0003803)	Central core regions in muscle fibers (HP:0030230)

Congenital muscular dystrophy clinical tests (30953)

<table>
<tr><td>Level 3 Title</td><td>Neuromuscular disorders (10994)</td></tr>
<tr><td>Level 4 Title</td><td>Congenital myopathy (11103)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Biopsy refers to muscle biopsy Imaging diagnostics refers to Brain, Spine and/or Muscle MRI and Muscle Ultrasound
Non-imaging diagnostics refers to EMG, Motor nerve conduction studies, Sensory nerve conduction studies
Other enzymes refers to CK
Entries ordered left to right in table
<table><tr><td>Forced vital capacity (30180.2)</td><td>Other enzymes (33123.1)</td><td>General Biopsy (33614.1)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td>General Non-imaging Diagnostics (34838.1)</td><td></td></tr></table></td></tr>
</table>

Congenital myaesthenia (15136)

CMS phenotypes (28650)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Congenital myaesthenia (15136)
Phenotypes	Entries ordered left to right in table

Skeletal muscle atrophy (HP:0003202)	Abnormality of muscle morphology (HP:0011805)	Lower limb amyotrophy (HP:0007210)
Upper limb amyotrophy (HP:0009129)	Progressive muscle weakness (HP:0003323)	Limb muscle weakness (HP:0003690)
Axial muscle weakness (HP:0003327)	Fatigable weakness (HP:0003473)	Facial hypotonia (HP:0000297)
Dysphonia (HP:0001618)	Dysphagia (HP:0002015)	Loss of ability to walk (HP:0006957)
Inability to walk (HP:0002540)	Peripheral neuropathy (HP:0009830)	Respiratory insufficiency (HP:0002093)
Decreased fetal movement (HP:0001558)	Polyhydramnios (HP:0001561)	Arthrogryposis multiplex congenita (HP:0002804)
Motor delay (HP:0001270)	Abnormality of the eye (HP:0000478)	Ptosis (HP:0000508)
Cataract (HP:0000518)	Ophthalmoparesis (HP:0000597)	Nystagmus (HP:0000639)
Bulbar signs (HP:0002483)	Hearing impairment (HP:0000365)	Intellectual disability (HP:0001249)
Seizures (HP:0001250)	Flexion contracture (HP:0001371)	Scapular winging (HP:0003691)
Scoliosis (HP:0002650)	Spinal rigidity (HP:0003306)	Sensory impairment (HP:0003474)
Arrhythmia (HP:0011675)	Cardiomyopathy (HP:0001638)	Abnormality of the cerebrum (HP:0002060)
Abnormality of the cerebellum (HP:0001317)	Muscle fibrillation (HP:0010546)	

CMS clinical tests (30954)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Congenital myaesthenia (15136)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to muscle biopsy Imaging diagnostics refers to Brain, Spine and/or Muscle MRI and Muscle Ultrasound Non-imaging diagnostics refers to EMG, Motor nerve conduction studies, Sensory nerve conduction studies Other enzymes refers to CK Entries ordered left to right in table

Forced vital capacity (30180.2)	Congenital Myaesthenia Antibodies (33147.2)	General Non-imaging Diagnostics (34838.1)
General Imaging Diagnostics (33633.1)		

Rhabdomyolysis and metabolic muscle disorders (15137)

Rhabdomyolysis and metabolic muscle disorders phenotypes (29181)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Rhabdomyolysis and metabolic muscle disorders (15137)
Phenotypes	Entries ordered left to right in table

Rhabdomyolysis (HP:0003201)	Acute rhabdomyolysis (HP:0008942)	Alcohol-induced rhabdomyolysis (HP:0011440)
Anesthetic-induced rhabdomylosis (HP:0011439)	Exercise-induced rhabdomyolysis (HP:0009045)	Myalgia (HP:0003326)
Muscle weakness (HP:0001324)	Progressive muscle weakness (HP:0003323)	Proximal muscle weakness in upper limbs (HP:0008997)
Proximal muscle weakness in lower limbs (HP:0008994)	Fatigable weakness (HP:0003473)	Progressive inability to walk (HP:0002505)
Difficulty walking (HP:0002355)	Skeletal muscle hypertrophy (HP:0003712)	Skeletal muscle atrophy (HP:0003202)
Acute kidney injury (HP:0001919)	Abnormal renal physiology (HP:0012211)	Recurrent myoglobinuria (HP:0003652)
Abnormal urinary color (HP:0012086)	Elevated serum creatine phosphokinase (HP:0003236)	Malignant hyperthermia (HP:0002047)
Cognitive impairment (HP:0100543)		

Rhabdomyolysis and metabolic muscle disorders clinical tests (30955)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Rhabdomyolysis and metabolic muscle disorders (15137)
Clinical Tests	NB. Clinical test guidance: Non-imaging diagnostics refers to EMG Other enzymes refers to CK Entries ordered left to right in table

Forced vital capacity (30180.2)	Other enzymes (33123.1)	General Non-imaging Diagnostics (34838.1)

Distal myopathies (11104)

Distal myopathies phenotypes (29182)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Distal myopathies (11104)

Phenotypes

Entries ordered left to right in table

Myopathy (HP:0003198)	Skeletal muscle atrophy (HP:0003202)	Skeletal muscle hypertrophy (HP:0003712)
Muscle hypertrophy of the lower extremities (HP:0008968)	Abnormality of muscle morphology (HP:0011805)	Lower limb amyotrophy (HP:0007210)
Upper limb amyotrophy (HP:0009129)	Progressive muscle weakness (HP:0003323)	Limb muscle weakness (HP:0003690)
Axial muscle weakness (HP:0003327)	Fatigable weakness (HP:0003473)	Facial hypotonia (HP:0000297)
Dysphonia (HP:0001618)	Dysphagia (HP:0002015)	Dysarthria (HP:0001260)
Loss of ability to walk (HP:0006957)	Inability to walk (HP:0002540)	Peripheral neuropathy (HP:0009830)
Respiratory insufficiency (HP:0002093)	Motor delay (HP:0001270)	Abnormality of the eye (HP:0000478)
Ptosis (HP:0000508)	Cataract (HP:0000518)	Ophthalmoparesis (HP:0000597)
Optic atrophy (HP:0000648)	Nystagmus (HP:0000639)	Cherry red spot of the macula (HP:0010729)
Bulbar signs (HP:0002483)	Hearing impairment (HP:0000365)	Intellectual disability (HP:0001249)
Seizures (HP:0001250)	Flexion contracture (HP:0001371)	Scoliosis (HP:0002650)
Spinal rigidity (HP:0003306)	Sensory impairment (HP:0003474)	Arrhythmia (HP:0011675)
Cardiomyopathy (HP:0001638)	Diabetes mellitus (HP:0000819)	Myofibrillar myopathy (HP:0003715)
Abnormal mitochondria in muscle tissue (HP:0008316)	Rimmed vacuoles (HP:0003805)	Reduced muscle fiber alpha dystroglycan (HP:0030099)
Reduced muscle fiber alpha sarcoglycan (HP:0030102)	Reduced muscle fiber calpain-3 (HP:0030121)	Reduced muscle fiber merosin (HP:0030092)
Reduced muscle fiber dysferlin (HP:0030115)	Reduced muscle dystrophin expression (HP:0030098)	Reduced muscle fiber emerin (HP:0030118)
Muscle fibrillation (HP:0010546)		

Distal Myopathy clinical tests (30956)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Distal myopathies (11104)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to muscle biopsy Imaging diagnostics refers to Muscle MRI Non-imaging diagnostics refers to EMG, Motor nerve conduction studies, Sensory nerve conduction studies Other enzymes refers to CK Entries ordered left to right in table

Forced vital capacity (30180.2)	General Laboratory Test Report (34329.1)	General Biopsy (33614.1)
General Imaging Diagnostics (33633.1)	General Non-imaging Diagnostics (34838.1)	

Arthrogryposis (15138)

Arthrogryposis phenotypes (29183)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Arthrogryposis (15138)
Phenotypes	Entries ordered left to right in table

Arthrogryposis multiplex congenita (HP:0002804)	Contractures of the joints of the upper limbs (HP:0100360)	Contractures of the joints of the lower limbs (HP:0005750)
Elbow flexion contracture (HP:0002987)	Hip contracture (HP:0003273)	Knee flexion contracture (HP:0006380)
Distal arthrogryposis (HP:0005684)	Camptodactyly of finger (HP:0100490)	Ankle contracture (HP:0006466)
Skeletal muscle atrophy (HP:0003202)	Abnormality of muscle morphology (HP:0011805)	Limb muscle weakness (HP:0003690)
Axial muscle weakness (HP:0003327)	Fatigable weakness (HP:0003473)	Decreased fetal movement (HP:0001558)
Polyhydramnios (HP:0001561)	Oligohydramnios (HP:0001562)	Neonatal hypotonia (HP:0001319)
Inability to walk (HP:0002540)	Intellectual disability (HP:0001249)	Short stature (HP:0004322)
Microcephaly (HP:0000252)	Abnormal facial shape (HP:0001999)	Global developmental delay (HP:0001263)
Skeletal dysplasia (HP:0002652)	Scoliosis (HP:0002650)	Blepharophimosis (HP:0000581)
Cataract (HP:0000518)	Cleft palate (HP:0000175)	Respiratory insufficiency (HP:0002093)
Arrhythmia (HP:0011675)	Cardiomyopathy (HP:0001638)	Abnormal heart morphology (HP:0001627)
Seizures (HP:0001250)	Abnormality of the cerebrum (HP:0002060)	Abnormality of the cerebral white matter (HP:0002500)
Abnormality of the cerebellum (HP:0001317)		

Arthrogryposis clinical tests (30958)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Arthrogryposis (15138)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to muscle biopsy Imaging diagnostics refers to Muscle MRI Non-imaging diagnostics refers to EMG, Motor nerve conduction studies, Sensory nerve conduction studies Other enzymes refers to CK Entries ordered left to right in table

Forced vital capacity (30180.2)	Other enzymes (33123.1)	General Biopsy (33614.1)
General Imaging Diagnostics (33633.1)	General Non-imaging Diagnostics (34838.1)	

Limb girdle muscular dystrophy (11106)

Limb girdle muscular dystrophy phenotypes (27786)

Level 3 Title	Neuromuscular disorders (10994)
Level 4 Title	Limb girdle muscular dystrophy (11106)
Phenotypes	Entries ordered left to right in table

Limb-girdle muscular dystrophy (HP:0006785)	Skeletal muscle atrophy (HP:0003202)	Skeletal muscle hypertrophy (HP:0003712)
Abnormality of muscle morphology (HP:0011805)	Lower limb amyotrophy (HP:0007210)	Upper limb amyotrophy (HP:0009129)
Progressive muscle weakness (HP:0003323)	Limb muscle weakness (HP:0003690)	Axial muscle weakness (HP:0003327)
Fatigable weakness (HP:0003473)	Facial hypotonia (HP:0000297)	Dysphonia (HP:0001618)
Dysphagia (HP:0002015)	Dysarthria (HP:0001260)	Loss of ability to walk (HP:0006957)
Inability to walk (HP:0002540)	Peripheral neuropathy (HP:0009830)	Respiratory insufficiency (HP:0002093)
Motor delay (HP:0001270)	Abnormality of the eye (HP:0000478)	Ptosis (HP:0000508)
Cataract (HP:0000518)	Ophthalmoparesis (HP:0000597)	Bulbar signs (HP:0002483)
Hearing impairment (HP:0000365)	Intellectual disability (HP:0001249)	Seizures (HP:0001250)
Flexion contracture (HP:0001371)	Scapular winging (HP:0003691)	Scoliosis (HP:0002650)
Spinal rigidity (HP:0003306)	Skeletal dysplasia (HP:0002652)	Sensory impairment (HP:0003474)
Arrhythmia (HP:0011675)	Cardiomyopathy (HP:0001638)	Diabetes mellitus (HP:0000819)
Abnormality of the cerebrum (HP:0002060)	Abnormality of the cerebellum (HP:0001317)	Muscle fibrillation (HP:0010546)
Abnormal muscle fiber alpha dystroglycan (HP:0030112)	Abnormal muscle fiber calpain-3 (HP:0030119)	Abnormal muscle fiber dysferlin (HP:0030113)
Abnormal muscle fiber merosin expression (HP:0030090)	Reduced muscle dystrophin expression (HP:0030098)	Abnormal muscle fiber emerin (HP:0030116)

Limb girdle muscular dystrophy clinical tests (30957)

<table>
<tr><td>Level 3 Title</td><td>Neuromuscular disorders (10994)</td></tr>
<tr><td>Level 4 Title</td><td>Limb girdle muscular dystrophy (11106)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Biopsy refers to muscle biopsy Imaging diagnostics refers to Muscle MRI
Non-imaging diagnostics refers to EMG, Motor nerve conduction studies, Sensory nerve conduction studies
Other enzymes refers to CK
Entries ordered left to right in table

<table><tr><td>Forced vital capacity (30180.2)</td><td>Other enzymes (33123.1)</td><td>General Biopsy (33614.1)</td></tr><tr><td>General Imaging Diagnostics (33633.1)</td><td>General Non-imaging Diagnostics (34838.1)</td><td></td></tr></table></td></tr>
</table>

Skeletal Muscle Channelopathies (15139)

MCP phenotypes (27787)

Level 3 Title	Channelopathies (11097)
Level 4 Title	Skeletal Muscle Channelopathies (15139)
Phenotypes	Entries ordered left to right in table

Periodic paralysis (HP:0003768)	Episodic ataxia (HP:0002131)	Muscle weakness (HP:0001324)
Upper limb muscle weakness (HP:0003484)	Lower limb muscle weakness (HP:0007340)	Bulbar signs (HP:0002483)
Facial hypotonia (HP:0000297)	Weakness of muscles of respiration (HP:0004347)	Prenatal maternal abnormality (HP:0002686)
Myotonia (HP:0002486)	Myotonia with warm-up phenomenon (HP:0003740)	Exercise-induced muscle cramps (HP:0003710)
Cold-induced muscle cramps (HP:0003449)	Myalgia (HP:0003326)	Skeletal muscle hypertrophy (HP:0003712)
Progressive muscle weakness (HP:0003323)	Myokymia (HP:0002411)	Migraine (HP:0002076)
Migraine with aura (HP:0002077)	Migraine without aura (HP:0002083)	Prolonged QT interval (HP:0001657)
EMG abnormality (HP:0003457)	Abnormality of muscle morphology (HP:0011805)	Morphological abnormality of the central nervous system (HP:0002011)
Short stature (HP:0004322)	Abnormal facial shape (HP:0001999)	Abnormality of the lower limb (HP:0002814)
Abnormality of the upper limb (HP:0002817)	Hypertrophy of the lower limb (HP:0010496)	Hypertrophy of the upper limb (HP:0010484)
Generalized limb muscle atrophy (HP:0009055)	Percussion myotonia (HP:0010548)	Reduced tendon reflexes (HP:0001315)
Areflexia (HP:0001284)	Brisk reflexes (HP:0001348)	Gait disturbance (HP:0001288)
Elevated serum creatine phosphokinase (HP:0003236)	Mildly elevated creatine phosphokinase (HP:0008180)	

Skeletal Muscle Channelopathies clinical tests (30959)

Level 3 Title	Channelopathies (11097)
Level 4 Title	Skeletal Muscle Channelopathies (15139)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to muscle biopsy Non-imaging diagnostics refers to ECG, EMG Imaging diagnostics refers to Brain CT or MRI Other enzymes refers to CK Entries ordered left to right in table

Exercise test (30214.1)	General Biopsy (33614.1)	General Non-imaging Diagnostics (34838.1)
General Imaging Diagnostics (33633.1)		

Brain channelopathy (15140)

Brain channelopathy phenotypes (29184)

Level 3 Title	Channelopathies (11097)
Level 4 Title	Brain channelopathy (15140)
Phenotypes	Entries ordered left to right in table

Episodic ataxia (HP:0002131)	Headache (HP:0002315)	Cluster headache (HP:0012199)
Migraine (HP:0002076)	Trigeminal neuralgia (HP:0100661)	Nausea (HP:0002018)
Postural instability (HP:0002172)	Abnormality of eye movement (HP:0000496)	Difficulty walking (HP:0002355)
Abnormality of movement (HP:0100022)	Dystonia (HP:0001332)	Dysarthria (HP:0001260)

Brain Channelopathy clinical tests (30965)

Level 3 Title	Channelopathies (11097)
Level 4 Title	Brain channelopathy (15140)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to Brain MRI Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Sleep disorders (10995)

Kleine-Levin syndrome and other inherited sleep disorders (11108)

Kleine-Levin syndrome phenotypes (29185)

Level 3 Title	Sleep disorders (10995)
Level 4 Title	Kleine-Levin syndrome and other inherited sleep disorders (11108)
Phenotypes	Entries ordered left to right in table

