The 100,000 Genomes Project – Current Rare Disease List v 1.9.0

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1. Rare disease list

Diseases live for recruitment 240

Category	Subcategory	Disease	Status	Non-penetrant condition	Unaffected individual recruitment*
Cardiovascular disorders (10950)	Arteriopathies (33332)	Familial cerebral small vessel disease (36469)	Live	Yes	Typically not recruited
		Familial Hypercholesterolaemia (33666)	Live	Yes	Typically not recruited
		Severe hypertriglyceridaemia (42185)	Live	Yes	Typically not recruited
	Connective Tissues Disorders and Aortopathies (10951)	Familial Thoracic Aortic Aneurysm Disease (11021)	Live	Yes	Typically not recruited
	Cardiac arrhythmia (10952)	Brugada syndrome (11022)	Live	Yes	Typically not recruited
		Long QT syndrome (11023)	Live	Yes	Typically not recruited
		Catecholaminergic Polymorphic Ventricular Tachycardia (11024)	Live	Yes	Typically not recruited
		Unexplained sudden death in the young (38566)	Live	Yes	Recruited in paediatric cases, typically not recruited in adult cases
		Idiopathic ventricular fibrillation (42161)	Live	Yes	Typically not recruited
		Short QT syndrome (55487)	Live	Yes	Typically not recruited
	Cardiomyopathy (10953)	Arrhythmogenic Right Ventricular Cardiomyopathy (11025)	Live	Yes	Typically not recruited
		Left Ventricular Noncompaction Cardiomyopathy (15044)	Live	Yes	Typically not recruited
		Dilated Cardiomyopathy (31340)	Live	Yes	Typically not recruited
		Dilated Cardiomyopathy and conduction defects (11027)	Live	Yes	Typically not recruited
		Hypertrophic Cardiomyopathy (11028)	Live	Yes	Typically not recruited
	Congenital heart disease (10954)	Familial congenital heart disease (42212)	Live	Yes	Typically not recruited

		Syndromic congenital heart disease (42213)	Live	No	Should be recruited
	Lymphatic disorders (33334)	Meige disease (34328)	Live	Yes	Typically not recruited
		Milroy disease (37604)	Live	Yes	Typically not recruited
		Lymphoedema distichiasis (37612)	Live	Yes	Typically not recruited
		Lipoedema disease (55456)	Live	Yes	Typically not recruited
		Primary lymphoedema (55517)	Live	Yes	Typically not recruited
	Pulmonary heart disease (55662)	Pulmonary arterial hypertension (55499)	Live	Yes	Typically not recruited
Ciliopathies (10963)	Congenital malformations caused by ciliopathies (15091)	Bardet-Biedl Syndrome (11046)	Live	No	Should be recruited
		Joubert syndrome (36478)	Live	No	Should be recruited
		Rare multisystem ciliopathy disorders (36488)	Live	No	Should be recruited
	Respiratory ciliopathies (15092)	Primary ciliary dyskinesia (11047)	Live	No	Should be recruited
		Non-CF bronchiectasis (11048)	Live	Yes	Typically not recruited
Dermatological disorders (10956)	Atopy (15084)	Severe multi-system atopic disease with high IgE (15085)	Live	Yes	Typically not recruited
	Autoimmune skin disorders (33336)	Generalised pustular psoriasis (33646)	Live	Yes	Typically not recruited
	Ectodermal dysplasias (33338)	Ectodermal dysplasia without a known gene mutation (33699)	Live	No	Should be recruited
	Ichthyoses (33340)	Autosomal recessive congenital ichthyosis (33700)	Live	No	Should be recruited
	Keratodermas (33342)	Palmoplantar keratoderma and erythrokeratodermas (33701)	Live	Yes	Typically not recruited
		Familial disseminated superficial actinic porokeratosis (37644)	Live	Yes	Typically not recruited
	Neurocutaneous disorders (33344)	Undiagnosed neurocutaneous disorders (33686)	Live	No	Should be recruited
	Skin adnexa disorders (36587)	Familial cicatricial alopecia (36588)	Live	Yes	Typically not recruited

