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100,000 Genomes Project

Cardiology

Routine genetic testing should have been performed prior to recruitment for most conditions

Familial Thoracic Aortic Aneurysm Disease

- Familial thoracic aortic aneurysm and dissection
- Thoracic aortopathy < 50 years (no prior genetic testing required)
- Marfan syndrome

Cardiomyopathies

ARVC, HCM, LVNC, DCM

- Patients with a clear diagnosis and at least one affected relative
- Single case of HCM < 40 years of age
- Severe or syndromic cases with un-affected parents

Brugada Syndrome

- Type I Brugada ECG (spontaneous or after provocation)

AND at least one of the following

- FH of premature sudden death < 45 or SADS < 65 years
- Family member with Brugada ECG
- Survivor of cardiac arrest, spontaneous type I ECG, unaffected parents

Familial Hypercholesterolaemia (FH)

Simon Broome criteria for definite FH

- Chol > 7.5 mmol/l, or LDL > 4.9 mmol/ l and tendon xanthomas in patient or relative

OR

- Chol > 8.5 mmol/l, or LDL > 5.5 mmol/l and FH of premature MI or raised Chol

Congenital Heart Disease

Familial Congenital Heart Disease

- Congenital heart disease

AND at least one of the following:

- ≥ 1 first degree relative with congenital heart disease
- Parental consanguinity

Syndromic Congenital Heart Disease

- Congenital heart disease

AND

- ≥ 1 malformations outside the cardiovascular system or neurodevelopmental delay

Other:

Unexplained sudden death in the young

Idiopathic VF

Catecholaminergic polymorphic VT

LQT syndrome

Full Rare Disease Eligibility Criteria

https://www.genomicsengland.co.uk/information-for-gmc-staff/rare-disease-documents/

For any questions please contact the
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