Episodic hypersomnia (HP:0007200)	Abnormality of prenatal development or birth (HP:0001197)	Global developmental delay (HP:0001263)
Behavioral abnormality (HP:0000708)	Polyphagia (HP:0002591)	Hypersexuality (HP:0030214)
Irritability (HP:0000737)	Personality changes (HP:0000751)	Abnormality of the nervous system (HP:0000707)
Aggressive behavior (HP:0000718)	Abnormal fear/anxiety-related behavior (HP:0100852)	EEG abnormality (HP:0002353)
EEG with generalized slow activity (HP:0010845)		

Kleine-Levin syndrome and other inherited sleep disorders clinical tests (30966)

Level 3 Title	Sleep disorders (10995)
Level 4 Title	Kleine-Levin syndrome and other inherited sleep disorders (11108)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to Brain MRI Entries ordered left to right in table

Sleep test (30184.2)	General Imaging Diagnostics (33633.1)

Cerebrovascular disorders (36610)

Moyamoya disease (36611)

Moyamoya disease phenotypes (36652)

Level 3 Title	Cerebrovascular disorders (36610)
Level 4 Title	Moyamoya disease (36611)
Phenotypes	Entries ordered left to right in table

Transient ischemic attack (HP:0002326)	Ischemic stroke (HP:0002140)	Cerebral hemorrhage (HP:0001342)
Chorea (HP:0002072)	Cognitive impairment (HP:0100543)	Seizures (HP:0001250)
Cafe-au-lait spot (HP:0000957)	Hyperpigmented/hypopigmented macules (HP:0007441)	Hemangioma (HP:0001028)
Cutis marmorata (HP:0000965)	Abnormal facial shape (HP:0001999)	Abnormal heart morphology (HP:0001627)
Hypertension (HP:0000822)	Renovascular hypertension (HP:0100817)	Abnormality of the aorta (HP:0001679)
Aortic aneurysm (HP:0004942)	Coarctation of aorta (HP:0001680)	Cataract (HP:0000518)
Bilateral congenital mydriasis (HP:0007932)	Short stature (HP:0004322)	Hypothyroidism (HP:0000821)
Hypopituitarism (HP:0040075)	Hypogonadism (HP:0000135)	

Moyamoya disease clinical tests (36613)

Level 3 Title	Cerebrovascular disorders (36610)
Level 4 Title	Moyamoya disease (36611)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to MRI brain, MRA brain, cerebral angiography and CT brain Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Vein of Galen malformation (42174)

Vein of Galen malformation phenotypes (42179)

Level 3 Title	Cerebrovascular disorders (36610)
Level 4 Title	Vein of Galen malformation (42174)
Phenotypes	Entries ordered left to right in table

Cerebral arteriovenous malformation (HP:0002408)	Intracranial hemorrhage (HP:0002170)	Hydrocephalus (HP:0000238)
High-output congestive heart failure (HP:0001722)	Capillary hemangiomas (HP:0005306)	Diffuse telangiectasia (HP:0007489)

Vein of Galen malformation clinical tests (42180)

Level 3 Title	Cerebrovascular disorders (36610)
Level 4 Title	Vein of Galen malformation (42174)
Clinical Tests	Clinical tests guidance General imaging diagnostics refers to MRI brain, MRA brain, cerebral angiography and CT brain Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Parenchymal brain disorders (36618)

Intracerebral calcification disorders (36619)

Intracerebral calcification disorders phenotypes (36653)

Level 3 Title	Parenchymal brain disorders (36618)
Level 4 Title	Intracerebral calcification disorders (36619)
Phenotypes	Entries ordered left to right in table

Cerebral calcification (HP:0002514)	Basal ganglia calcification (HP:0002135)	Bilateral intracranial calcifications (HP:0005671)
Intracerebral periventricular calcifications (HP:0007229)	Subcortical white matter calcifications (HP:0007346)	Intracranial cystic lesion (HP:0010576)
Cerebral atrophy (HP:0002059)	Abnormal cortical gyration (HP:0002536)	Abnormality of brainstem morphology (HP:0002363)
Cerebellar hypoplasia (HP:0001321)	Cerebellar atrophy (HP:0001272)	Ventriculomegaly (HP:0002119)
Abnormality of the corpus callosum (HP:0001273)	Microcephaly (HP:0000252)	Global developmental delay (HP:0001263)
Developmental regression (HP:0002376)	Intellectual disability (HP:0001249)	Cortical visual impairment (HP:0100704)
Optic atrophy (HP:0000648)	Abnormality of the retina (HP:0000479)	Cataract (HP:0000518)
Microphthalmia (HP:0000568)	Sensorineural hearing impairment (HP:0000407)	Abnormality of the cardiovascular system (HP:0001626)
Cardiomyopathy (HP:0001638)	Recurrent lower respiratory tract infections (HP:0002783)	Skeletal dysplasia (HP:0002652)
Chilblain lesions (HP:0009710)	Abnormality of the endocrine system (HP:0000818)	Spastic diplegia (HP:0001264)
Tetraplegia (HP:0002445)	Peripheral neuropathy (HP:0009830)	Bulbar palsy (HP:0001283)
Parkinsonism (HP:0001300)	Dystonia (HP:0001332)	Chorea (HP:0002072)
Seizures (HP:0001250)	Dementia (HP:0000726)	

Intracerebral calcification disorders clinical tests (36621)

Level 3 Title	Parenchymal brain disorders (36618)
Level 4 Title	Intracerebral calcification disorders (36619)
Clinical Tests	Entries ordered left to right in table General Imaging Diagnostics (33633.1)

White matter disorders (36626)

Inherited white matter disorders (36627)

Inherited white matter disorders phenotypes (36654)

Level 3 Title	White matter disorders (36626)
Level 4 Title	Inherited white matter disorders (36627)
Phenotypes	Entries ordered left to right in table

Diffuse white matter abnormalities (HP:0007204)	Focal white matter lesions (HP:0007042)	Leukoencephalopathy (HP:0002352)
Subcortical white matter calcifications (HP:0007346)	Adrenal insufficiency (HP:0000846)	Growth hormone deficiency (HP:0000824)
Failure to thrive (HP:0001508)	Hypogonadotrophic hypogonadism (HP:0000044)	Abnormal facial shape (HP:0001999)
Coarse facial features (HP:0000280)	Abnormality of the teeth (HP:0000164)	Hyperpigmentation of the skin (HP:0000953)
Angiokeratoma (HP:0001014)	Cherry red spot of the macula (HP:0010729)	Glaucoma (HP:0000501)
Optic atrophy (HP:0000648)	Rod-cone dystrophy (HP:0000510)	Visual impairment (HP:0000505)
Sensorineural hearing impairment (HP:0000407)	Diarrhea (HP:0002014)	Gallbladder dysfunction (HP:0005609)
Abnormality of the liver (HP:0001392)	Hepatosplenomegaly (HP:0001433)	Abnormal joint morphology (HP:0001367)
Abnormality of the skeletal system (HP:0000924)	Myopathy (HP:0003198)	Primary ovarian failure (HP:0001587)
Macrocephaly (HP:0000256)	Microcephaly (HP:0000252)	Cognitive impairment (HP:0100543)
Autism (HP:0000717)	Behavioral abnormality (HP:0000708)	Irritability (HP:0000737)
Severe muscular hypotonia (HP:0006829)	Spasticity (HP:0001257)	Tremor (HP:0001337)
Chorea (HP:0002072)	Dystonia (HP:0001332)	Parkinsonism (HP:0001300)
Spastic paraparesis (HP:0002313)	Ataxia (HP:0001251)	Peripheral neuropathy (HP:0009830)

	Seizures (HP:0001250)	Dysautonomia (HP:0002459)	Developmental regression (HP:0002376)
	CNS hypomyelination (HP:0003429)	Abnormal myelination (HP:0012447)	Abnormality of the basal ganglia (HP:0002134)
	Cerebellar atrophy (HP:0001272)	Cerebral atrophy (HP:0002059)	Cerebellar calcifications (HP:0007352)
	Cerebral calcification (HP:0002514)	Intracranial cystic lesion (HP:0010576)	Abnormality of the spinal cord (HP:0002143)
	Premature birth (HP:0001622)	Abnormal delivery (HP:0001787)	Hypothyroidism (HP:0000821)
	Ichthyosis (HP:0008064)	Cataract (HP:0000518)	

Inherited white matter disorders clinical tests (36629)

Level 3 Title	White matter disorders (36626)
Level 4 Title	Inherited white matter disorders (36627)
Clinical Tests	Entries ordered left to right in table APGAR score (30207.1)

Ophthalmological disorders (10996)

Anterior segment abnormalities (10997)

Corneal abnormalities (11110)

Corneal abnormalities phenotypes (29187)

Level 3 Title	Anterior segment abnormalities (10997)
Level 4 Title	Corneal abnormalities (11110)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Slow decrease in visual acuity (HP:0007924)	Corneal dystrophy (HP:0001131)	Lattice corneal dystrophy (HP:0001149)
Granular corneal dystrophy (HP:0007802)	Band-shaped corneal dystrophy (HP:0007709)	Central corneal dystrophy (HP:0007881)
Congenital corneal dystrophy (HP:0008005)	Crystalline corneal dystrophy (HP:0007760)	Juvenile epithelial corneal dystrophy (HP:0007755)
Map-dot-fingerprint corneal dystrophy (HP:0007690)	Marginal corneal dystrophy (HP:0007880)	Mosaic central corneal dystrophy (HP:0100690)
Mosaic corneal dystrophy (HP:0007836)	Nodular corneal dystrophy (HP:0007827)	Polymorphous posterior corneal dystrophy (HP:0007915)
Punctate corneal dystrophy (HP:0007809)	Speckled corneal dystrophy (HP:0007962)	Ocular pain (HP:0200026)
Corneal erosion (HP:0200020)	Astigmatism (HP:0000483)	

Corneal Abnormality clinical tests (30971)

Level 3 Title	Anterior segment abnormalities (10997)
Level 4 Title	Corneal abnormalities (11110)
Clinical Tests	Entries ordered left to right in table

Ocular Malformation Metrics (30260.2)	Visual Acuity (30970.2)

Glaucoma (developmental) (11111)

Glaucoma (developmental) phenotypes (29188)

Level 3 Title	Anterior segment abnormalities (10997)
Level 4 Title	Glaucoma (developmental) (11111)
Phenotypes	Entries ordered left to right in table

Glaucoma (HP:0000501)	Primary open angle glaucoma (HP:0012108)	Primary angle closure glaucoma (HP:0012109)
Congenital glaucoma (HP:0001087)	Abnormality of the anterior segment of the globe (HP:0004328)	Posterior embryotoxon (HP:0000627)
Central posterior corneal opacity (HP:0008511)	Microcornea (HP:0000482)	Megalocornea (HP:0000485)
Abnormality of the iris (HP:0000525)	Polycoria (HP:0011500)	Ectopia pupillae (HP:0009918)
Hypoplasia of the iris (HP:0007676)	Cataract (HP:0000518)	Increased cup-to-disc ratio (HP:0012796)
Microphthalmia (HP:0000568)		

Glaucoma clinical tests (30972)

Level 3 Title	Anterior segment abnormalities (10997)
Level 4 Title	Glaucoma (developmental) (11111)
Clinical Tests	Entries ordered left to right in table

Ocular Pressure (30265.2)	Ocular Malformation Metrics (30260.2)	Visual Acuity (30970.2)

Cataracts (11112)

Cataracts phenotypes (28641)

Level 3 Title	Anterior segment abnormalities (10997)
Level 4 Title	Cataracts (11112)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Cataract (HP:0000518)	Progressive cataract (HP:0007834)
Total cataract (HP:0010700)	Nuclear cataract (HP:0100018)	Cortical cataract (HP:0100019)
Membranous cataract (HP:0010922)	Polar cataract (HP:0010696)	Subcapsular cataract (HP:0000523)
Lamellar cataract (HP:0007971)	Pulverulent cataract (HP:0010693)	Microphthalmia (HP:0000568)
Microcornea (HP:0000482)	Microphakia (HP:0012376)	Abnormality of the anterior segment of the globe (HP:0004328)
Anterior segment dysgenesis (HP:0007700)	Abnormality of the posterior segment of the globe (HP:0004329)	Remnants of the hyaloid vascular system (HP:0007968)
Retinal degeneration (HP:0000546)	Maternal teratogenic exposure (HP:0011438)	

Cataracts clinical tests (30973)

Level 3 Title	Anterior segment abnormalities (10997)
Level 4 Title	Cataracts (11112)
Clinical Tests	Entries ordered left to right in table

TORCH screen (30122.2)	Ocular Pressure (30265.2)	Ocular Malformation Metrics (30260.2)
Visual Acuity (30970.2)	Urine metabolic tests (33189.1)	

Posterior segment abnormalities (10998)

Inherited optic neuropathies (11114)

Inherited optic neuropathies phenotypes (29190)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Inherited optic neuropathies (11114)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Central scotoma (HP:0000603)	Constriction of peripheral visual field (HP:0001133)	Optic neuropathy (HP:0001138)
Abnormality of color vision (HP:0000551)	Cataract (HP:0000518)	Nystagmus (HP:0000639)
Hearing impairment (HP:0000365)	Ataxia (HP:0001251)	Diabetes mellitus (HP:0000819)
Diabetes insipidus (HP:0000873)		

Inherited optic neuropathies clinical tests (30975)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Inherited optic neuropathies (11114)
Clinical Tests	Entries ordered left to right in table

Colour Plate Test (30269.2)	Visual Acuity (30970.2)

Rod-cone dystrophy (29268)

Retinal dystrophies phenotypes (29189)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Rod-cone dystrophy (29268)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Central scotoma (HP:0000603)	Constriction of peripheral visual field (HP:0001133)	Abnormality of color vision (HP:0000551)
Reduced visual acuity (HP:0007663)	Retinal dystrophy (HP:0000556)	Cone/cone-rod dystrophy (HP:0000548)
Abnormal light-adapted electroretinogram (HP:0008275)	Rod-cone dystrophy (HP:0000510)	Abnormality of retinal pigmentation (HP:0007703)
Macular dystrophy (HP:0007754)	Macular atrophy (HP:0007401)	Hypoplasia of the fovea (HP:0007750)
Abnormality of the retinal vasculature (HP:0008046)	Retinal exudate (HP:0001147)	

Retinal dystrophy clinical tests (30974)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Rod-cone dystrophy (29268)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography Entries ordered left to right in table

Colour Plate Test (30269.2)	Electroretinogram (30291.2)	Visual Field (30264.2)
Refraction Error (30978.2)	Dark Adaptation Test (30276.2)	Electro-oculogram (30271.2)
General Imaging Diagnostics (33633.1)		

Rod Dysfunction Syndrome (29269)

Retinal dystrophies phenotypes (29189)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Rod Dysfunction Syndrome (29269)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Central scotoma (HP:0000603)	Constriction of peripheral visual field (HP:0001133)	Abnormality of color vision (HP:0000551)
Reduced visual acuity (HP:0007663)	Retinal dystrophy (HP:0000556)	Cone/cone-rod dystrophy (HP:0000548)
Abnormal light-adapted electroretinogram (HP:0008275)	Rod-cone dystrophy (HP:0000510)	Abnormality of retinal pigmentation (HP:0007703)
Macular dystrophy (HP:0007754)	Macular atrophy (HP:0007401)	Hypoplasia of the fovea (HP:0007750)
Abnormality of the retinal vasculature (HP:0008046)	Retinal exudate (HP:0001147)	

Retinal dystrophy clinical tests (30974)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Rod Dysfunction Syndrome (29269)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography Entries ordered left to right in table

Colour Plate Test (30269.2)	Electroretinogram (30291.2)	Visual Field (30264.2)
Refraction Error (30978.2)	Dark Adaptation Test (30276.2)	Electro-oculogram (30271.2)
General Imaging Diagnostics (33633.1)		

Cone Dysfunction Syndrome (29270)

Retinal dystrophies phenotypes (29189)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Cone Dysfunction Syndrome (29270)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Central scotoma (HP:0000603)	Constriction of peripheral visual field (HP:0001133)	Abnormality of color vision (HP:0000551)
Reduced visual acuity (HP:0007663)	Retinal dystrophy (HP:0000556)	Cone/cone-rod dystrophy (HP:0000548)
Abnormal light-adapted electroretinogram (HP:0008275)	Rod-cone dystrophy (HP:0000510)	Abnormality of retinal pigmentation (HP:0007703)
Macular dystrophy (HP:0007754)	Macular atrophy (HP:0007401)	Hypoplasia of the fovea (HP:0007750)
Abnormality of the retinal vasculature (HP:0008046)	Retinal exudate (HP:0001147)	

Retinal dystrophy clinical tests (30974)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Cone Dysfunction Syndrome (29270)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography Entries ordered left to right in table

Colour Plate Test (30269.2)	Electroretinogram (30291.2)	Visual Field (30264.2)
Refraction Error (30978.2)	Dark Adaptation Test (30276.2)	Electro-oculogram (30271.2)
General Imaging Diagnostics (33633.1)		

Inherited macular dystrophy (29271)