		Familial hidradenitis suppurativa (41844)	Live	Yes	Typically not recruited
		Non-syndromic hypotrichosis (36849)	Live	Yes	Typically not recruited
	Skin fragility disorders (33346)	Epidermolysis bullosa (33684)	Live	No	Should be recruited
		Peeling skin syndrome (36540)	Live	No	Should be recruited
	Sun-exposure related conditions (10958)	Erythropoietic protoporphyria, mild variant (11037)	Live	Yes	Typically not recruited
		Hydroa vacciniforme (15083)	Live	Yes	Typically not recruited
Dysmorphic and congenital abnormality syndromes (10959)	Kabuki (28664)	Kabuki syndrome (10960)	Live	No	Should be recruited
	RASopathies (10961)	Noonan syndrome (11039)	Live	Yes	Should be recruited
		Noonan syndrome plus other features (11040)	Live	Yes	Should be recruited
		Cardio-facio-cutaneous syndrome (11041)	Live	Yes	Should be recruited
		LEOPARD syndrome (11042)	Live	Yes	Should be recruited
		Costello syndrome (11043)	Live	No	Should be recruited
		Legius syndrome (11044)	Live	Yes	Should be recruited
	Balanced translocations (10962)	Balanced translocations with an unusual phenotype (11045)	Live	No	Should be recruited
	Limb disorders (15087)	VACTERL-like phenotypes (10964)	Live	Yes	Should be recruited
	DNA repair disorders (10965)	Cockayne syndrome (36497)	Live	No	Should be recruited
		Non-Fanconi anaemia (11050)	Live	No	Should be recruited
		Xeroderma Pigmentosum-like disorders (15089)	Live	No	Should be recruited
		Primary Microcephaly - Microcephalic Dwarfism Spectrum (36505)	Live	No	Should be recruited
	Autophagy disorders (10966)	Vici Syndrome and other autophagy disorders (11051)	Live	No	Should be recruited
	Dysmorphic disorders (36595)	Coarse facial features including Coffin-Siris-like disorders (36596)	Live	No	Should be recruited

		Familial non-syndromic cleft lip and or familial cleft palate (37565)	Live	Yes	Typically not recruited
		Syndromic cleft lip and or cleft palate (37573)	Live	No	Should be recruited
		PHACE(S) syndrome (37578)	Live	No	Should be recruited
		Radial dysplasia (37636)	Live	Yes	Should be recruited
	Fetal disorders (38586)	Fetal hydrops (37586)	Live	No	Should be recruited
		Unexplained monogenic fetal disorders (38665)	Live	No	Should be recruited
Endocrine disorders (10967)	Adrenal disorders (10969)	Congenital adrenal hypoplasia (11053)	Live	No	Should be recruited
	Disorders of calcium homeostasis (10970)	Familial or syndromic hypoparathyroidism (11054)	Live	Yes	Typically not recruited
	Gonadal and sex development disorders (36923)	Disorders of sex development (36852)	Live	No	Should be recruited
		Early onset familial premature ovarian insufficiency (36851)	Live	Yes	Typically not recruited
	Growth hormone disorders (10971)	IUGR and IGF abnormalities (11057)	Live	Yes	Should be recruited
	Hypothalamic and pituitary disorders (42204)	Idiopathic hypogonadotropic hypogonadism (41827)	Live	Yes	Typically not recruited
	Obesity syndromes (10973)	Significant early-onset obesity with or without other endocrine features and short stature (11060)	Live	Yes	Should be recruited
	Rare subtypes of diabetes (15099)	Familial young-onset non-insulin-dependent diabetes (15103)	Live	Yes	Unaffected family members should only be recruited if they have had testing to rule out diabetes (e.g. normal HbA1c) at or over the age of 40.
		Hyperinsulinism (15105)	Live	No	Should be recruited

