GOALS:

- ✓ **To identify** the cause of rare genetic diseases, provide a clinical diagnosis and, in time, new or more effective treatments.
- ✓ **To accelerate** the uptake of genomic medicine in the NHS.
- ✓ **To provide** new scientific insights and discovery.
- ✓ **To stimulate** and enhance the UK genomics industry.
- ✓ **To increase** public knowledge and support for genomic medicine by delivering an ethical and transparent program.

For more information see:
http://www.genomicsengland.co.uk

Enquiries to
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Genomics england

Genomics england

NHS Wessex
NHS Genomic Medicine Centre

A Cardiologist's guide to the

100,000 Genomes Project

PROJECT OVERVIEW

We will sequence 100,000 genomes from 70,000 people; NHS patients with rare disease and their families, or patients with cancer.

We will create a new genomic medicine service for the NHS, transforming the way people are cared for. Patients may be offered a diagnosis where there wasn't one before. In time, there may also be the potential of new and more effective treatments.

A Cardiologist's Guide

HOW TO REFER:

Discuss with the family in clinic

Explain purpose of study and that blood samples will be needed from affected individuals and unaffected parent(s) or other affected family members.

Data will be made available in anonymized form to research/commercial bodies.

Patients who already have a molecular genetic diagnosis are excluded from this project because the aim is to make diagnoses that have not been previously possible.

Where the inclusion criteria include 'familial', two or more family members should be affected, even if only one is available for DNA testing.

Once verbal consent has been obtained, refer via the Genomics tab on **equest**.

Please complete both Request Details tab and Service Details tab with patient and relatives' full names and DOBs unless it is a singleton case.

If you have any concerns about the case fulfilling the referral criteria, please contact the rare disease team or Dr Catherine Mercer, Cardiac Genetics Lead.

WHO TO REFER:

1. Thoracic aortic aneurysm disease

a. Familial Thoracic Aortic Aneurysm and dissection

b. Thoracic aortopathy <50 years with no established risk factors

c. Clinically diagnosed Marfan syndrome with no FBN1 mutation

d. Loeys-Dietz syndrome and similar conditions

2. Cardiac arrhythmias, either familial or severely affected singleton suggestive of autosomal recessive inheritance

a. Brugada syndrome (or mixture of FH of SADS and Brugada)

b. Long QT syndrome

c. Catecholaminergic polymorphic ventricular tachycardia

3. Cardiomyopathy, either familial or with clear diagnosis <40 years

a. Hypertrophic Cardiomyopathy

b. Dilated Cardiomyopathy, +/- conduction defects

c. Arrhythmogenic right ventricular cardiomyopathy

d. Left ventricular non compaction

4. Congenital Heart Disease; must have CHD *plus*:

- consanguineous family history or
  - at least one first degree relative with a structural cardiac anomaly or
  - One or more additional non-cardiac abnormalities

a. Tetralogy of Fallot

b. Hypoplastic Left Heart Syndrome/LVOTO disorders

c. Pulmonary atresia

d. Transposition of the great vessels

e. Isomerism and laterality disorders

If condition appears dominantly inherited, recruit as many affected family members as possible.

If the case is a singleton with no other affected family members, consistent with autosomal recessive disease, recruit affected proband and parents where possible.

For possible X-linked or mitochondrial diseases, discuss with rare disease team re which patients to recruit.

If you have a patient with a likely inherited condition which is not included in this leaflet, please get in touch as we may be able to recruit them under an 'ultra-rare monogenetic disorders' category.