Retinal dystrophies phenotypes (29189)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Inherited macular dystrophy (29271)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Central scotoma (HP:0000603)	Constriction of peripheral visual field (HP:0001133)	Abnormality of color vision (HP:0000551)
Reduced visual acuity (HP:0007663)	Retinal dystrophy (HP:0000556)	Cone/cone-rod dystrophy (HP:0000548)
Abnormal light-adapted electroretinogram (HP:0008275)	Rod-cone dystrophy (HP:0000510)	Abnormality of retinal pigmentation (HP:0007703)
Macular dystrophy (HP:0007754)	Macular atrophy (HP:0007401)	Hypoplasia of the fovea (HP:0007750)
Abnormality of the retinal vasculature (HP:0008046)	Retinal exudate (HP:0001147)	

Retinal dystrophy clinical tests (30974)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Inherited macular dystrophy (29271)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography Entries ordered left to right in table

Colour Plate Test (30269.2)	Electroretinogram (30291.2)	Visual Field (30264.2)
Refraction Error (30978.2)	Dark Adaptation Test (30276.2)	Electro-oculogram (30271.2)
General Imaging Diagnostics (33633.1)		

Leber Congenital Amaurosis or Early-Onset Severe Retinal Dystrophy (29272)

Retinal dystrophies phenotypes (29189)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Leber Congenital Amaurosis or Early-Onset Severe Retinal Dystrophy (29272)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Central scotoma (HP:0000603)	Constriction of peripheral visual field (HP:0001133)	Abnormality of color vision (HP:0000551)
Reduced visual acuity (HP:0007663)	Retinal dystrophy (HP:0000556)	Cone/cone-rod dystrophy (HP:0000548)
Abnormal light-adapted electroretinogram (HP:0008275)	Rod-cone dystrophy (HP:0000510)	Abnormality of retinal pigmentation (HP:0007703)
Macular dystrophy (HP:0007754)	Macular atrophy (HP:0007401)	Hypoplasia of the fovea (HP:0007750)
Abnormality of the retinal vasculature (HP:0008046)	Retinal exudate (HP:0001147)	

Retinal dystrophy clinical tests (30974)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Leber Congenital Amaurosis or Early-Onset Severe Retinal Dystrophy (29272)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography Entries ordered left to right in table

Colour Plate Test (30269.2)	Electroretinogram (30291.2)	Visual Field (30264.2)
Refraction Error (30978.2)	Dark Adaptation Test (30276.2)	Electro-oculogram (30271.2)
General Imaging Diagnostics (33633.1)		

Developmental macular and foveal dystrophy (29273)

Retinal dystrophies phenotypes (29189)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Developmental macular and foveal dystrophy (29273)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Central scotoma (HP:0000603)	Constriction of peripheral visual field (HP:0001133)	Abnormality of color vision (HP:0000551)
Reduced visual acuity (HP:0007663)	Retinal dystrophy (HP:0000556)	Cone/cone-rod dystrophy (HP:0000548)
Abnormal light-adapted electroretinogram (HP:0008275)	Rod-cone dystrophy (HP:0000510)	Abnormality of retinal pigmentation (HP:0007703)
Macular dystrophy (HP:0007754)	Macular atrophy (HP:0007401)	Hypoplasia of the fovea (HP:0007750)
Abnormality of the retinal vasculature (HP:0008046)	Retinal exudate (HP:0001147)	

Retinal dystrophy clinical tests (30974)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Developmental macular and foveal dystrophy (29273)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography Entries ordered left to right in table

Colour Plate Test (30269.2)	Electroretinogram (30291.2)	Visual Field (30264.2)
Refraction Error (30978.2)	Dark Adaptation Test (30276.2)	Electro-oculogram (30271.2)
General Imaging Diagnostics (33633.1)		

Familial exudative vitreoretinopathy (41900)

Retinal dystrophies phenotypes (29189)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Familial exudative vitreoretinopathy (41900)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)
Central scotoma (HP:0000603)	Constriction of peripheral visual field (HP:0001133)	Abnormality of color vision (HP:0000551)
Reduced visual acuity (HP:0007663)	Retinal dystrophy (HP:0000556)	Cone/cone-rod dystrophy (HP:0000548)
Abnormal light-adapted electroretinogram (HP:0008275)	Rod-cone dystrophy (HP:0000510)	Abnormality of retinal pigmentation (HP:0007703)
Macular dystrophy (HP:0007754)	Macular atrophy (HP:0007401)	Hypoplasia of the fovea (HP:0007750)
Abnormality of the retinal vasculature (HP:0008046)	Retinal exudate (HP:0001147)	

Retinal dystrophy clinical tests (30974)

Level 3 Title	Posterior segment abnormalities (10998)
Level 4 Title	Familial exudative vitreoretinopathy (41900)
Clinical Tests	Clinical test guidance: Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography Entries ordered left to right in table

Colour Plate Test (30269.2)	Electroretinogram (30291.2)	Visual Field (30264.2)
Refraction Error (30978.2)	Dark Adaptation Test (30276.2)	Electro-oculogram (30271.2)
General Imaging Diagnostics (33633.1)		

Anophthalmia or microphthamia (11115)

Microphthalmia phenotypes (27788)

Level 3 Title	Ocular malformations (10999)
Level 4 Title	Anophthalmia or microphthamia (11115)
Phenotypes	Entries ordered left to right in table

Microphthalmia (HP:0000568)	Anophthalmia (HP:0000528)	Visual impairment (HP:0000505)
Iris coloboma (HP:0000612)	Retinal coloboma (HP:0000480)	Chorioretinal coloboma (HP:0000567)
Optic nerve coloboma (HP:0000588)	Abnormality of the anterior segment of the globe (HP:0004328)	Anterior segment dysgenesis (HP:0007700)
Aplasia/Hypoplasia of the lens (HP:0008063)	Sclerocornea (HP:0000647)	Central opacification of the cornea (HP:0011493)
Cataract (HP:0000518)	Total cataract (HP:0010700)	Nuclear cataract (HP:0100018)
Cortical cataract (HP:0100019)	Membranous cataract (HP:0010922)	Polar cataract (HP:0010696)
Subcapsular cataract (HP:0000523)	Lamellar cataract (HP:0007971)	Pulverulent cataract (HP:0010693)
Abnormality of the posterior segment of the globe (HP:0004329)	Abnormality of the fovea (HP:0000493)	Aplasia/Hypoplasia of the optic nerve (HP:0008058)
Optic nerve dysplasia (HP:0001093)	Retinal dysplasia (HP:0007973)	Remnants of the hyaloid vascular system (HP:0007968)
Maternal fever in pregnancy (HP:0030244)		

Ocular malformation clinical tests (30979)

<table>
<tr><td>Level 3 Title</td><td>Ocular malformations (10999)</td></tr>
<tr><td>Level 4 Title</td><td>Anophthalmia or microphthamia (11115)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography and medical photography of the face and/or other body parts if relevant
Entries ordered left to right in table
<table><tr><td>Ocular Pressure (30265.2)</td><td>Ocular Malformation Metrics (30260.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr><tr><td>Visual Acuity (30970.2)</td><td></td><td></td></tr></table></td></tr>
</table>

Ocular coloboma (15141)

Ocular coloboma phenotypes (29191)

Level 3 Title	Ocular malformations (10999)
Level 4 Title	Ocular coloboma (15141)
Phenotypes	Entries ordered left to right in table

Visual impairment (HP:0000505)	Iris coloboma (HP:0000612)	Chorioretinal coloboma (HP:0000567)
Retinal coloboma (HP:0000480)	Optic nerve coloboma (HP:0000588)	Cataract (HP:0000518)
Total cataract (HP:0010700)	Nuclear cataract (HP:0100018)	Cortical cataract (HP:0100019)
Membranous cataract (HP:0010922)	Polar cataract (HP:0010696)	Subcapsular cataract (HP:0000523)
Lamellar cataract (HP:0007971)	Pulverulent cataract (HP:0010693)	Anterior segment dysgenesis (HP:0007700)
Central opacification of the cornea (HP:0011493)	Sclerocornea (HP:0000647)	Aplasia/Hypoplasia of the lens (HP:0008063)
Abnormality of the fovea (HP:0000493)	Aplasia/Hypoplasia of the optic nerve (HP:0008058)	Optic nerve dysplasia (HP:0001093)
Remnants of the hyaloid vascular system (HP:0007968)	Retinal dysplasia (HP:0007973)	Ptosis (HP:0000508)
Abnormality of the lacrimal duct (HP:0011481)	Strabismus (HP:0000486)	Abnormality of the outer ear (HP:0000356)
Preauricular pit (HP:0004467)	Preauricular skin tag (HP:0000384)	Abnormality of the inner ear (HP:0000359)
Sensorineural hearing impairment (HP:0000407)	Conductive hearing impairment (HP:0000405)	Abnormality of the kidney (HP:0000077)
Horseshoe kidney (HP:0000085)	Renal cyst (HP:0000107)	Abnormality of the ureter (HP:0000069)
Abnormality of the urethra (HP:0000795)	Atrial septal defect (HP:0001631)	Ventricular septal defect (HP:0001629)
Complete atrioventricular canal defect (HP:0001674)	Tetralogy of Fallot (HP:0001636)	Coarctation of aorta (HP:0001680)
Seizures (HP:0001250)	Facial palsy (HP:0010628)	Focal dermal aplasia/hypoplasia (HP:0007510)
Choanal atresia (HP:0000453)	Hypoplasia of teeth (HP:0000685)	Nail dysplasia (HP:0002164)
Oral cleft (HP:0000202)		

Ocular malformation clinical tests (30979)

<table>
<tr><td>Level 3 Title</td><td>Ocular malformations (10999)</td></tr>
<tr><td>Level 4 Title</td><td>Ocular coloboma (15141)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to medical photography of the fundus and/or Indocyanin green [ICG] angiography, fluorescein angiography and medical photography of the face and/or other body parts if relevant
Entries ordered left to right in table
<table><tr><td>Ocular Pressure (30265.2)</td><td>Ocular Malformation Metrics (30260.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr><tr><td>Visual Acuity (30970.2)</td><td></td><td></td></tr></table></td></tr>
</table>

Ocular movement disorders (33350)

Infantile nystagmus (33662)

Infantile nystagmus phenotypes (33664)

Level 3 Title	Ocular movement disorders (33350)
Level 4 Title	Infantile nystagmus (33662)
Phenotypes	Entries ordered left to right in table

Nystagmus (HP:0000639)	Visual impairment (HP:0000505)	Abnormality of refraction (HP:0000539)
Myopia (HP:0000545)	Hypermetropia (HP:0000540)	Astigmatism (HP:0000483)
Amblyopia (HP:0000646)	Strabismus (HP:0000486)	Congenital nystagmus (HP:0006934)
Pendular nystagmus (HP:0012043)	Horizontal opticokinetic nystagmus (HP:0008026)	

Infantile nystagmus clinical tests (33665)

Level 3 Title	Ocular movement disorders (33350)
Level 4 Title	Infantile nystagmus (33662)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to MRI brain. Non-imaging diagnostics refers to ERG and VEP Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	Colour Plate Test (30269.2)	General Non-imaging Diagnostics (34838.1)
Visual Acuity (30970.2)		

Psychiatric disorders (71735)

Feeding and eating disorders (71737)

Severe familial anorexia (29278)

Severe familial anorexia phenotypes (29173)

Level 3 Title	Feeding and eating disorders (71737)
Level 4 Title	Severe familial anorexia (29278)
Phenotypes	Entries ordered left to right in table

Anorexia (HP:0002039)	Abnormality of dental enamel (HP:0000682)	Abnormality of dentin (HP:0010299)
Abnormality of parotid gland (HP:0000197)	Enlargement of parotid gland (HP:0011801)	Temperature instability (HP:0005968)
Constipation (HP:0002019)	Acute constipation (HP:0012451)	Chronic constipation (HP:0012450)
Cholelithiasis (HP:0001081)	Pancreatitis (HP:0001733)	Hypotension (HP:0002615)
Orthostatic hypotension (HP:0001278)	Postural hypotension with compensatory tachycardia (HP:0005307)	Prolonged QT interval (HP:0001657)
Sinus bradycardia (HP:0001688)	Paroxysmal vertigo (HP:0010532)	Delayed puberty (HP:0000823)
Abnormality of circulating leptin level (HP:0004361)	Decreased serum leptin (HP:0003292)	Amenorrhea (HP:0000141)
Primary amenorrhea (HP:0000786)	Secondary amenorrhea (HP:0000869)	Infertility (HP:0000789)
Female infertility (HP:0008222)	Acrocyanosis (HP:0001063)	Brittle hair (HP:0002299)
Congenital, generalized hypertrichosis (HP:0004540)	Dry skin (HP:0000958)	Erythroderma (HP:0001019)
Exercise-induced muscle fatigue (HP:0009020)	Fatigable weakness (HP:0003473)	Generalized muscle weakness (HP:0003324)
Abnormal fear/anxiety-related behavior (HP:0100852)	Abnormality of taste sensation (HP:0000223)	Abnormality of the sense of smell (HP:0004408)

	Abnormality of the nervous system (HP:0000707)	Specific learning disability (HP:0001328)	Attention deficit hyperactivity disorder (HP:0007018)
	Anxiety (HP:0000739)	Bipolar affective disorder (HP:0007302)	Depression (HP:0000716)
	Obsessive-compulsive behavior (HP:0000722)	Psychosis (HP:0000709)	Schizophrenia (HP:0100753)
	Lack of insight (HP:0000757)	Bulimia (HP:0100739)	Headache (HP:0002315)
	Fatigue (HP:0012378)	Migraine (HP:0002076)	Chest pain (HP:0100749)
	Growth delay (HP:0001510)	Osteopenia (HP:0000938)	Osteoporosis (HP:0000939)

Schizophrenia and other psychotic disorders (71736)

Schizophrenia plus additional features (71740)

Schizophrenia plus additional features phenotypes (71768)

Level 3 Title	Schizophrenia and other psychotic disorders (71736)
Level 4 Title	Schizophrenia plus additional features (71740)
Phenotypes	Entries ordered left to right in table

Schizophrenia (HP:0100753)	Hallucinations (HP:0000738)	Delusions (HP:0000746)
Delayed speech and language development (HP:0000750)	Intellectual disability (HP:0001249)	Behavioral abnormality (HP:0000708)
Autistic behavior (HP:0000729)	Bipolar affective disorder (HP:0007302)	Microcephaly (HP:0000252)
Abnormality of the nervous system (HP:0000707)	Seizures (HP:0001250)	Narcolepsy (HP:0030050)
Cardiomyopathy (HP:0001638)	Autoimmunity (HP:0002960)	Abnormality of the skeletal system (HP:0000924)
Hearing impairment (HP:0000365)	Abnormal facial shape (HP:0001999)	

Schizophrenia plus additional features clinical tests (71743)

Level 3 Title	Schizophrenia and other psychotic disorders (71736)
Level 4 Title	Schizophrenia plus additional features (71740)
Clinical Tests	Clinical test guidance: General Imaging Diagnostics refers to MRI Brain Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Renal and urinary tract disorders (11000)

Syndromes with prominent renal abnormalities (11001)

Proteinuric renal disease (30732)

Renal phenotypes (30761)

Level 3 Title	Syndromes with prominent renal abnormalities (11001)
Level 4 Title	Proteinuric renal disease (30732)
Phenotypes	Entries ordered left to right in table

Microscopic hematuria (HP:0002907)	Macroscopic hematuria (HP:0012587)	Edema (HP:0000969)
Proteinuria (HP:0000093)	Thin glomerular basement membrane (HP:0012577)	Thickening of the glomerular basement membrane (HP:0004722)
Global developmental delay (HP:0001263)	Abnormality of the eye (HP:0000478)	Abnormality of the outer ear (HP:0000356)
Hearing impairment (HP:0000365)	Anosmia (HP:0000458)	Abnormal facial shape (HP:0001999)
Branchial anomaly (HP:0009794)	Morphological abnormality of the central nervous system (HP:0002011)	Situs inversus totalis (HP:0001696)
Abnormality of the respiratory system (HP:0002086)	Abnormal heart morphology (HP:0001627)	Abnormality of the abdominal organs (HP:0002012)
Cryptorchidism (HP:0000028)	Abnormality of the immune system (HP:0002715)	Abnormality of skeletal morphology (HP:0011842)
Abnormality of the hair (HP:0001595)	Abnormality of the nail (HP:0001597)	Abnormality of the skin (HP:0000951)
Abnormality of the endocrine system (HP:0000818)	Diabetes mellitus (HP:0000819)	Hypogonadism (HP:0000135)
Ambiguous genitalia (HP:0000062)	Abnormality of the female genitalia (HP:0010460)	Abnormality of the male genitalia (HP:0010461)

Renal clinical tests (30982)

<table>
<tr><td>Level 3 Title</td><td>Syndromes with prominent renal abnormalities (11001)</td></tr>
<tr><td>Level 4 Title</td><td>Proteinuric renal disease (30732)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to renal imaging
Entries ordered left to right in table