		Neonatal diabetes (diagnosed less than 6 months) (30553)	Live	No	Should be recruited
		Diabetes with additional phenotypes suggestive of a monogenic aetiology (30559)	Live	Yes	Should be recruited
		Insulin resistance (including lipodystrophy) (30561)	Live	Yes	Should be recruited
		Multi-organ autoimmune diabetes (30563)	Live	Yes	Typically not recruited
	Thyroid disorders (42208)	Congenital hypothyroidism (41908)	Live	No	Should be recruited
		Resistance to thyroid hormone (41916)	Live	No	Should be recruited
Gastroenterological disorders (38581)	Gastrointestinal disorders (38582)	Infantile enterocolitis and monogenic inflammatory bowel disease (37490)	Live	No	Should be recruited
		Gastrointestinal epithelial barrier disorders (37772)	Live	No	Should be recruited
		Non-syndromic familial congenital anorectal malformations (41868)	Live	Yes	Typically not recruited
		Early onset or familial intestinal pseudo obstruction (41876)	Live	Yes	Typically not recruited
		Familial Hirschsprung Disease (55463)	Live	Yes	Typically not recruited
	Liver disease (55663)	Ductal plate malformation (55469)	Live	Yes	Should be recruited
		Neonatal cholestasis (71744)	Live	No	Should be recruited
Growth disorders (10974)	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)	Classical Beckwith-Wiedemann syndrome (11063)	Live	No	Should be recruited
		Atypical Beckwith-Wiedemann syndrome (11064)	Live	No	Should be recruited
		Simpson-Golabi-Behmel syndrome (11065)	Live	No	Should be recruited
		Sotos syndrome (11066)	Live	No	Should be recruited
		Weaver syndrome (11067)	Live	No	Should be recruited
	Growth restriction (38585)	Silver Russell syndrome (37553)	Live	No	Should be recruited

Haematological and immunological disorders (10977)	Anaemias and red cell disorders (10979)	Congenital anaemias (11075)	Live	No	Should be recruited
		Hereditary erythrocytosis (55505)	Live	No	Should be recruited
	Primary immunodeficiency disorders (10978)	Primary immunodeficiency (55674)	Live	Yes	Should be recruited
	Haemostasis disorders (55664)	Inherited bleeding and or platelet disorders (55475)	Live	Yes	Should be recruited
		Monogenic venous thrombosis (55523)	Live	Yes	Typically not recruited
	Myeloid and marrow failure disorders (71739)	Cytopenia and pancytopenia (71752)	Live	No	Should be recruited
Hearing and ear disorders (10980)	Non-syndromic hearing loss (10981)	Congenital hearing impairment (11076)	Live	No	Should be recruited
		Auditory Neuropathy Spectrum Disorder (30607)	Live	No	Should be recruited
		Autosomal dominant deafness (36848)	Live	Yes	Typically not recruited
	Deafness and congenital structural abnormalities (10982)	Bilateral microtia (11077)	Live	No	Should be recruited
		Familial hemifacial microsomia (37649)	Live	No	Should be recruited
		Ear malformations with hearing impairment (37657)	Live	No	Should be recruited
	Other hearing and ear disorders (71738)	Familial Meniere Disease (71748)	Live	Yes	Typically not recruited
Infectious diseases (42209)	Bacterial disorders (42210)	Disseminated non-tuberculous mycobacterial infection (41932)	Live	Yes	Should be recruited
	Sepsis (55671)	GAinS study (55665)	Live	Yes	Typically not recruited
Metabolic disorders (10983)	Specific metabolic abnormalities (10984)	Ketotic hypoglycaemia (11080)	Live	No	Should be recruited
		Lactic acidosis (11081)	Live	No	Should be recruited
		Cerebral folate deficiency (11083)	Live	No	Should be recruited
		Undiagnosed metabolic disorders (37620)	Live	No	Should be recruited
		Congenital disorders of glycosylation (37628)	Live	No	Should be recruited
	Urea Cycle disorders (15108)	Hyperammonaemia (11079)	Live	No	Should be recruited

	Lysosomal storage disorders (10985)	Mucopolysaccharideosis, Gaucher, Fabry (11084)	Live	No	Should be recruited
	Mitochondrial (10986)	Mitochondrial disorders (11085)	Live	No	Should be recruited
	Peroxisomal disorders (10987)	Peroxisomal biogenesis disorders (11086)	Live	No	Should be recruited
		Other peroxisomal disorders (15109)	Live	No	Should be recruited
Neurology and neurodevelopmental disorders (10988)	Motor Disorders of the CNS (10989)	Cerebellar hypoplasia (36512)	Live	No	Should be recruited
		Hereditary ataxia (11087)	Live	Yes	Typically not recruited
		Early onset dystonia (11088)	Live	No	Should be recruited
		Hereditary spastic paraplegia (11089)	Live	Yes	Typically not recruited
		Neurotransmitter disorders (37779)	Live	No	Should be recruited
		Structural basal ganglia disorders (37786)	Live	No	Should be recruited
	Inherited Epilepsy Syndromes (10990)	Genetic Epilepsies with Febrile Seizures Plus (11091)	Live	No	Should be recruited
		Familial Genetic Generalised Epilepsies (11092)	Live	Yes	Typically not recruited
		Familial Focal Epilepsies (11093)	Live	Yes	Typically not recruited
		Epileptic encephalopathy (11094)	Live	No	Should be recruited
		Epilepsy plus other features (41924)	Live	Yes	Should be recruited
	Motor and Sensory Disorders of the PNS (10991)	Charcot-Marie-Tooth disease (15111)	Live	Yes	Typically not recruited
		Paediatric motor neuronopathies (11099)	Live	No	Should be recruited
		Pain channelopathies (82148)	Live	Yes	Typically not recruited
	Neurodegenerative disorders (10992)	Early onset and familial Parkinson's Disease (11100)	Live	Yes	Typically not recruited
		Complex Parkinsonism (includes pallido-pyramidal syndromes) (15112)	Live	Yes	Typically not recruited
		Early onset dementia (15113)	Live	Yes	Typically not recruited

		Amyotrophic lateral sclerosis or motor neuron disease (15114)	Live	Yes	Typically not recruited
	Neurodevelopmental disorders (10993)	Classical tuberous sclerosis (11101)	Live	Yes	Should be recruited
		Intellectual disability (11102)	Live	No	Should be recruited
		Holoprosencephaly (36519)	Live	Yes	Should be recruited
		Rhomboencephalosynapsis (36603)	Live	No	Should be recruited
		Malformations of cortical development (36526)	Live	No	Should be recruited
		Fetal structural CNS abnormalities (36850)	Live	No	Should be recruited
		Pontine tegmental cap dysplasia (55493)	Live	No	Should be recruited
	Neuromuscular disorders (10994)	Congenital muscular dystrophy (15135)	Live	No	Should be recruited
		Congenital myopathy (11103)	Live	No	Should be recruited
		Congenital myaesthenia (15136)	Live	No	Should be recruited
		Rhabdomyolysis and metabolic muscle disorders (15137)	Live	Yes	Typically not recruited
		Distal myopathies (11104)	Live	No	Should be recruited
		Arthrogryposis (15138)	Live	No	Should be recruited
		Limb girdle muscular dystrophy (11106)	Live	No	Should be recruited
	Channelopathies (11097)	Skeletal Muscle Channelopathies (15139)	Live	No	Should be recruited
		Brain channelopathy (15140)	Live	Yes	Typically not recruited
	Sleep disorders (10995)	Kleine-Levin syndrome and other inherited sleep disorders (11108)	Live	Yes	Typically not recruited
	Cerebrovascular disorders (36610)	Moyamoya disease (36611)	Live	No	Should be recruited
		Vein of Galen malformation (42174)	Live	Yes	Should be recruited
	Parenchymal brain disorders (36618)	Intracerebral calcification disorders (36619)	Live	No	Should be recruited
	White matter disorders (36626)	Inherited white matter disorders (36627)	Live	No	Should be recruited