<table><tr><td>Blood pressure (30245.1)</td><td>Full Blood Count (30318.2)</td><td>Autoantibodies (30112.2)</td></tr><tr><td>Renal Biopsy (30153.3)</td><td>Age at ESRD (30199.2)</td><td>Renal biochemistry (30355.2)</td></tr><tr><td>Extended renal biochemistry (33152.1)</td><td>Bone profile (30317.2)</td><td>Glucose (33153.1)</td></tr><tr><td>Renin and aldosterone (33166.1)</td><td>Inflammatory markers (33156.1)</td><td>Lipids (33158.1)</td></tr><tr><td>Complement (33146.1)</td><td>Urine electrolytes (33187.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr><tr><td>Metabolic biochemistry (33160.2)</td><td>Urine Protein (33190.1)</td><td>Vitamin B12 (33172.1)</td></tr></table></td></tr>
</table>

Familial haematuria (30733)

Renal phenotypes (30761)

Level 3 Title	Syndromes with prominent renal abnormalities (11001)
Level 4 Title	Familial haematuria (30733)
Phenotypes	Entries ordered left to right in table

Microscopic hematuria (HP:0002907)	Macroscopic hematuria (HP:0012587)	Edema (HP:0000969)
Proteinuria (HP:0000093)	Thin glomerular basement membrane (HP:0012577)	Thickening of the glomerular basement membrane (HP:0004722)
Global developmental delay (HP:0001263)	Abnormality of the eye (HP:0000478)	Abnormality of the outer ear (HP:0000356)
Hearing impairment (HP:0000365)	Anosmia (HP:0000458)	Abnormal facial shape (HP:0001999)
Branchial anomaly (HP:0009794)	Morphological abnormality of the central nervous system (HP:0002011)	Situs inversus totalis (HP:0001696)
Abnormality of the respiratory system (HP:0002086)	Abnormal heart morphology (HP:0001627)	Abnormality of the abdominal organs (HP:0002012)
Cryptorchidism (HP:0000028)	Abnormality of the immune system (HP:0002715)	Abnormality of skeletal morphology (HP:0011842)
Abnormality of the hair (HP:0001595)	Abnormality of the nail (HP:0001597)	Abnormality of the skin (HP:0000951)
Abnormality of the endocrine system (HP:0000818)	Diabetes mellitus (HP:0000819)	Hypogonadism (HP:0000135)
Ambiguous genitalia (HP:0000062)	Abnormality of the female genitalia (HP:0010460)	Abnormality of the male genitalia (HP:0010461)

Renal clinical tests (30982)

Level 3 Title	Syndromes with prominent renal abnormalities (11001)
Level 4 Title	Familial haematuria (30733)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to renal imaging Entries ordered left to right in table

Blood pressure (30245.1)	Full Blood Count (30318.2)	Autoantibodies (30112.2)
Renal Biopsy (30153.3)	Age at ESRD (30199.2)	Renal biochemistry (30355.2)
Extended renal biochemistry (33152.1)	Bone profile (30317.2)	Glucose (33153.1)
Renin and aldosterone (33166.1)	Inflammatory markers (33156.1)	Lipids (33158.1)
Complement (33146.1)	Urine electrolytes (33187.2)	General Imaging Diagnostics (33633.1)
Metabolic biochemistry (33160.2)	Urine Protein (33190.1)	Vitamin B12 (33172.1)

Atypical haemolytic uraemic syndrome (33489)

Atypical haemolytic uraemic syndrome phenotypes (33669)

Level 3 Title	Syndromes with prominent renal abnormalities (11001)
Level 4 Title	Atypical haemolytic uraemic syndrome (33489)
Phenotypes	Entries ordered left to right in table

Microangiopathic hemolytic anemia (HP:0001937)	Thrombocytopenia (HP:0001873)	Acute kidney injury (HP:0001919)
Chronic kidney disease (HP:0012622)	Seizures (HP:0001250)	Stroke (HP:0001297)
Transient ischemic attack (HP:0002326)	Confusion (HP:0001289)	Angina pectoris (HP:0001681)
Gastrointestinal hemorrhage (HP:0002239)	Hypertension (HP:0000822)	Hemolytic-uremic syndrome (HP:0005575)
Mesangial hypercellularity (HP:0012574)	Thickening of the glomerular basement membrane (HP:0004722)	Thin glomerular basement membrane (HP:0012577)

Atypical haemolytic uraemic syndrome clinical tests (33670)

Level 3 Title	Syndromes with prominent renal abnormalities (11001)
Level 4 Title	Atypical haemolytic uraemic syndrome (33489)
Clinical Tests	Entries ordered left to right in table

Age at ESRD (30199.2)	Full Blood Count (30318.2)	Extended haematology investigations (33151.1)
Renal biochemistry (30355.2)	Liver biochemistry (30328.2)	Inflammatory markers (33156.1)
Complement (33146.1)	Urine dip - standard (31317.2)	Urine electrolytes (33187.2)
General Biopsy (33614.1)	Renal Biopsy (30153.3)	Blood pressure (30245.1)
Urine Protein (33190.1)		

Primary membranoproliferative glomerulonephritis (55481)

Primary membranoproliferative glomerulonephritis phenotypes (68117)

Level 3 Title	Syndromes with prominent renal abnormalities (11001)
Level 4 Title	Primary membranoproliferative glomerulonephritis (55481)
Phenotypes	Entries ordered left to right in table

Membranoproliferative glomerulonephritis (HP:0000793)	Nephrotic syndrome (HP:0000100)	Stage 5 chronic kidney disease (HP:0003774)
Glomerular subendothelial electron-dense deposits (HP:0004746)	Glomerular C3 deposition (HP:0012576)	Hypertension (HP:0000822)
Proteinuria (HP:0000093)	Hematuria (HP:0000790)	Edema (HP:0000969)
Hypoalbuminemia (HP:0003073)	Decreased urine output (HP:0011037)	Drusen (HP:0011510)
Lipodystrophy (HP:0009125)	Decreased serum complement C3 (HP:0005421)	Decreased serum complement C4 (HP:0045042)
C3 nephritic factor positivity (HP:0030888)	Chronic hepatitis (HP:0200123)	Respiratory tract infection (HP:0011947)
Recurrent viral infections (HP:0004429)	Recurrent fungal infections (HP:0002841)	Autoimmunity (HP:0002960)
Abnormal immunoglobulin level (HP:0010701)		

Primary membranoproliferative glomerulonephritis clinical tests (55482)

Level 3 Title	Syndromes with prominent renal abnormalities (11001)
Level 4 Title	Primary membranoproliferative glomerulonephritis (55481)
Clinical Tests	NB Clinical test guidance: Urine Protein refers to Urine Protein Creatinine ratio Extended Renal Biochemistry refers to Creatinine (clearance) and eGFR Entries ordered left to right in table

Urine Protein (33190.1)	Extended renal biochemistry (33152.1)

Familial IgA nephropathy and IgA vasculitis (82147)

Familial IgA nephropathy and IgA vasculitis phenotypes (82290)

Level 3 Title	Syndromes with prominent renal abnormalities (11001)
Level 4 Title	Familial IgA nephropathy and IgA vasculitis (82147)
Phenotypes	Entries ordered left to right in table

IgA deposition in the glomerulus (HP:0000794)	Vasculitis (HP:0002633)	Macroscopic hematuria (HP:0012587)
Microscopic hematuria (HP:0002907)	Proteinuria (HP:0000093)	Nephrotic syndrome (HP:0000100)
Glomerulonephritis (HP:0000099)	Acute kidney injury (HP:0001919)	Chronic kidney disease (HP:0012622)

Structural renal and urinary tract disease (11003)

Cystic kidney disease (11120)

Cystic kidney disease phenotypes (29194)

Level 3 Title	Structural renal and urinary tract disease (11003)
Level 4 Title	Cystic kidney disease (11120)
Phenotypes	Entries ordered left to right in table

Multiple renal cysts (HP:0005562)	Multiple small medullary renal cysts (HP:0008659)	Renal cortical cysts (HP:0000803)
Multiple glomerular cysts (HP:0100611)	Tubulointerstitial fibrosis (HP:0005576)	Tubular atrophy (HP:0000092)
Tubulointerstitial nephritis (HP:0001970)	Enlarged kidney (HP:0000105)	Renal atrophy (HP:0012585)
Nephronophthisis (HP:0000090)	Pancreatic cysts (HP:0001737)	Hepatic cysts (HP:0001407)
Cerebral aneurysm (HP:0004944)	Gout (HP:0001997)	Hypertension (HP:0000822)
Obesity (HP:0001513)	Polydactyly (HP:0010442)	Global developmental delay (HP:0001263)
Morphological abnormality of the central nervous system (HP:0002011)	Abnormality of the eye (HP:0000478)	Anosmia (HP:0000458)
Hypogonadism (HP:0000135)	Abnormal facial shape (HP:0001999)	Branchial anomaly (HP:0009794)
Hearing impairment (HP:0000365)	Abnormality of the outer ear (HP:0000356)	Abnormality of the respiratory system (HP:0002086)
Abnormal heart morphology (HP:0001627)	Diabetes mellitus (HP:0000819)	Abnormality of skeletal morphology (HP:0011842)
Abnormality of the immune system (HP:0002715)	Abnormality of the hair (HP:0001595)	Abnormality of the nail (HP:0001597)
Abnormality of the skin (HP:0000951)	Abnormal genital system morphology (HP:0012243)	

Cystic kidney disease clinical tests (30983)

Level 3 Title	Structural renal and urinary tract disease (11003)
Level 4 Title	Cystic kidney disease (11120)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to renal and liver imaging Entries ordered left to right in table

Blood pressure (30245.1)	Age at ESRD (30199.2)	Kidney Imaging (29807.2)
Liver Imaging (29823.2)	Full Blood Count (30318.2)	Renal Biopsy (30153.3)
Liver biochemistry (30328.2)	Renal biochemistry (30355.2)	Lipids (33158.1)
Renin and aldosterone (33166.1)	Urine electrolytes (33187.2)	Urine dip - standard (31317.2)
General Imaging Diagnostics (33633.1)	Metabolic biochemistry (33160.2)	Urine Protein (33190.1)

Congenital Anomaly of the Kidneys and Urinary Tract (CAKUT) (29277)

CAKUT phenotypes (29193)

Level 3 Title	Structural renal and urinary tract disease (11003)
Level 4 Title	Congenital Anomaly of the Kidneys and Urinary Tract (CAKUT) (29277)
Phenotypes	Entries ordered left to right in table

Renal agenesis (HP:0000104)	Renal duplication (HP:0000075)	Ectopic kidney (HP:0000086)
Horseshoe kidney (HP:0000085)	Cystic renal dysplasia (HP:0000800)	Hydronephrosis (HP:0000126)
Ureteral stenosis (HP:0000071)	Ureteral duplication (HP:0000073)	Hydroureter (HP:0000072)
Vesicoureteral reflux (HP:0000076)	Abnormality of the bladder (HP:0000014)	Megacystis (HP:0000021)
Bladder exstrophy (HP:0002836)	Congenital posterior urethral valve (HP:0010957)	Abnormality of the male genitalia (HP:0010461)
Abnormality of the female genitalia (HP:0010460)	Global developmental delay (HP:0001263)	Abnormal facial shape (HP:0001999)
Anosmia (HP:0000458)	Abnormality of the eye (HP:0000478)	Abnormality of the outer ear (HP:0000356)
Hearing impairment (HP:0000365)	Branchial anomaly (HP:0009794)	Abnormality of the respiratory system (HP:0002086)
Abnormal heart morphology (HP:0001627)	Abnormality of the abdominal organs (HP:0002012)	Abnormality of skeletal morphology (HP:0011842)
Situs inversus totalis (HP:0001696)	Abnormality of the skin (HP:0000951)	Abnormality of the hair (HP:0001595)
Abnormality of the nail (HP:0001597)	Abnormality of the immune system (HP:0002715)	Abnormality of the endocrine system (HP:0000818)
Hypogonadism (HP:0000135)	Diabetes mellitus (HP:0000819)	Morphological abnormality of the central nervous system (HP:0002011)

CAKUT clinical tests (30985)

Level 3 Title	Structural renal and urinary tract disease (11003)
Level 4 Title	Congenital Anomaly of the Kidneys and Urinary Tract (CAKUT) (29277)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to renal and liver imaging including ultrasound, MCUG, DMSA Entries ordered left to right in table

Blood pressure (30245.1)	Renal biochemistry (30355.2)	Extended renal biochemistry (33152.1)
Glucose (33153.1)	Bone profile (30317.2)	Urine electrolytes (33187.2)
Urine dip - standard (31317.2)	General Biopsy (33614.1)	General Imaging Diagnostics (33633.1)
Metabolic biochemistry (33160.2)	Urine Protein (33190.1)	

Renal tubular acidosis (11123)

Renal tubular acidosis phenotypes (29195)

Level 3 Title	Disorders of function (11004)
Level 4 Title	Renal tubular acidosis (11123)
Phenotypes	Entries ordered left to right in table

Renal tubular dysfunction (HP:0000124)	Renal tubular acidosis (HP:0001947)	Abnormality of renal resorption (HP:0011038)
Abnormality of renal excretion (HP:0011036)	Nephrocalcinosis (HP:0000121)	Nephrolithiasis (HP:0000787)
Hypertension (HP:0000822)	Hypokalemic alkalosis (HP:0001949)	Hypomagnesemia (HP:0002917)
Hypocalciuria (HP:0003127)	Hypercalciuria (HP:0002150)	Hyperkalemia (HP:0002153)
Abnormality of renin-angiotensin system (HP:0000847)		

Renal Tubular Acidosis clinical tests (30986)

Level 3 Title	Disorders of function (11004)
Level 4 Title	Renal tubular acidosis (11123)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to Abdominal X-ray, Renal tract ultrasound and/or CT and DEXA scan Non-imaging diagnostics refers to water deprivation test and/or stone analysis Entries ordered left to right in table

Glucose (33153.1)	Bone profile (30317.2)	Extended renal biochemistry (33152.1)
Renal biochemistry (30355.2)	Urine electrolytes (33187.2)	Urine dip - standard (31317.2)
General Imaging Diagnostics (33633.1)	General Non-imaging Diagnostics (34838.1)	Metabolic biochemistry (33160.2)
Urine Protein (33190.1)		

Renal tract calcification (or Nephrolithiasis or nephrocalcinosis) (11124)

Renal tract calcification phenotypes (29196)

Level 3 Title	Disorders of function (11004)
Level 4 Title	Renal tract calcification (or Nephrolithiasis or nephrocalcinosis) (11124)
Phenotypes	Entries ordered left to right in table

Nephrocalcinosis (HP:0000121)	Nephrolithiasis (HP:0000787)	Renal tubular dysfunction (HP:0000124)
Renal tubular acidosis (HP:0001947)	Abnormality of renal resorption (HP:0011038)	Abnormality of renal excretion (HP:0011036)
Renal insufficiency (HP:0000083)	Metabolic acidosis (HP:0001942)	Metabolic alkalosis (HP:0200114)
Hypocalcemia (HP:0002901)	Hypercalcemia (HP:0003072)	Hypoparathyroidism (HP:0000829)
Hyperparathyroidism (HP:0000843)	Hypercalciuria (HP:0002150)	Hyperphosphaturia (HP:0003109)
Hyperoxaluria (HP:0003159)	Aminoaciduria (HP:0003355)	Hypocitraturia (HP:0012405)
Hypomagnesiuria (HP:0012609)		

Renal Tract Calcification Test clinical tests (30987)

Level 3 Title	Disorders of function (11004)
Level 4 Title	Renal tract calcification (or Nephrolithiasis or nephrocalcinosis) (11124)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to Abdominal X-ray, Renal tract ultrasound and/or CT and DEXA scan Non-imaging diagnostics refers to water deprivation test and/or stone analysis Entries ordered left to right in table

Blood pressure (30245.1)	Renal biochemistry (30355.2)	Bone profile (30317.2)
Glucose (33153.1)	Extended renal biochemistry (33152.1)	Urine dip - standard (31317.2)
General Imaging Diagnostics (33633.1)	General Non-imaging Diagnostics (34838.1)	Metabolic biochemistry (33160.2)
Urine Protein (33190.1)	Urine metabolic tests (33189.1)	

Extreme early-onset hypertension (15142)

Extreme early-onset hypertension phenotypes (29197)

Level 3 Title	Disorders of function (11004)
Level 4 Title	Extreme early-onset hypertension (15142)
Phenotypes	Entries ordered left to right in table

Hypertension (HP:0000822)	Cerebral aneurysm (HP:0004944)	Microangiopathic hemolytic anemia (HP:0001937)
Renovascular hypertension (HP:0100817)	Hypokalemia (HP:0002900)	Hyperkalemia (HP:0002153)
Metabolic acidosis (HP:0001942)	Metabolic alkalosis (HP:0200114)	Abnormality of acid-base homeostasis (HP:0004360)
Abnormality of thyroid physiology (HP:0002926)	Hypogonadism (HP:0000135)	Ambiguous genitalia (HP:0000062)

Extreme early-onset hypertension clinical tests (30988)

Level 3 Title	Disorders of function (11004)
Level 4 Title	Extreme early-onset hypertension (15142)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to renal and/or adrenal imaging Entries ordered left to right in table