Ophthalmological disorders (10996)	Anterior segment abnormalities (10997)	Corneal abnormalities (11110)	Live	No	Should be recruited
		Glaucoma (developmental) (11111)	Live	No	Should be recruited
		Cataracts (11112)	Live	No	Should be recruited
	Posterior segment abnormalities (10998)	Inherited optic neuropathies (11114)	Live	No	Should be recruited
		Rod-cone dystrophy (29268)	Live	No	Should be recruited
		Rod Dysfunction Syndrome (29269)	Live	No	Should be recruited
		Cone Dysfunction Syndrome (29270)	Live	No	Should be recruited
		Inherited macular dystrophy (29271)	Live	No	Should be recruited
		Leber Congenital Amaurosis or Early-Onset Severe Retinal Dystrophy (29272)	Live	No	Should be recruited
		Developmental macular and foveal dystrophy (29273)	Live	No	Should be recruited
		Familial exudative vitreoretinopathy (41900)	Live	No	Should be recruited
	Ocular malformations (10999)	Anophthalmia or microphthamia (11115)	Live	Yes	Should be recruited
		Ocular coloboma (15141)	Live	Yes	Should be recruited
	Ocular movement disorders (33350)	Infantile nystagmus (33662)	Live	No	Should be recruited
Psychiatric disorders (71735)	Schizophrenia and other psychotic disorders (71736)	Schizophrenia plus additional features (71740)	Live	Yes	Typically not recruited
	Feeding and eating disorders (71737)	Severe familial anorexia (29278)	Live	Yes	Typically not recruited
Renal and urinary tract disorders (11000)	Syndromes with prominent renal abnormalities (11001)	Proteinuric renal disease (30732)	Live	Yes	Typically not recruited
		Familial haematuria (30733)	Live	Yes	Typically not recruited
		Atypical haemolytic uraemic syndrome (33489)	Live	Yes	Typically not recruited
		Primary membranoproliferative glomerulonephritis (55481)	Live	Yes	Typically not recruited

		Familial IgA nephropathy and IgA vasculitis (82147)	Live	Yes	Typically not recruited
	Structural renal and urinary tract disease (11003)	Cystic kidney disease (11120)	Live	Yes	Typically not recruited
		Congenital Anomaly of the Kidneys and Urinary Tract (CAKUT) (29277)	Live	Yes	Typically not recruited
	Disorders of function (11004)	Renal tubular acidosis (11123)	Live	Yes	Typically not recruited
		Renal tract calcification (or Nephrolithiasis or nephrocalcinosis) (11124)	Live	Yes	Typically not recruited
		Extreme early-onset hypertension (15142)	Live	Yes	Typically not recruited
		Unexplained kidney failure in young people (36855)	Live	Yes	Typically not recruited
Respiratory disorders (33353)	Interstitial lung disorders (33354)	Familial pulmonary fibrosis (33671)	Live	Yes	Typically not recruited
	Vascular lung disorders (33355)	Hereditary haemorrhagic telangiectasia (33674)	Live	Yes	Typically not recruited
		Familial and multiple pulmonary arteriovenous malformations (33677)	Live	Yes	Typically not recruited
	Structural lung disorders (42203)	Familial primary spontaneous pneumothorax (41819)	Live	Yes	Typically not recruited
Rheumatological disorders (11009)	Multi-system inflammatory or autoimmune disorders (11008)	Periodic fever syndromes and amyloidosis (11127)	Live	No	Should be recruited
		Juvenile dermatomyositis (29219)	Live	Yes	Typically not recruited
	Connective tissues disorders (36930)	Kyphoscoliotic Ehlers-Danlos syndrome (36853)	Live	No	Should be recruited
		Classical Ehlers-Danlos Syndrome (41860)	Live	No	Should be recruited
Skeletal disorders (11005)	Skeletal dysplasias (11007)	Multiple Epiphyseal Dysplasia (11125)	Live	No	Should be recruited
		Chondrodysplasia punctata (15143)	Live	No	Should be recruited
		Thoracic dystrophies (11126)	Live	No	Should be recruited
		Stickler syndrome (11129)	Live	Yes	Should be recruited
		Osteogenesis imperfecta (30627)	Live	No	Should be recruited
		Unexplained skeletal dysplasia (36854)	Live	No	Should be recruited

		Amelogenesis imperfecta (55449)	Live	Yes	Typically not recruited
	Craniosynostosis (30775)	Craniosynostosis syndromes (11006)	Live	Yes	Should be recruited
	Choanal anomalies (31500)	Choanal atresia (11078)	Live	Yes	Should be recruited
Tumour syndromes (11012)	Breast and endocrine (11013)	Familial breast and or ovarian cancer (11131)	Live	Yes	Typically not recruited
		Multiple endocrine tumours (11132)	Live	Yes	Typically not recruited
		Neuro-endocrine Tumours- PCC and PGL (11133)	Live	Yes	Typically not recruited
		Parathyroid cancer (30611)	Live	Yes	Typically not recruited
		Inherited non-medullary thyroid cancer (41884)	Live	Yes	Typically not recruited
	GI tract (11014)	Familial colon cancer (11135)	Live	Yes	Typically not recruited
		Multiple bowel polyps (30615)	Live	Yes	Typically not recruited
		Peutz-Jeghers syndrome (36533)	Live	Yes	Typically not recruited
	Muscle and nerve (11015)	Familial rhabdomyosarcoma or sarcoma (11138)	Live	Yes	Typically not recruited
		Familial tumour syndromes of the central and peripheral nervous system (30619)	Live	Yes	Typically not recruited
		Neurofibromatosis Type 1 (38874)	Live	No	Where the NF1 gene has not been tested before recruitment, participants should be recruited initially as singletons. In families where analysis as a singleton does not lead to identification of the

					underlying causative mutations, recruitment of additional affected family members is encouraged.
	Skin (11016)	Genodermatoses with malignancies (30623)	Live	Yes	Typically not recruited
	Young onset tumour syndromes (30781)	Paediatric congenital malformation-dysmorphism-tumour syndromes (30686)	Live	No	Should be recruited
		Exceptionally young adult onset cancer (41892)	Live	Yes	Typically not recruited
	Multiple Primaries (30782)	Multiple Tumours (30685)	Live	Yes	Typically not recruited
Ultra-rare disorders (30783)	Undescribed disorders (30784)	Ultra-rare undescribed monogenic disorders (30785)	Live	No	Should be recruited
	Multi-system groups (38589)	Neonatal or paediatric intensive care admission with a likely monogenic disease (38558)	Live	No	Depends on clinical situation
		Single autosomal recessive mutation in rare disease (38672)	Live	No	Should be recruited
		Undiagnosed monogenic disorder seen in a specialist genetics clinic (42193)	Live	Yes	Depends on clinical situation
Genomic medicine service indications (82157)	Whole genome sequencing indications (82159)	GMS R14 Acutely unwell infants with a likely monogenic disorder (82160)	Live	No	Should be recruited
		GMS R27 Congenital malformation and dysmorphism syndromes - likely monogenic (82161)	Live	No	Should be recruited
		GMS R69 Floppy infant with a likely central cause (82162)	Live	No	Should be recruited
		GMS R29 Moderate, severe or profound intellectual disability (82163)	Live	No	Should be recruited

		GMS R89 Ultra-rare and atypical monogenic disorders (82164)	Live	No	Depends on clinical situation
		GMS R100 Rare syndromic craniosynostosis or isolated multisuture synostosis (82165)	Live	Yes	Depends on clinical situation
		GMS R104 Skeletal dysplasia (82166)	Live	No	Should be recruited
		GMS R143 Neonatal diabetes (82167)	Live	No	Should be recruited
		GMS R98 Likely inborn error of metabolism - targeted testing not possible (82168)	Live	No	Should be recruited
		GMS R83 Arthrogryposis (82185)	Live	Yes	Should be recruited
		GMS R84 Cerebellar anomalies (82169)	Live	No	Should be recruited
		GMS R87 Cerebral malformation (82170)	Live	No	Should be recruited
		GMS R61 Childhood onset hereditary spastic paraplegia (82171)	Live	Yes	Should be recruited
		GMS R109 Childhood onset leukodystrophy (82172)	Live	No	Should be recruited
		GMS R59 Early onset or syndromic epilepsy (82173)	Live	No	Should be recruited
		GMS R54 Hereditary ataxia with onset in adulthood (82174)	Live	No	Typically not recruited
		GMS R55 Hereditary ataxia with onset in childhood (82175)	Live	No	Should be recruited
		GMS R85 Holoprosencephaly - NOT chromosomal (82176)	Live	No	Should be recruited
		GMS R86 Hydrocephalus (82177)	Live	No	Should be recruited
		GMS R381 Other rare neuromuscular disorders (82178)	Live	No	Should be recruited
		GMS R88 Severe microcephaly (82179)	Live	No	Should be recruited

		GMS R193 Cystic renal disease (82180)	Live	Yes	Typically not recruited

*please refer to relevant Eligibility Statement