Kidney Imaging (29807.2)	Renal biochemistry (30355.2)	Full Blood Count (30318.2)
Age at diagnosis of chronic kidney disease (30198.2)	Age at ESRD (30199.2)	Extended haematology investigations (33151.1)
Extended renal biochemistry (33152.1)	Metabolic biochemistry (33160.2)	Renin and aldosterone (33166.1)
Sex hormones (33167.2)	Cortisol (33148.1)	Urine electrolytes (33187.2)
Urine dip - standard (31317.2)	General Imaging Diagnostics (33633.1)	Urine Protein (33190.1)

Unexplained kidney failure in young people (36855)

Unexplained kidney failure in young people phenotypes (36911)

Level 3 Title	Disorders of function (11004)
Level 4 Title	Unexplained kidney failure in young people (36855)

Phenotypes

Entries ordered left to right in table

Microscopic hematuria (HP:0002907)	Macroscopic hematuria (HP:0012587)	Edema (HP:0000969)
Proteinuria (HP:0000093)	Abnormality of the abdominal organs (HP:0002012)	Abnormality of the female genitalia (HP:0010460)
Abnormality of the male genitalia (HP:0010461)	Abnormality of the respiratory system (HP:0002086)	Abnormality of skeletal morphology (HP:0011842)
Morphological abnormality of the central nervous system (HP:0002011)	Abnormality of the endocrine system (HP:0000818)	Abnormality of the hair (HP:0001595)
Abnormal heart morphology (HP:0001627)	Abnormality of the immune system (HP:0002715)	Abnormality of the nail (HP:0001597)
Abnormality of the skin (HP:0000951)	Ambiguous genitalia (HP:0000062)	Anosmia (HP:0000458)
Branchial anomaly (HP:0009794)	Cryptorchidism (HP:0000028)	Diabetes mellitus (HP:0000819)
Abnormality of the outer ear (HP:0000356)	Abnormal facial shape (HP:0001999)	Global developmental delay (HP:0001263)
Hearing impairment (HP:0000365)	Hypogonadism (HP:0000135)	Abnormality of the eye (HP:0000478)
Situs inversus totalis (HP:0001696)	Chronic kidney disease (HP:0012622)	Renal cyst (HP:0000107)
Nephronophthisis (HP:0000090)	Gout (HP:0001997)	Hypertension (HP:0000822)
Renal atrophy (HP:0012585)	Multiple glomerular cysts (HP:0100611)	Obesity (HP:0001513)
Intellectual disability (HP:0001249)	Seizures (HP:0001250)	

Unexplained kidney failure in young people clinical tests (36898)

<table>
<tr><td>Level 3 Title</td><td>Disorders of function (11004)</td></tr>
<tr><td>Level 4 Title</td><td>Unexplained kidney failure in young people (36855)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to renal imaging
Entries ordered left to right in table

<table><tr><td>Blood pressure (30245.1)</td><td>Full Blood Count (30318.2)</td><td>Autoantibodies (30112.2)</td></tr><tr><td>Renal biochemistry (30355.2)</td><td>Extended renal biochemistry (33152.1)</td><td>Bone profile (30317.2)</td></tr><tr><td>Glucose (33153.1)</td><td>Renin and aldosterone (33166.1)</td><td>Vitamin B12 (33172.1)</td></tr><tr><td>Lipids (33158.1)</td><td>Inflammatory markers (33156.1)</td><td>Urine Protein (33190.1)</td></tr><tr><td>Complement (33146.1)</td><td>Urine electrolytes (33187.2)</td><td></td></tr></table></td></tr>
</table>

Respiratory disorders (33353)

Interstitial lung disorders (33354)

Familial pulmonary fibrosis (33671)

Familial pulmonary fibrosis phenotypes (33673)

Level 3 Title	Interstitial lung disorders (33354)
Level 4 Title	Familial pulmonary fibrosis (33671)
Phenotypes	Entries ordered left to right in table

Interstitial pulmonary abnormality (HP:0006530)	Pulmonary fibrosis (HP:0002206)	Acute respiratory tract infection (HP:0011948)
Asthma (HP:0002099)	Tachypnea (HP:0002789)	Dyspnea (HP:0002094)
Cough (HP:0012735)	Hemoptysis (HP:0002105)	Aspiration (HP:0002835)
Hypertension (HP:0000822)	Angina pectoris (HP:0001681)	Coronary artery disease (HP:0001677)
Stroke (HP:0001297)	Diabetes mellitus (HP:0000819)	Deep venous thrombosis (HP:0002625)
Pulmonary embolism (HP:0002204)	Neoplasm (HP:0002664)	Renal insufficiency (HP:0000083)
Thromboembolism (HP:0001907)	Rheumatoid arthritis (HP:0001370)	Osteoarthritis (HP:0002758)
Depression (HP:0000716)	Anxiety (HP:0000739)	Gastroesophageal reflux (HP:0002020)

Familial pulmonary fibrosis clinical tests (33672)

<table>
<tr><td>Level 3 Title</td><td>Interstitial lung disorders (33354)</td></tr>
<tr><td>Level 4 Title</td><td>Familial pulmonary fibrosis (33671)</td></tr>
<tr><td>Clinical Tests</td><td>NB. Clinical test guidance:
Imaging diagnostics refers to Chest CT and chest radiograph
Non-imaging diagnostics refers to Lung function tests
Autoantibodies refers to ANA, c-ANCA and a-ANCA
Biopsy refers to lung biopsy
Entries ordered left to right in table

<table><tr><td>Full Blood Count (30318.2)</td><td>Autoantibodies (30112.2)</td><td>General Imaging Diagnostics (33633.1)</td></tr><tr><td>General Biopsy (33614.1)</td><td>Microbiology antibodies (33173.1)</td><td></td></tr></table></td></tr>
</table>

Vascular lung disorders (33355)

Hereditary haemorrhagic telangiectasia (33674)

Hereditary haemorrhagic telangiectasia phenotypes (33675)

Level 3 Title	Vascular lung disorders (33355)
Level 4 Title	Hereditary haemorrhagic telangiectasia (33674)
Phenotypes	Entries ordered left to right in table

Epistaxis (HP:0000421)	Hemoptysis (HP:0002105)	Gastrointestinal hemorrhage (HP:0002239)
Iron deficiency anemia (HP:0001891)	Nasal mucosa telangiectasia (HP:0000434)	Lip telangiectasia (HP:0000214)
Tongue telangiectasia (HP:0000227)	Palate telangiectasia (HP:0002707)	Nail bed telangiectasia (HP:0001232)
Retinal telangiectasia (HP:0007763)	Mucosal telangiectasiae (HP:0100579)	Gastrointestinal telangiectasia (HP:0002604)
Arteriovenous malformation (HP:0100026)	Cerebral arteriovenous malformation (HP:0002408)	Pulmonary arteriovenous malformation (HP:0006548)
Gastrointestinal arteriovenous malformation (HP:0002629)	Hepatic arteriovenous malformation (HP:0006574)	Spinal arteriovenous malformation (HP:0002390)
High-output congestive heart failure (HP:0001722)	Hypoxemia (HP:0012418)	Polycythemia (HP:0001901)
Brain abscess (HP:0030049)	Recurrent abscess formation (HP:0002722)	Ischemic stroke (HP:0002140)
Migraine with aura (HP:0002077)	Migraine without aura (HP:0002083)	Intracranial hemorrhage (HP:0002170)
Pulmonary embolism (HP:0002204)	Deep venous thrombosis (HP:0002625)	Pulmonary arterial hypertension (HP:0002092)
Abnormality of the aorta (HP:0001679)	Juvenile colonic polyposis (HP:0012198)	Juvenile gastrointestinal polyposis (HP:0004784)
Graves disease (HP:0100647)	Hyperthyroidism (HP:0000836)	Hypothyroidism (HP:0000821)
Gout (HP:0001997)		

Hereditary haemorrhagic telangiectasia clinical tests (33676)

Level 3 Title	Vascular lung disorders (33355)
Level 4 Title	Hereditary haemorrhagic telangiectasia (33674)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to Chest CT and liver ultrasound scan Full blood count refers to highest and lowest known haemoglobin Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	Liver biochemistry (30328.2)	Full Blood Count (30318.2)

Familial and multiple pulmonary arteriovenous malformations (33677)

Familial and multiple pulmonary arteriovenous malformations phenotypes (33678)

Level 3 Title	Vascular lung disorders (33355)
Level 4 Title	Familial and multiple pulmonary arteriovenous malformations (33677)
Phenotypes	Entries ordered left to right in table

Pulmonary arteriovenous malformation (HP:0006548)	Hypoxemia (HP:0012418)	Polycythemia (HP:0001901)
Brain abscess (HP:0030049)	Recurrent abscess formation (HP:0002722)	Ischemic stroke (HP:0002140)
Hemoptysis (HP:0002105)	Migraine with aura (HP:0002077)	Migraine without aura (HP:0002083)
Epistaxis (HP:0000421)	Gastrointestinal hemorrhage (HP:0002239)	Iron deficiency anemia (HP:0001891)
Nasal mucosa telangiectasia (HP:0000434)	Lip telangiectasia (HP:0000214)	Tongue telangiectasia (HP:0000227)
Palate telangiectasia (HP:0002707)	Nail bed telangiectasia (HP:0001232)	Retinal telangiectasia (HP:0007763)
Mucosal telangiectasiae (HP:0100579)	Gastrointestinal telangiectasia (HP:0002604)	Arteriovenous malformation (HP:0100026)
Hepatic arteriovenous malformation (HP:0006574)	Cerebral arteriovenous malformation (HP:0002408)	Gastrointestinal arteriovenous malformation (HP:0002629)
Spinal arteriovenous malformation (HP:0002390)	High-output congestive heart failure (HP:0001722)	Abnormality of the liver (HP:0001392)
Intracranial hemorrhage (HP:0002170)	Pulmonary embolism (HP:0002204)	Deep venous thrombosis (HP:0002625)
Pulmonary arterial hypertension (HP:0002092)	Abnormality of the aorta (HP:0001679)	Juvenile colonic polyposis (HP:0012198)
Juvenile gastrointestinal polyposis (HP:0004784)	Choriocarcinoma (HP:0100768)	Gout (HP:0001997)
Graves disease (HP:0100647)	Hyperthyroidism (HP:0000836)	Hypothyroidism (HP:0000821)

Level 3 Title	Vascular lung disorders (33355)
Level 4 Title	Familial and multiple pulmonary arteriovenous malformations (33677)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to Chest CT and liver ultrasound scan Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	Liver biochemistry (30328.2)

Structural lung disorders (42203)

Familial primary spontaneous pneumothorax (41819)

Familial primary spontaneous pneumothorax phenotypes (42104)

Level 3 Title	Structural lung disorders (42203)
Level 4 Title	Familial primary spontaneous pneumothorax (41819)
Phenotypes	Entries ordered left to right in table

Pneumothorax (HP:0002107)	Thoracic aortic aneurysm (HP:0012727)	Ectopia lentis (HP:0001083)
Dental crowding (HP:0000678)	Arachnodactyly (HP:0001166)	Disproportionate tall stature (HP:0001519)
Reduced upper to lower segment ratio (HP:0012773)	Scoliosis (HP:0002650)	Joint hypermobility (HP:0001382)
Pectus carinatum (HP:0000768)	Pectus excavatum (HP:0000767)	Pes planus (HP:0001763)
Striae distensae (HP:0001065)	Soft skin (HP:0000977)	Thin skin (HP:0000963)
Cutis laxa (HP:0000973)	Atypical scarring of skin (HP:0000987)	Abnormal facial shape (HP:0001999)
High palate (HP:0000218)	Neoplasm (HP:0002664)	Angiofibromas (HP:0010615)
Clear cell renal cell carcinoma (HP:0006770)	Papillary renal cell carcinoma (HP:0006766)	Renal oncocytoma (HP:0011798)
Renal cell carcinoma (HP:0005584)		

Familial primary spontaneous pneumothorax clinical tests (41820)

<table>
<tr><td>Level 3 Title</td><td>Structural lung disorders (42203)</td></tr>
<tr><td>Level 4 Title</td><td>Familial primary spontaneous pneumothorax (41819)</td></tr>
<tr><td>Clinical Tests</td><td>Clinical test guidance:
Additional Measurements refers to Parental heights, Arm span, Lower segment, Upper segment (Height-Lower segment)
General Imaging Diagnostics refers to chest X-Rays
Entries ordered left to right in table

<table><tr><td>Smoking status (31464.1)</td><td>Additional body measurements (30247.2)</td><td>Beighton test (31476.1)</td></tr><tr><td>Blood pressure (30245.1)</td><td>General Imaging Diagnostics (33633.1)</td><td></td></tr></table></td></tr>
</table>

Rheumatological disorders (11009)

Multi-system inflammatory or autoimmune disorders (11008)

Periodic fever syndromes and amyloidosis (11127)

Periodic fever syndromes phenotypes (29221)

Level 3 Title	Multi-system inflammatory or autoimmune disorders (11008)
Level 4 Title	Periodic fever syndromes and amyloidosis (11127)
Phenotypes	Entries ordered left to right in table

Amyloidosis (HP:0011034)	Unexplained fevers (HP:0001955)	Lymphadenopathy (HP:0002716)
Arthritis (HP:0001369)	Skin rash (HP:0000988)	Inflammatory abnormality of the eye (HP:0100533)
Proteinuria (HP:0000093)	Chronic kidney disease (HP:0012622)	Decreased liver function (HP:0001410)
Peripheral neuropathy (HP:0009830)	Abnormality of the autonomic nervous system (HP:0002270)	Orthostatic hypotension (HP:0001278)
Congestive heart failure (HP:0001635)	Arrhythmia (HP:0011675)	Functional abnormality of the gastrointestinal tract (HP:0012719)
Abdominal pain (HP:0002027)		

Periodic fever syndromes clinical tests (30991)

Level 3 Title	Multi-system inflammatory or autoimmune disorders (11008)
Level 4 Title	Periodic fever syndromes and amyloidosis (11127)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to biopsies of kidney, skin and/or peritoneum Imaging diagnostics refers to SAP scan, MRI brain, renal ultrasound, and other relevant modalities Non-imaging diagnostics refers to nerve conduction studies Entries ordered left to right in table

Blood pressure (30245.1)	Inflammation metrics (30200.2)	Renal Biopsy (30153.3)
Serum immunoglobulins (30338.2)	Renal biochemistry (30355.2)	Inflammatory markers (33156.1)
Bone profile (30317.2)	Extended renal biochemistry (33152.1)	Urine electrolytes (33187.2)
Urine dip - standard (31317.2)	General Biopsy (33614.1)	General Imaging Diagnostics (33633.1)
General Non-imaging Diagnostics (34838.1)	Metabolic biochemistry (33160.2)	Urine Protein (33190.1)

Juvenile dermatomyositis (29219)

Juvenile dermatomyositis phenotypes (29220)

Level 3 Title	Multi-system inflammatory or autoimmune disorders (11008)
Level 4 Title	Juvenile dermatomyositis (29219)
Phenotypes	Entries ordered left to right in table

Fever (HP:0001945)	Muscle weakness (HP:0001324)	Generalized muscle weakness (HP:0003324)
Proximal muscle weakness (HP:0003701)	Myositis (HP:0100614)	EMG: myopathic abnormalities (HP:0003458)
Arthritis (HP:0001369)	Flexion contracture (HP:0001371)	Skin rash (HP:0000988)
Skin ulcer (HP:0200042)	Edema (HP:0000969)	Generalized edema (HP:0007430)
Periorbital edema (HP:0100539)	Erythema (HP:0010783)	Facial erythema (HP:0001041)
Cutis marmorata (HP:0000965)	Cutaneous photosensitivity (HP:0000992)	Abnormality of the periungual region (HP:0100803)
Alopecia (HP:0001596)	Panniculitis (HP:0012490)	Lipodystrophy (HP:0009125)
Calcinosis (HP:0003761)	Abnormality of the cardiovascular system (HP:0001626)	Abnormal heart morphology (HP:0001627)
Dysphonia (HP:0001618)	Dysphagia (HP:0002015)	Interstitial pulmonary abnormality (HP:0006530)
Abnormality of the gastrointestinal tract (HP:0011024)	Abdominal pain (HP:0002027)	Weight loss (HP:0001824)
Neoplasm (HP:0002664)	Fatigue (HP:0012378)	Irritability (HP:0000737)
Autoimmune antibody positivity (HP:0030057)		

Juvenile dermatomyositis clinical tests (30992)

Level 3 Title	Multi-system inflammatory or autoimmune disorders (11008)
Level 4 Title	Juvenile dermatomyositis (29219)
Clinical Tests	NB. Clinical test guidance: Biopsy refers to muscle biopsy Imaging diagnostics refers to as muscle MRI Entries ordered left to right in table

Autoantibodies (30112.2)	Childhood Myositis Assessment (30222.1)	Manual Muscle Testing 8 (MMT8) Assessment (30232.1)
VAS Assessment (30239.2)	Childhood Health Assessment (30220.1)	Liver biochemistry (30328.2)
Other enzymes (33123.1)	General Biopsy (33614.1)	General Imaging Diagnostics (33633.1)
General Non-imaging Diagnostics (34838.1)		

Connective tissues disorders (36930)

Kyphoscoliotic Ehlers-Danlos syndrome (36853)

Kyphoscoliotic Ehlers-Danlos syndrome phenotypes (36909)

Level 3 Title	Connective tissues disorders (36930)
Level 4 Title	Kyphoscoliotic Ehlers-Danlos syndrome (36853)
Phenotypes	Entries ordered left to right in table

Kyphosis (HP:0002808)	Scoliosis (HP:0002650)	Kyphoscoliosis (HP:0002751)
Aneurysm (HP:0002617)	Arterial dissection (HP:0005294)	Hyperextensible skin (HP:0000974)
Atrophic scars (HP:0001075)	Excessive wrinkling of palmar skin (HP:0007605)	Multiple plantar creases (HP:0008113)
Multiple palmar creases (HP:0006114)	Muscular hypotonia (HP:0001252)	Hearing impairment (HP:0000365)
Joint hypermobility (HP:0001382)	Abnormality of the voice (HP:0001608)	Glaucoma (HP:0000501)
Spontaneous rupture of the globe (HP:0010727)	Short stature (HP:0004322)	Tall stature (HP:0000098)
Arachnodactyly (HP:0001166)	Flexion contracture (HP:0001371)	Increased susceptibility to fractures (HP:0002659)
Joint dislocation (HP:0001373)	Microcornea (HP:0000482)	Osteopenia (HP:0000938)
Abnormality of the sternum (HP:0000766)	Umbilical hernia (HP:0001537)	Inguinal hernia (HP:0000023)
Retinal detachment (HP:0000541)	Blue sclerae (HP:0000592)	Aortic root dilatation (HP:0002616)

Classical Ehlers-Danlos Syndrome (41860)

Classical Ehlers-Danlos Syndrome phenotypes (42109)

Level 3 Title	Connective tissues disorders (36930)
Level 4 Title	Classical Ehlers-Danlos Syndrome (41860)
Phenotypes	Entries ordered left to right in table

Bruising susceptibility (HP:0000978)	Hyperextensible skin (HP:0000974)	Fragile skin (HP:0001030)
Atrophic scars (HP:0001075)	Molluscoid pseudotumors (HP:0000993)	Poor wound healing (HP:0001058)
Subcutaneous spheroids (HP:0025014)	Joint hypermobility (HP:0001382)	Hernia (HP:0100790)
Scoliosis (HP:0002650)	Arterial rupture (HP:0025019)	Capillary fragility (HP:0025017)
Epicanthus (HP:0000286)		

Classical Ehlers-Danlos Syndrome clinical tests (41862)

Level 3 Title	Connective tissues disorders (36930)
Level 4 Title	Classical Ehlers-Danlos Syndrome (41860)
Clinical Tests	Clinical test guidance: General Imaging Diagnostic refers to electron microscopy Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Skeletal disorders (11005)

Skeletal dysplasias (11007)

Multiple Epiphyseal Dysplasia (11125)

Multiple Epiphyseal Dysplasia phenotypes (29199)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Multiple Epiphyseal Dysplasia (11125)
Phenotypes	Entries ordered left to right in table

Multiple epiphyseal dysplasia (HP:0002654)	Mild short stature (HP:0003502)	Disproportionate short stature (HP:0003498)
Brachydactyly syndrome (HP:0001156)	Short finger (HP:0009381)	Short toe (HP:0001831)
Genu varum (HP:0002970)	Genu valgum (HP:0002857)	Arthralgia (HP:0002829)
Premature osteoarthritis (HP:0003088)	Back pain (HP:0003418)	Gait disturbance (HP:0001288)
Joint hypermobility (HP:0001382)	Abnormal bone ossification (HP:0011849)	

Chondrodysplasia punctata (15143)

Chondrodysplasia punctata phenotypes (29201)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Chondrodysplasia punctata (15143)
Phenotypes	Entries ordered left to right in table

Disproportionate short stature (HP:0003498)	Rhizomelia (HP:0008905)	Brachydactyly syndrome (HP:0001156)
Mesomelia (HP:0003027)	Microcephaly (HP:0000252)	Short finger (HP:0009381)
Short distal phalanx of finger (HP:0009882)	Short metacarpal (HP:0010049)	Short toe (HP:0001831)
Short distal phalanx of toe (HP:0001857)	Scoliosis (HP:0002650)	Short humerus (HP:0005792)
Radial bowing (HP:0002986)	Hypoplasia of the ulna (HP:0003022)	Short femur (HP:0003097)
Short tibia (HP:0005736)	Kyphosis (HP:0002808)	Flexion contracture (HP:0001371)
Talipes (HP:0001883)	Joint hypermobility (HP:0001382)	Intellectual disability (HP:0001249)
Seizures (HP:0001250)	Hypertonia (HP:0001276)	Midface retrusion (HP:0011800)
Malar flattening (HP:0000272)	Depressed nasal bridge (HP:0005280)	Short nose (HP:0003196)
Underdeveloped nasal alae (HP:0000430)	Cleft upper lip (HP:0000204)	Corneal opacity (HP:0007957)
Cataract (HP:0000518)	Abnormality of the optic nerve (HP:0000587)	Dry skin (HP:0000958)
Hyperkeratosis (HP:0000962)	Ichthyosis (HP:0008064)	Erythroderma (HP:0001019)
Fine hair (HP:0002213)	Coarse hair (HP:0002208)	Sparse hair (HP:0008070)
Alopecia (HP:0001596)	Small nail (HP:0001792)	Sensorineural hearing impairment (HP:0000407)
Tracheobronchomalacia (HP:0002786)	Abnormality of cardiovascular system morphology (HP:0030680)	Atrial septal defect (HP:0001631)
Ventricular septal defect (HP:0001629)	Tetralogy of Fallot (HP:0001636)	Pulmonary artery hypoplasia (HP:0004971)
Patent ductus arteriosus (HP:0001643)	Mitral valve prolapse (HP:0001634)	Elevated levels of phytanic acid (HP:0010571)
Hyperemesis gravidarum (HP:0012188)		

Thoracic dystrophies (11126)

Thoracic dystrophies phenotypes (29202)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Thoracic dystrophies (11126)
Phenotypes	Entries ordered left to right in table

Short stature (HP:0004322)	Disproportionate short stature (HP:0003498)	Mesomelia (HP:0003027)
Acromelia (HP:0010884)	Acromesomelia (HP:0003086)	Brachydactyly syndrome (HP:0001156)
Hand polydactyly (HP:0001161)	Postaxial hand polydactyly (HP:0001162)	Preaxial hand polydactyly (HP:0001177)
Mesoaxial hand polydactyly (HP:0006159)	Foot polydactyly (HP:0001829)	Postaxial foot polydactyly (HP:0001830)
Preaxial foot polydactyly (HP:0001841)	Short toe (HP:0001831)	Narrow chest (HP:0000774)
Long thorax (HP:0100818)	Genu valgum (HP:0002857)	Corner fracture of metaphysis (HP:0003908)
Platyspondyly (HP:0000926)	Narrow greater sacrosciatic notches (HP:0003375)	Respiratory distress (HP:0002098)
Retinal dystrophy (HP:0000556)	Rod-cone dystrophy (HP:0000510)	Dextrocardia (HP:0001651)
Situs inversus totalis (HP:0001696)	Polysplenia (HP:0001748)	Asplenia (HP:0001746)
Polycystic kidney dysplasia (HP:0000113)	Polycystic liver disease (HP:0006557)	Hepatic fibrosis (HP:0001395)
Bile duct proliferation (HP:0001408)	Jaundice (HP:0000952)	Pancreatic fibrosis (HP:0100732)
Pancreatic cysts (HP:0001737)		

Stickler syndrome (11129)

Stickler syndrome phenotypes (29203)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Stickler syndrome (11129)
Phenotypes	Entries ordered left to right in table

Myopia (HP:0000545)	Severe Myopia (HP:0011003)	Visual impairment (HP:0000505)
Nonprogressive visual loss (HP:0200068)	Progressive visual loss (HP:0000529)	Slow decrease in visual acuity (HP:0007924)
Blindness (HP:0000618)	Glaucoma (HP:0000501)	Cataract (HP:0000518)
Abnormality of the vitreous humor (HP:0004327)	Retinal detachment (HP:0000541)	Rhegmatogenous retinal detachment (HP:0012230)
Abnormality of the fundus (HP:0001098)	Abnormality of head or neck (HP:0000152)	Abnormality of the ear (HP:0000598)
Hearing impairment (HP:0000365)	Conductive hearing impairment (HP:0000405)	Sensorineural hearing impairment (HP:0000407)
Short nose (HP:0003196)	Midface retrusion (HP:0011800)	Malar flattening (HP:0000272)
Depressed nasal bridge (HP:0005280)	Anteverted nares (HP:0000463)	Pierre-Robin sequence (HP:0000201)
Micrognathia (HP:0000347)	Cleft palate (HP:0000175)	Bifid uvula (HP:0000193)
Macroglossia (HP:0000158)	Mitral valve prolapse (HP:0001634)	Abnormality of the musculature (HP:0003011)
Abnormality of the skeletal system (HP:0000924)	Pectus excavatum (HP:0000767)	Scoliosis (HP:0002650)
Kyphosis (HP:0002808)	Long fingers (HP:0100807)	Arachnodactyly (HP:0001166)
Joint hypermobility (HP:0001382)	Arthralgia (HP:0002829)	Arthropathy (HP:0003040)
Premature osteoarthritis (HP:0003088)	Abnormality of femoral epiphysis (HP:0006499)	Irregular femoral epiphysis (HP:0006361)
Enlarged epiphyses (HP:0010580)	Epiphyseal dysplasia (HP:0002656)	Platyspondyly (HP:0000926)
Beaking of vertebral bodies (HP:0004568)	Spondyloepiphyseal dysplasia (HP:0002655)	

Stickler clinical tests (30989)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Stickler syndrome (11129)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to skeletal survey and medical photography as appropriate Entries ordered left to right in table

Refraction Error (30978.2)	Visual Acuity (30970.2)

Osteogenesis imperfecta (30627)

Osteogenesis imperfecta phenotypes (30628)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Osteogenesis imperfecta (30627)
Phenotypes	Entries ordered left to right in table

Joint hypermobility (HP:0001382)	Corneal perforation (HP:0100583)	Blue sclerae (HP:0000592)
Cutis laxa (HP:0000973)	Joint dislocation (HP:0001373)	Bruising susceptibility (HP:0000978)
Abnormality of the teeth (HP:0000164)	Dentinogenesis imperfecta (HP:0000703)	Sensorineural hearing impairment (HP:0000407)
Low-set ears (HP:0000369)	Pointed chin (HP:0000307)	Abnormal heart morphology (HP:0001627)
Abnormality of the respiratory system (HP:0002086)	Gastroesophageal reflux (HP:0002020)	Gastric ulcer (HP:0002592)
Nephrolithiasis (HP:0000787)	Global developmental delay (HP:0001263)	Autistic behavior (HP:0000729)
Contractures of the joints of the upper limbs (HP:0100360)	Contractures of the joints of the lower limbs (HP:0005750)	Abnormality of the vertebral column (HP:0000925)
Scoliosis (HP:0002650)	Kyphosis (HP:0002808)	Osteopenia (HP:0000938)
Osteoarthritis (HP:0002758)	Craniosynostosis (HP:0001363)	Turricephaly (HP:0000262)
Wormian bones (HP:0002645)	Basilar impression (HP:0005758)	Basilar invagination (HP:0012366)
Platybasia (HP:0002691)	Periosteal new bone of humeral diaphysis (HP:0003931)	Periosteal new bone of humerus (HP:0003878)
Coarse humeral trabeculae (HP:0003866)	Calcification of the interosseus membrane of the forearm (HP:0030267)	Abnormality of the hip bone (HP:0003272)
Platyspondyly (HP:0000926)	Protrusio acetabuli (HP:0003179)	Spondylolysis (HP:0003304)
Spondylolisthesis (HP:0003302)	Multiple prenatal fractures (HP:0005855)	Fractures of the long bones (HP:0003084)
Multiple rib fractures (HP:0006640)	Multiple small vertebral fractures (HP:0005877)	

Osteogenesis imperfecta clinical tests (33368)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Osteogenesis imperfecta (30627)
Clinical Tests	NB. Clinical test guidance: Imaging diagnostics refers to skeletal survey and medical photography as appropriate Non-imaging diagnostics refers to hearing test Entries ordered left to right in table

General Imaging Diagnostics (33633.1)	General Non-imaging Diagnostics (34838.1)

Unexplained skeletal dysplasia (36854)

Unexplained skeletal dysplasia phenotypes (36910)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Unexplained skeletal dysplasia (36854)
Phenotypes	Entries ordered left to right in table

Short stature (HP:0004322)	Joint hypermobility (HP:0001382)	Brachydactyly syndrome (HP:0001156)
Mesomelia (HP:0003027)	Acromelia (HP:0010884)	Acromesomelia (HP:0003086)
Hand polydactyly (HP:0001161)	Foot polydactyly (HP:0001829)	Gait disturbance (HP:0001288)
Abnormality of the vertebral column (HP:0000925)	Premature osteoarthritis (HP:0003088)	Back pain (HP:0003418)
Abnormality of the thorax (HP:0000765)	Craniosynostosis (HP:0001363)	Abnormality of the eye (HP:0000478)
Intellectual disability (HP:0001249)	Hearing impairment (HP:0000365)	Abnormal facial shape (HP:0001999)
Irregular vertebral endplates (HP:0003301)	Kyphosis (HP:0002808)	Osteoarthritis (HP:0002758)
Platyspondyly (HP:0000926)	Pointed proximal second through fifth metacarpals (HP:0001223)	Pseudoepiphyses (HP:0010584)
Talipes (HP:0001883)	Corner fracture of metaphysis (HP:0003908)	Genu valgum (HP:0002857)
Mesoaxial hand polydactyly (HP:0006159)	Narrow greater sacrosciatic notches (HP:0003375)	Postaxial polydactyly (HP:0100259)
Preaxial polydactyly (HP:0100258)	Cleft palate (HP:0000175)	Hip dysplasia (HP:0001385)
Flexion contracture (HP:0001371)	Scoliosis (HP:0002650)	Hypodontia (HP:0000668)
Increased number of teeth (HP:0011069)	Natal tooth (HP:0000695)	Macrocephaly (HP:0000256)
Microcephaly (HP:0000252)		

Unexplained skeletal dysplasia clinical tests (40316)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Unexplained skeletal dysplasia (36854)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to skeletal survey and medical photographs as relevant Entries ordered left to right in table

Bone profile (30317.2)	General Imaging Diagnostics (33633.1)	Growth hormones (33154.1)

Amelogenesis imperfecta (55449)

Amelogenesis imperfecta phenotypes (68111)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Amelogenesis imperfecta (55449)
Phenotypes	Entries ordered left to right in table

Amelogenesis imperfecta (HP:0000705)	Hypoplasia of dental enamel (HP:0006297)	Hypomature dental enamel (HP:0011085)
Hypocalcification of dental enamel (HP:0011084)	Dental enamel pits (HP:0009722)	Taurodontia (HP:0000679)
Yellow-brown discoloration of the teeth (HP:0006286)	Anterior open bite (HP:0200095)	Overbite (HP:0011094)
Abnormality of the hair (HP:0001595)	Nail dysplasia (HP:0002164)	Hypoparathyroidism (HP:0000829)
Chronic mucocutaneous candidiasis (HP:0002728)	Rickets (HP:0002748)	Cone/cone-rod dystrophy (HP:0000548)
Sensorineural hearing impairment (HP:0000407)	Nephrocalcinosis (HP:0000121)	Intellectual disability (HP:0001249)
Seizures (HP:0001250)	Immunodeficiency (HP:0002721)	Short stature (HP:0004322)

Amelogenesis imperfecta clinical tests (55450)

Level 3 Title	Skeletal dysplasias (11007)
Level 4 Title	Amelogenesis imperfecta (55449)
Clinical Tests	NB clinical testing guidance: General Imaging Diagnostics refers to dental radiography Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Craniosynostosis (30775)

Craniosynostosis syndromes (11006)

Craniosynostosis syndromes phenotypes (29198)

Level 3 Title	Craniosynostosis (30775)
Level 4 Title	Craniosynostosis syndromes (11006)
Phenotypes	Entries ordered left to right in table

Craniosynostosis (HP:0001363)	Multiple suture craniosynostosis (HP:0011324)	Unicoronal synostosis (HP:0011315)
Bicoronal synostosis (HP:0011318)	Left unicoronal synostosis (HP:0011316)	Right unicoronal synostosis (HP:0011317)
Sagittal craniosynostosis (HP:0004442)	Metopic synostosis (HP:0011330)	Unilambdoid synostosis (HP:0011320)
Left unilambdoid synostosis (HP:0011321)	Right unilambdoid synostosis (HP:0011322)	Increased intracranial pressure (HP:0002516)
Oral cleft (HP:0000202)	Ptosis (HP:0000508)	Strabismus (HP:0000486)
Hearing impairment (HP:0000365)	Premature birth (HP:0001622)	Maternal teratogenic exposure (HP:0011438)

Craniosynostosis clinical tests (30990)

Level 3 Title	Craniosynostosis (30775)
Level 4 Title	Craniosynostosis syndromes (11006)
Clinical Tests	Entries ordered left to right in table

Development Quotient Assessment (30244.2)

Choanal anomalies (31500)

Choanal atresia (11078)

Choanal atresia phenotypes (29232)

Level 3 Title	Choanal anomalies (31500)
Level 4 Title	Choanal atresia (11078)
Phenotypes	Entries ordered left to right in table

Choanal atresia (HP:0000453)	Membranous choanal atresia (HP:0011820)	Choanal stenosis (HP:0000452)
Global developmental delay (HP:0001263)	Intellectual disability (HP:0001249)	Growth delay (HP:0001510)
Microcephaly (HP:0000252)	Abnormality of the thyroid gland (HP:0000820)	Craniosynostosis (HP:0001363)
Abnormal facial shape (HP:0001999)	Abnormality of the pinna (HP:0000377)	Microtia (HP:0008551)
Abnormality of the auditory canal (HP:0000372)	Morphological abnormality of the semicircular canal (HP:0011380)	Conductive hearing impairment (HP:0000405)
Sensorineural hearing impairment (HP:0000407)	Underdeveloped nasal alae (HP:0000430)	Abnormality of the eye (HP:0000478)
Hypertelorism (HP:0000316)	Nasolacrimal duct obstruction (HP:0000579)	Lower eyelid coloboma (HP:0000652)
Upper eyelid coloboma (HP:0000636)	Malar flattening (HP:0000272)	Cleft upper lip (HP:0000204)
Cleft palate (HP:0000175)	Bifid uvula (HP:0000193)	Micrognathia (HP:0000347)
Abnormality of the teeth (HP:0000164)	Polydactyly (HP:0010442)	Abnormality of cardiovascular system morphology (HP:0030680)
Abnormality of the genitourinary system (HP:0000119)	Renal dysplasia (HP:0000110)	Abnormality of the anus (HP:0004378)

Tumour syndromes (11012)

Breast and endocrine (11013)

Familial breast and or ovarian cancer (11131)

Familial breast and or Ovarian cancer phenotypes (40359)

Level 3 Title	Breast and endocrine (11013)
Level 4 Title	Familial breast and or ovarian cancer (11131)
Phenotypes	Entries ordered left to right in table

Breast carcinoma (HP:0003002)	Ovarian neoplasm (HP:0100615)

Multiple endocrine tumours (11132)

Multiple endocrine tumours phenotypes (40365)

Level 3 Title	Breast and endocrine (11013)
Level 4 Title	Multiple endocrine tumours (11132)
Phenotypes	Entries ordered left to right in table

Parathyroid hyperplasia (HP:0008208)	Medullary thyroid carcinoma (HP:0002865)	Pituitary adenoma (HP:0002893)
Adrenocortical adenoma (HP:0008256)	Adrenocortical carcinoma (HP:0006744)	Neuroendocrine neoplasm (HP:0100634)
Carcinoid tumor (HP:0100570)	Pheochromocytoma (HP:0002666)	Paraganglioma (HP:0002668)
Nodular goiter (HP:0005994)	Growth hormone excess (HP:0000845)	Hypoglycemia (HP:0001943)
Hyperparathyroidism (HP:0000843)	Parathyroid adenoma (HP:0002897)	Hypercalcemia (HP:0003072)
Elevated calcitonin (HP:0003528)	Insulinoma (HP:0012197)	Zollinger-Ellison syndrome (HP:0002044)
Thick lower lip vermilion (HP:0000179)	Ganglioneuroma (HP:0003005)	Thick eyebrow (HP:0000574)
Subcutaneous lipoma (HP:0001031)	Cafe-au-lait spot (HP:0000957)	Kyphoscoliosis (HP:0002751)
Pectus excavatum (HP:0000767)	High palate (HP:0000218)	Disproportionate tall stature (HP:0001519)
Prominent corneal nerve fibers (HP:0010726)		

Neuro-endocrine Tumours- PCC and PGL (11133)

Neuro-endocrine Tumours- PCC and PGL phenotypes (40367)

Level 3 Title	Breast and endocrine (11013)
Level 4 Title	Neuro-endocrine Tumours- PCC and PGL (11133)
Phenotypes	Entries ordered left to right in table

Pheochromocytoma (HP:0002666)	Paraganglioma (HP:0002668)

Parathyroid cancer (30611)

Parathyroid cancer phenotypes (40369)

Level 3 Title	Breast and endocrine (11013)
Level 4 Title	Parathyroid cancer (30611)
Phenotypes	Entries ordered left to right in table

Parathyroid carcinoma (HP:0006780)	Hyperparathyroidism (HP:0000843)	Parathyroid adenoma (HP:0002897)
Hypercalcemia (HP:0003072)		

Inherited non-medullary thyroid cancer (41884)

Inherited non-medullary thyroid cancer phenotypes (42112)

Level 3 Title	Breast and endocrine (11013)
Level 4 Title	Inherited non-medullary thyroid cancer (41884)
Phenotypes	Entries ordered left to right in table

Follicular thyroid carcinoma (HP:0006731)	Papillary thyroid carcinoma (HP:0002895)	Anaplastic thyroid carcinoma (HP:0011779)
Thyroiditis (HP:0100646)	Multinodular goiter (HP:0005987)	Hashimoto thyroiditis (HP:0000872)
Graves disease (HP:0100647)	Goiter (HP:0000853)	

Inherited non-medullary thyroid cancer clinical tests (41886)

Level 3 Title	Breast and endocrine (11013)
Level 4 Title	Inherited non-medullary thyroid cancer (41884)
Clinical Tests	Clinical test guidance: Biopsy refers to biopsy or resection pathology results of the proband's tumour(s) Entries ordered left to right in table

General Biopsy (33614.1)

Familial colon cancer (11135)

Familial colon cancer phenotypes (40360)

Level 3 Title	GI tract (11014)
Level 4 Title	Familial colon cancer (11135)
Phenotypes	Entries ordered left to right in table

Colon cancer (HP:0003003)	Large intestinal polyposis (HP:0030255)	Small intestinal polyposis (HP:0030256)
Multiple gastric polyps (HP:0004394)	Endometrial carcinoma (HP:0012114)	Ovarian neoplasm (HP:0100615)
Pancreatic adenocarcinoma (HP:0006725)	Neoplasm of the ureter (HP:0100516)	Neoplasm of the skin (HP:0008069)
Stomach cancer (HP:0012126)	Biliary tract neoplasm (HP:0100574)	Neoplasm of the small intestine (HP:0100833)
Glioblastoma (HP:0100843)	Osteoma (HP:0100246)	

Multiple bowel polyps (30615)

Multiple bowel polyps phenotypes (40364)

Level 3 Title	GI tract (11014)
Level 4 Title	Multiple bowel polyps (30615)
Phenotypes	Entries ordered left to right in table

Colon cancer (HP:0003003)	Large intestinal polyposis (HP:0030255)	Small intestinal polyposis (HP:0030256)
Juvenile gastrointestinal polyposis (HP:0004784)	Adenomatous colonic polyposis (HP:0005227)	Hyperplastic colonic polyposis (HP:0012183)
Hamartomatous polyposis (HP:0004390)		

Peutz-Jeghers syndrome (36533)

Peutz-Jeghers syndrome phenotypes (36655)

Level 3 Title	GI tract (11014)
Level 4 Title	Peutz-Jeghers syndrome (36533)
Phenotypes	Entries ordered left to right in table

Abnormality of skin pigmentation (HP:0001000)	Perioral hyperpigmentation (HP:0010802)	Abnormal pigmentation of the oral mucosa (HP:0100669)
Large intestinal polyposis (HP:0030255)	Adenomatous colonic polyposis (HP:0005227)	Hamartomatous polyposis (HP:0004390)
Small intestinal polyposis (HP:0030256)	Intussusception (HP:0002576)	Intestinal bleeding (HP:0002584)
Cervix cancer (HP:0030079)	Testicular neoplasm (HP:0010788)	Colon cancer (HP:0003003)
Breast carcinoma (HP:0003002)	Neoplasm of the small intestine (HP:0100833)	Ovarian neoplasm (HP:0100615)

Muscle and nerve (11015)

Familial rhabdomyosarcoma or sarcoma (11138)

Familial rhabdomyosarcoma or sarcoma phenotypes (40361)

Level 3 Title	Muscle and nerve (11015)		
Level 4 Title	Familial rhabdomyosarcoma or sarcoma (11138)		
Phenotypes	Entries ordered left to right in table		
	Sarcoma (HP:0100242)	Rhabdomyosarcoma (HP:0002859)	Breast carcinoma (HP:0003002)
	Leukemia (HP:0001909)	Adrenocortical carcinoma (HP:0006744)	

Familial tumour syndromes of the central and peripheral nervous system (30619)

Familial tumour syndromes of the central and peripheral nervous system phenotypes (40362)

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<tr><td>Phenotypes</td><td>Entries ordered left to right in table
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<tr><td>Glioma (HP:0009733)</td><td>Astrocytoma (HP:0009592)</td><td>Meningioma (HP:0002858)</td></tr>
<tr><td>Schwannoma (HP:0100008)</td><td>Vestibular Schwannoma (HP:0009588)</td><td>Lisch nodules (HP:0009737)</td></tr>
<tr><td>Cafe-au-lait spot (HP:0000957)</td><td>Renal artery stenosis (HP:0001920)</td><td>Axillary freckling (HP:0000997)</td></tr>
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<tr><td>Hemangioblastoma (HP:0010797)</td><td></td><td></td></tr>
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Neurofibromatosis Type 1 (38874)

Tumour proband phenotypes (29217)

Level 3 Title	Muscle and nerve (11015)
Level 4 Title	Neurofibromatosis Type 1 (38874)
Phenotypes	Entries ordered left to right in table

Macrocephaly (HP:0000256)	Palmar pits (HP:0010610)	Plantar pits (HP:0010612)
Odontogenic keratocysts of the jaw (HP:0010603)	Calcification of falx cerebri (HP:0005462)	Bifid ribs (HP:0000892)
Meningioma (HP:0002858)	Schwannoma (HP:0100008)	Lisch nodules (HP:0009737)
Cafe-au-lait spot (HP:0000957)	Renal artery stenosis (HP:0001920)	Axillary freckling (HP:0000997)
Inguinal freckling (HP:0030052)	Scoliosis (HP:0002650)	Pseudoarthrosis (HP:0005864)
Neurofibromas (HP:0001067)	Intellectual disability (HP:0001249)	Delayed speech and language development (HP:0000750)
Autistic behavior (HP:0000729)	Peripheral neuropathy (HP:0009830)	Ataxia (HP:0001251)
Hydrocephalus (HP:0000238)	Aqueductal stenosis (HP:0002410)	Epiretinal membrane (HP:0100014)
Glaucoma (HP:0000501)	Juvenile posterior subcapsular lenticular opacities (HP:0007935)	Juvenile cortical cataract (HP:0007876)
Cerebral hamartomata (HP:0009731)	Retinal hamartoma (HP:0009594)	Hearing impairment (HP:0000365)
Tinnitus (HP:0000360)		

Genodermatoses with malignancies (30623)

Genodermatoses with malignancies phenotypes (40363)

Level 3 Title	Skin (11016)
Level 4 Title	Genodermatoses with malignancies (30623)
Phenotypes	Entries ordered left to right in table

Macrocephaly (HP:0000256)	Palmar pits (HP:0010610)	Plantar pits (HP:0010612)
Odontogenic keratocysts of the jaw (HP:0010603)	Calcification of falx cerebri (HP:0005462)	Bifid ribs (HP:0000892)
Intellectual disability (HP:0001249)	Colon cancer (HP:0003003)	Large intestinal polyposis (HP:0030255)
Adenomatous colonic polyposis (HP:0005227)	Hamartomatous polyposis (HP:0004390)	Endometrial carcinoma (HP:0012114)
Ovarian neoplasm (HP:0100615)	Neoplasm of the ureter (HP:0100516)	Neoplasm of the skin (HP:0008069)
Biliary tract neoplasm (HP:0100574)	Neoplasm of the small intestine (HP:0100833)	Glioblastoma (HP:0100843)
Hamartoma (HP:0010566)	Breast carcinoma (HP:0003002)	Multiple trichilemmomata (HP:0012846)
Multiple lipomas (HP:0001012)	Hyperpigmented genitalia (HP:0030258)	Uterine leiomyoma (HP:0000131)
Hemangioma (HP:0001028)	Fibroadenoma of the breast (HP:0010619)	Arteriovenous malformation (HP:0100026)
Palmoplantar hyperkeratosis (HP:0000972)		

Young onset tumour syndromes (30781)

Paediatric congenital malformation-dysmorphism-tumour syndromes (30686)

Paediatric congenital malformation-dysmorphism-tumour syndromes phenotypes (40368)

Level 3 Title	Young onset tumour syndromes (30781)
Level 4 Title	Paediatric congenital malformation-dysmorphism-tumour syndromes (30686)
Phenotypes	Entries ordered left to right in table

Neoplasm (HP:0002664)	Small for gestational age (HP:0001518)	Large for gestational age (HP:0001520)
Failure to thrive (HP:0001508)	Short stature (HP:0004322)	Tall stature (HP:0000098)
Microcephaly (HP:0000252)	Macrocephaly (HP:0000256)	Abnormal facial shape (HP:0001999)
Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)	Hypermelanotic macule (HP:0001034)
Hypopigmentation of the skin (HP:0001010)	Cutaneous photosensitivity (HP:0000992)	Abnormality of the cardiovascular system (HP:0001626)
Abnormality of the musculature (HP:0003011)	Abnormality of the gastrointestinal tract (HP:0011024)	Abnormality of the liver (HP:0001392)
Abnormality of the respiratory system (HP:0002086)	Abnormality of the endocrine system (HP:0000818)	Abnormality of metabolism/homeostasis (HP:0001939)
Abnormality of blood and blood-forming tissues (HP:0001871)	Abnormality of the immune system (HP:0002715)	Abnormality of the skeletal system (HP:0000924)
Abnormality of the genitourinary system (HP:0000119)	Abnormality of prenatal development or birth (HP:0001197)	

Exceptionally young adult onset cancer (41892)

Exceptionally young adult onset cancer phenotypes (42113)

Level 3 Title	Young onset tumour syndromes (30781)
Level 4 Title	Exceptionally young adult onset cancer (41892)
Phenotypes	Entries ordered left to right in table

Breast carcinoma (HP:0003002)	Ovarian neoplasm (HP:0100615)	Colon cancer (HP:0003003)
Stomach cancer (HP:0012126)	Prostate cancer (HP:0012125)	Neoplasm of the lung (HP:0100526)
Thyroid carcinoma (HP:0002890)	Thyroid adenoma (HP:0000854)	Neoplasm of the parathyroid gland (HP:0100733)
Glioma (HP:0009733)	Neuroblastic tumors (HP:0004376)	Spinal cord tumor (HP:0010302)
Leukemia (HP:0001909)	Neoplasm of the skin (HP:0008069)	Melanoma (HP:0002861)
Sarcoma (HP:0100242)	Neoplasm of the liver (HP:0002896)	Renal neoplasm (HP:0009726)
Bladder neoplasm (HP:0009725)	Testicular neoplasm (HP:0010788)	Pheochromocytoma (HP:0002666)
Paraganglioma (HP:0002668)	Large intestinal polyposis (HP:0030255)	Small intestinal polyposis (HP:0030256)
Multiple gastric polyps (HP:0004394)	Lymphoma (HP:0002665)	Neoplasm of the thyroid gland (HP:0100031)
Adenoma sebaceum (HP:0009720)	Neoplasm of the eye (HP:0100012)	Neoplasm of the small intestine (HP:0100833)
Endometrial carcinoma (HP:0012114)	Pancreatic adenocarcinoma (HP:0006725)	Biliary tract neoplasm (HP:0100574)
Neoplasm of the ureter (HP:0100516)	Nephroblastoma (HP:0002667)	Glioblastoma (HP:0100843)
Medulloblastoma (HP:0002885)		

Exceptionally young adult onset cancer clinical tests (41894)

Level 3 Title	Young onset tumour syndromes (30781)
Level 4 Title	Exceptionally young adult onset cancer (41892)
Clinical Tests	Clinical test guidance: Biopsy refers to biopsy or resection pathology results of the proband's tumour(s) Entries ordered left to right in table

General Biopsy (33614.1)

Multiple Primaries (30782)

Multiple Tumours (30685)

Multiple Tumours phenotypes (40366)

Level 3 Title	Multiple Primaries (30782)
Level 4 Title	Multiple Tumours (30685)
Phenotypes	Entries ordered left to right in table

Breast carcinoma (HP:0003002)	Neoplasm of the lung (HP:0100526)	Ovarian neoplasm (HP:0100615)
Endometrial carcinoma (HP:0012114)	Prostate cancer (HP:0012125)	Testicular neoplasm (HP:0010788)
Colon cancer (HP:0003003)	Stomach cancer (HP:0012126)	Neoplasm of the small intestine (HP:0100833)
Large intestinal polyposis (HP:0030255)	Neoplasm of the liver (HP:0002896)	Pancreatic adenocarcinoma (HP:0006725)
Biliary tract neoplasm (HP:0100574)	Renal neoplasm (HP:0009726)	Neoplasm of the ureter (HP:0100516)
Bladder neoplasm (HP:0009725)	Nephroblastoma (HP:0002667)	Glioblastoma (HP:0100843)
Neuroblastic tumors (HP:0004376)	Medulloblastoma (HP:0002885)	Glioma (HP:0009733)
Spinal cord tumor (HP:0010302)	Leukemia (HP:0001909)	Lymphoma (HP:0002665)
Neoplasm of the thyroid gland (HP:0100031)	Neoplasm of the parathyroid gland (HP:0100733)	Pheochromocytoma (HP:0002666)
Paraganglioma (HP:0002668)	Sarcoma (HP:0100242)	Melanoma (HP:0002861)
Neoplasm of the skin (HP:0008069)	Adenoma sebaceum (HP:0009720)	Neoplasm of the eye (HP:0100012)

Ultra-rare disorders (30783)

Undescribed disorders (30784)

Ultra-rare undescribed monogenic disorders (30785)

Ultra-rare undescribed monogenic disorders phenotypes (30790)

Level 3 Title	Undescribed disorders (30784)
Level 4 Title	Ultra-rare undescribed monogenic disorders (30785)
Phenotypes	Entries ordered left to right in table

Abnormality of the cardiovascular system (HP:0001626)	Abnormality of the nervous system (HP:0000707)	Abnormality of the musculature (HP:0003011)
Abnormality of the gastrointestinal tract (HP:0011024)	Abnormality of the liver (HP:0001392)	Abnormality of the respiratory system (HP:0002086)
Abnormality of the endocrine system (HP:0000818)	Abnormality of metabolism/homeostasis (HP:0001939)	Abnormality of blood and blood-forming tissues (HP:0001871)
Abnormality of the immune system (HP:0002715)	Abnormality of the skeletal system (HP:0000924)	Abnormality of the integument (HP:0001574)
Abnormality of the genitourinary system (HP:0000119)	Abnormality of prenatal development or birth (HP:0001197)	Growth abnormality (HP:0001507)
Abnormality of the ear (HP:0000598)	Abnormality of the eye (HP:0000478)	

Neonatal or paediatric intensive care admission with a likely monogenic disease (38558)

Neonatal or paediatric intensive care admission with a likely monogenic disease phenotypes (38565)

Level 3 Title	Multi-system groups (38589)
Level 4 Title	Neonatal or paediatric intensive care admission with a likely monogenic disease (38558)
Phenotypes	Entries ordered left to right in table

Small for gestational age (HP:0001518)	Large for gestational age (HP:0001520)	Failure to thrive (HP:0001508)
Short stature (HP:0004322)	Tall stature (HP:0000098)	Microcephaly (HP:0000252)
Macrocephaly (HP:0000256)	Abnormal facial shape (HP:0001999)	Intellectual disability (HP:0001249)
Global developmental delay (HP:0001263)	Hyperpigmented streaks (HP:0007572)	Hypermelanotic macule (HP:0001034)
Macular hypopigmented whorls, streaks, and patches (HP:0005593)	Hypopigmentation of the skin (HP:0001010)	Cutaneous photosensitivity (HP:0000992)
Abnormality of the cardiovascular system (HP:0001626)	Abnormality of the musculature (HP:0003011)	Abnormality of the gastrointestinal tract (HP:0011024)
Abnormality of the liver (HP:0001392)	Abnormality of the respiratory system (HP:0002086)	Abnormality of the endocrine system (HP:0000818)
Abnormality of metabolism/homeostasis (HP:0001939)	Abnormality of blood and blood-forming tissues (HP:0001871)	Abnormality of the immune system (HP:0002715)
Abnormality of the skeletal system (HP:0000924)	Abnormality of the genitourinary system (HP:0000119)	Abnormality of prenatal development or birth (HP:0001197)

Neonatal or paediatric intensive care admission with a likely monogenic disease clinical tests (38564)

Level 3 Title	Multi-system groups (38589)
Level 4 Title	Neonatal or paediatric intensive care admission with a likely monogenic disease (38558)
Clinical Tests	NB. Clinical test guidance: General imaging diagnostics refers to medical photographs where relevant and other relevant imaging Entries ordered left to right in table

General Imaging Diagnostics (33633.1)

Single autosomal recessive mutation in rare disease (38672)

Ultra-rare undescribed monogenic disorders phenotypes (30790)

<table>
<tr><td>Level 3 Title</td><td>Multi-system groups (38589)</td></tr>
<tr><td>Level 4 Title</td><td>Single autosomal recessive mutation in rare disease (38672)</td></tr>
<tr><td>Phenotypes</td><td>Entries ordered left to right in table
<table>
<tr><td>Abnormality of the cardiovascular system (HP:0001626)</td><td>Abnormality of the nervous system (HP:0000707)</td><td>Abnormality of the musculature (HP:0003011)</td></tr>
<tr><td>Abnormality of the gastrointestinal tract (HP:0011024)</td><td>Abnormality of the liver (HP:0001392)</td><td>Abnormality of the respiratory system (HP:0002086)</td></tr>
<tr><td>Abnormality of the endocrine system (HP:0000818)</td><td>Abnormality of metabolism/homeostasis (HP:0001939)</td><td>Abnormality of blood and blood-forming tissues (HP:0001871)</td></tr>
<tr><td>Abnormality of the immune system (HP:0002715)</td><td>Abnormality of the skeletal system (HP:0000924)</td><td>Abnormality of the integument (HP:0001574)</td></tr>
<tr><td>Abnormality of the genitourinary system (HP:0000119)</td><td>Abnormality of prenatal development or birth (HP:0001197)</td><td>Growth abnormality (HP:0001507)</td></tr>
<tr><td>Abnormality of the ear (HP:0000598)</td><td>Abnormality of the eye (HP:0000478)</td><td></td></tr>
</table></td></tr>
</table>

Undiagnosed monogenic disorder seen in a specialist genetics clinic (42193)

Ultra-rare undescribed monogenic disorders phenotypes (30790)

Level 3 Title	Multi-system groups (38589)
Level 4 Title	Undiagnosed monogenic disorder seen in a specialist genetics clinic (42193)
Phenotypes	Entries ordered left to right in table

Abnormality of the cardiovascular system (HP:0001626)	Abnormality of the nervous system (HP:0000707)	Abnormality of the musculature (HP:0003011)
Abnormality of the gastrointestinal tract (HP:0011024)	Abnormality of the liver (HP:0001392)	Abnormality of the respiratory system (HP:0002086)
Abnormality of the endocrine system (HP:0000818)	Abnormality of metabolism/homeostasis (HP:0001939)	Abnormality of blood and blood-forming tissues (HP:0001871)
Abnormality of the immune system (HP:0002715)	Abnormality of the skeletal system (HP:0000924)	Abnormality of the integument (HP:0001574)
Abnormality of the genitourinary system (HP:0000119)	Abnormality of prenatal development or birth (HP:0001197)	Growth abnormality (HP:0001507)
Abnormality of the ear (HP:0000598)	Abnormality of the eye (HP:0000478)	

Genomic medicine service indications (82157)

Whole genome sequencing indications (82159)

GMS R14 Acutely unwell infants with a likely monogenic disorder (82160)

Free HPO entry

GMS R27 Congenital malformation and dysmorphism syndromes - likely monogenic (82161)

Free HPO entry

GMS R69 Floppy infant with a likely central cause (82162)

GMS R69 Floppy infant with a likely central cause phenotypes (82271)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R69 Floppy infant with a likely central cause (82162)
Phenotypes	Entries ordered left to right in table

Generalized hypotonia (HP:0001290)	Abnormal facial shape (HP:0001999)	Respiratory insufficiency (HP:0002093)
Feeding difficulties (HP:0011968)	Lethargy (HP:0001254)	

GMS R29 Moderate, severe or profound intellectual disability (82163)

GMS R29 Moderate, severe or profound intellectual disability phenotypes (82272)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R29 Moderate, severe or profound intellectual disability (82163)
Phenotypes	Entries ordered left to right in table

Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)	Global developmental delay (HP:0001263)

GMS R89 Ultra-rare and atypical monogenic disorders (82164)

Free HPO entry

GMS R100 Rare syndromic craniosynostosis or isolated multisuture synostosis (82165)

GMS R100 Rare syndromic craniosynostosis or isolated multisuture synostosis phenotypes (82274)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R100 Rare syndromic craniosynostosis or isolated multisuture synostosis (82165)
Phenotypes	Entries ordered left to right in table

Multiple suture craniosynostosis (HP:0011324)	Unicoronal synostosis (HP:0011315)	Bicoronal synostosis (HP:0011318)
Sagittal craniosynostosis (HP:0004442)	Metopic synostosis (HP:0011330)	Unilambdoid synostosis (HP:0011320)
Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)	Global developmental delay (HP:0001263)

GMS R104 Skeletal dysplasia (82166)

GMS R104 Skeletal dysplasia phenotypes (82275)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R104 Skeletal dysplasia (82166)
Phenotypes	Entries ordered left to right in table

Skeletal dysplasia (HP:0002652)	Short stature (HP:0004322)	Intellectual disability (HP:0001249)
Abnormal facial shape (HP:0001999)	Global developmental delay (HP:0001263)	

GMS R143 Neonatal diabetes (82167)

GMS R143 Neonatal diabetes phenotypes (82276)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R143 Neonatal diabetes (82167)
Phenotypes	Entries ordered left to right in table

Neonatal insulin-dependent diabetes mellitus (HP:0000857)	Transient neonatal diabetes mellitus (HP:0008255)

GMS R98 Likely inborn error of metabolism - targeted testing not possible (82168)

Free HPO entry

GMS R83 Arthrogryposis (82185)

GMS R83 Arthrogryposis phenotypes (82277)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R83 Arthrogryposis (82185)
Phenotypes	Entries ordered left to right in table

Arthrogryposis multiplex congenita (HP:0002804)	Distal arthrogryposis (HP:0005684)

GMS R84 Cerebellar anomalies (82169)

GMS R84 Cerebellar anomalies phenotypes (82278)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R84 Cerebellar anomalies (82169)
Phenotypes	Entries ordered left to right in table

Cerebellar hypoplasia (HP:0001321)	Cerebellar atrophy (HP:0001272)	Dandy-Walker malformation (HP:0001305)
Olivopontocerebellar hypoplasia (HP:0006955)	Intellectual disability (HP:0001249)	Global developmental delay (HP:0001263)

GMS R87 Cerebral malformation (82170)

GMS R87 Cerebral malformation phenotypes (82279)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R87 Cerebral malformation (82170)
Phenotypes	Entries ordered left to right in table

Lissencephaly (HP:0001339)	Polymicrogyria (HP:0002126)	Heterotopia (HP:0002282)
Pachygyria (HP:0001302)	Schizencephaly (HP:0010636)	Cortical dysplasia (HP:0002539)
Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)	Global developmental delay (HP:0001263)
Seizures (HP:0001250)		

GMS R61 Childhood onset hereditary spastic paraplegia (82171)

GMS R61 Childhood onset hereditary spastic paraplegia phenotypes (82280)

<table>
<tr><td>Level 3 Title</td><td>Whole genome sequencing indications (82159)</td></tr>
<tr><td>Level 4 Title</td><td>GMS R61 Childhood onset hereditary spastic paraplegia (82171)</td></tr>
<tr><td>Phenotypes</td><td>Entries ordered left to right in table
<table><tr><td>Spasticity (HP:0001257)</td><td>Intellectual disability (HP:0001249)</td><td>Abnormal facial shape (HP:0001999)</td></tr><tr><td>Global developmental delay (HP:0001263)</td><td>Ataxia (HP:0001251)</td><td>Seizures (HP:0001250)</td></tr></table></td></tr>
</table>

GMS R109 Childhood onset leukodystrophy (82172)

GMS R109 Childhood onset leukodystrophy phenotypes (82281)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R109 Childhood onset leukodystrophy (82172)
Phenotypes	Entries ordered left to right in table

Diffuse white matter abnormalities (HP:0007204)	Focal white matter lesions (HP:0007042)	Leukoencephalopathy (HP:0002352)
Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)	Global developmental delay (HP:0001263)
Spasticity (HP:0001257)	Seizures (HP:0001250)	

GMS R59 Early onset or syndromic epilepsy (82173)

GMS R59 Early onset or syndromic epilepsy phenotypes (82282)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R59 Early onset or syndromic epilepsy (82173)
Phenotypes	Entries ordered left to right in table

Generalized seizures (HP:0002197)	Focal seizures (HP:0007359)	Epileptic spasms (HP:0011097)
Infantile encephalopathy (HP:0007105)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Global developmental delay (HP:0001263)		

GMS R54 Hereditary ataxia with onset in adulthood (82174)

GMS R54 Hereditary ataxia with onset in adulthood phenotypes (82283)

<table>
<tr><td>Level 3 Title</td><td>Whole genome sequencing indications (82159)</td></tr>
<tr><td>Level 4 Title</td><td>GMS R54 Hereditary ataxia with onset in adulthood (82174)</td></tr>
<tr><td>Phenotypes</td><td>Entries ordered left to right in table
<table><tr><td>Ataxia (HP:0001251)</td><td>Abnormality of eye movement (HP:0000496)</td><td>Spasticity (HP:0001257)</td></tr><tr><td>Dystonia (HP:0001332)</td><td>Cognitive impairment (HP:0100543)</td><td>Parkinsonism (HP:0001300)</td></tr></table></td></tr>
</table>

GMS R55 Hereditary ataxia with onset in childhood (82175)

GMS R55 Hereditary ataxia with onset in childhood phenotypes (82284)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R55 Hereditary ataxia with onset in childhood (82175)
Phenotypes	Entries ordered left to right in table

Ataxia (HP:0001251)	Abnormality of eye movement (HP:0000496)	Spasticity (HP:0001257)
Dystonia (HP:0001332)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Global developmental delay (HP:0001263)	Peripheral neuropathy (HP:0009830)	Optic atrophy (HP:0000648)

GMS R85 Holoprosencephaly - NOT chromosomal (82176)

GMS R85 Holoprosencephaly - NOT chromosomal phenotypes (82285)

<table>
<tr><td>Level 3 Title</td><td>Whole genome sequencing indications (82159)</td></tr>
<tr><td>Level 4 Title</td><td>GMS R85 Holoprosencephaly - NOT chromosomal (82176)</td></tr>
<tr><td>Phenotypes</td><td>Entries ordered left to right in table
<table><tr><td>Holoprosencephaly (HP:0001360)</td><td>Intellectual disability (HP:0001249)</td><td>Abnormal facial shape (HP:0001999)</td></tr><tr><td>Global developmental delay (HP:0001263)</td><td></td><td></td></tr></table></td></tr>
</table>

GMS R86 Hydrocephalus (82177)

GMS R86 Hydrocephalus phenotypes (82286)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R86 Hydrocephalus (82177)
Phenotypes	Entries ordered left to right in table

Hydrocephalus (HP:0000238)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Global developmental delay (HP:0001263)		

GMS R381 Other rare neuromuscular disorders (82178)

GMS R381 Other rare neuromuscular disorders phenotypes (82287)

<table>
<tr><td>Level 3 Title</td><td>Whole genome sequencing indications (82159)</td></tr>
<tr><td>Level 4 Title</td><td>GMS R381 Other rare neuromuscular disorders (82178)</td></tr>
<tr><td>Phenotypes</td><td>Entries ordered left to right in table
<table>
<tr><td>Myopathy (HP:0003198)</td><td>Muscular dystrophy (HP:0003560)</td><td>Fatigable weakness (HP:0003473)</td></tr>
<tr><td>Intellectual disability (HP:0001249)</td><td>Abnormal facial shape (HP:0001999)</td><td>Global developmental delay (HP:0001263)</td></tr>
<tr><td>Myotonia (HP:0002486)</td><td></td><td></td></tr>
</table>
</td></tr>
</table>

GMS R88 Severe microcephaly (82179)

GMS R88 Severe microcephaly phenotypes (82288)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R88 Severe microcephaly (82179)
Phenotypes	Entries ordered left to right in table

Congenital microcephaly (HP:0011451)	Postnatal microcephaly (HP:0005484)	Progressive microcephaly (HP:0000253)
Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)	Global developmental delay (HP:0001263)

GMS R193 Cystic renal disease (82180)

GMS R193 Cystic renal disease phenotypes (82289)

Level 3 Title	Whole genome sequencing indications (82159)
Level 4 Title	GMS R193 Cystic renal disease (82180)
Phenotypes	Entries ordered left to right in table

Multiple renal cysts (HP:0005562)	Nephronophthisis (HP:0000090)	Hepatic cysts (HP:0001407)
Cerebral aneurysm (HP:0004944)	Intellectual disability (HP:0001249)	Abnormal facial shape (HP:0001999)
Global developmental delay (HP:0001263)		