

Molecular Genetic Testing Request Form

For detailed lab and referral information please see our website: https://www.rbht.nhs.uk/our-services/clinical_support/laboratories/clinical-genetics-and-genomics-laboratory

All fields are mandatory. Illegible, unclear or incomplete forms or incorrect blood containers will result in delayed processing or no tests being performed

Patient Details *(Affix sticker if available. A minimum of three identifiers are required)*

Family name: Sex assigned at birth: **Billing: NHS/PP**
First name(s): Hospital Number:
Date of Birth: NHS number:
Postcode: CGGL Family Number:

Ethnicity: ☐ White- British ☐ White- Irish ☐ White- Other ☐ Mixed- White/Black Caribbean
☐ Mixed- White/Black African ☐ Mixed-White/Asian ☐ Mixed - Any Other (please state:)
☐ Asian- Indian ☐ Asian- Pakistani ☐ Asian- Bangladeshi ☐ Asian- Any Other (please state:)
☐ Black- Caribbean ☐ Black- African ☐ Black- any Other ☐ Chinese ☐ Arab
☐ Any Other Ethnic Group (please state:) ☐ Not Stated ☐ Not Known

Referrer Details

Referrer: Tel:
Named Consultant:
Hospital & Dept:
NHS email address:

CC reports to:
(name and email)

Clinical Information - PLEASE STATE HOW THIS PATIENT MEETS ELIGIBILITY CRITERIA FOR GENOMIC TESTING
see: <https://www.england.nhs.uk/publication/national-genomic-test-directories/> **Testing will NOT be initiated if this information is not provided**

If possible, for familial cases please include a pedigree with the patient clearly marked:

Have other family members been tested by our lab? Please provide details:

Is this sample urgent? Please indicate why:

CONSENT STATEMENT: The results of a genetic test may have implications both for the person being tested and for other members of that person's family. It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test, that the sample may be stored for future diagnostic testing, and that the sample may be used to inform appropriate healthcare of members of the patient's family. In sending this form and sample for testing, the clinician has obtained consent for testing, storage and for the use of this sample and the information gathered from it to be shared with members of the patient's family through their health professionals (if appropriate). The patient should be advised that the sample may be used anonymously for quality assurance and training purposes. **If the patient does not wish information to be shared, or does not wish the sample to be stored, or to be used for quality assurance and training purposes, please write this clearly in the clinical summary box.** In the course of genetic analysis, we generate sequence data on many genes. It is foreseeable, that in a small proportion of cases, that while not actively sought, we may identify "incidental" findings in genes unrelated to the initial presenting clinical phenotype. Incidental Pathogenic/Likely Pathogenic variants in genes listed in the ACMG SF v3.1 list of secondary findings may be reported, following discussion with the referring clinician.

I consent for any surplus diagnostic samples to be used in ethical research projects approved by the Trust's research office. Some research projects involve collaboration with commercial companies. Samples will not be used for any animal experiments, or any research that benefits non-healthcare industry. Clinical data will only be accessed by authorised staff in relation to approved research projects and will be anonymised to any person not involved my direct clinical care. ☐ Yes ☐ No

I consent to genetic testing on my sample and understand the above information:

.....
Patient/parent's signature

..... / /
Date

Consent undertaken by:

.....
Clinician's name

.....
Clinician's signature

PHLEBOTOMY/REFERRER: Please take 2x 4ml EDTA blood
A minimum of 2x 1ml of EDTA Blood is acceptable for paediatric samples

Date of collection:

LAB USE ONLY

Sample(s) received:

Aliquot
checked:

Samples and forms should be sent to the lab packaged according to UN3373 guidance. All samples should be sent by first class post, courier or hospital transport.

Diagnostic testing is by Next Generation Sequencing (NGS) using custom panels. Data is generated and stored on all genes in each panel. Analysis, including CNV calling, will be reported on the genes of clinical relevance to the disease category requested below. Incidental findings may also be reported (see consent statement on page 1)

For full details of genes on each subpanel, please refer to our website (see page 1). National Genomic Test Directory codes ('R' no.) are included for cardiac and respiratory specialist test groups (in bold) only. **NOTE:** for NHS commissioned testing, requests **MUST** be for one of the Test directory coded panels.

CARDIAC *Please select a panel(s) for testing using tick boxes below*

Aortopathy disorders

- ☐ R125 Familial thoracic aortic aneurysm (FTAA)
☐ R140.1 Elastin-related phenotypes

Arrhythmias

- ☐ R127 Long QT syndrome (LQTS)
☐ R128 Brugada syndrome (BrS)
☐ R129 Catecholaminergic polymorphic VT (CPVT)
☐ R130 Short QT syndrome
☐ R328 Progressive cardiac conduction disease

Cardiomyopathies

- ☐ R131 Hypertrophic cardiomyopathy (HCM)
☐ R132 Dilated/arrhythmogenic cardiomyopathy (DCM/ACM)
☐ R133 Arrhythmogenic right ventricular cardiomyopathy (ARVC)
☐ R138 Sudden unexplained death or survivors of a cardiac event
☐ R135.2 Paediatric or syndromic cardiomyopathy

WGS - requires other consent and request forms - see:

<https://southeastgenomics.nhs.uk/professionals/whole-genome-sequencing/#HowToOrderATest>

- ☐ R135.3 Paediatric or syndromic cardiomyopathy
Semi-urgent in-house - please contact laboratory to discuss)

Other cardiac conditions

- ☐ R384 Generalised arterial calcification in infancy
☐ R391 Barth syndrome
☐ R134 Familial Hypercholesterolaemia including PRS

Primary Lymphoedema

- ☐ R136 Primary Lymphoedema

RESPIRATORY *Please select a panel(s) for testing using tick boxes below*

Bronchiectasis/Cystic Fibrosis/Ciliopathies

- ☐ R184 Cystic Fibrosis, *CFTR* full gene including introns
☐ R189 Respiratory ciliopathies including non-CF bronchiectasis
☐ R139 Laterality disorders & isomerism (heterotaxy)

Congenital respiratory conditions

- ☐ R330 Alveolar capillary dysplasia
☐ R333 Central Congenital Hypoventilation syndrome
☐ R426 Pulmonary alveolar microlithiasis (PAM)

Emphysema

- ☐ R191 Alpha-1-Antitrypsin deficiency (AAT)
☐ All Emphysema genes (small panel)

Interstitial Lung Disease (ILD)

- ☐ R192 Surfactant deficiency (includes childhood ILD)
☐ R421 Familial Pulmonary Fibrosis

Pulmonary Hypertension

- ☐ R188 Pulmonary Arterial Hypertension

Vasculopathies

- ☐ R190 Familial Pneumothorax
☐ R186 Hereditary Haemorrhagic Telangiectasia (HHT)

HPO terms

Please indicate any relevant HPO terms from the lists below IF APPLICABLE (major HPO terms only are listed)

Cardiac related

- | | |
|---|--|
| <input type="checkbox"/> Aortic aneurysm | <input type="checkbox"/> Arachnodactyly |
| <input type="checkbox"/> Aortic dissection | <input type="checkbox"/> Joint dislocation |
| <input type="checkbox"/> Arterial dissection | <input type="checkbox"/> Pectus excavatum |
| <input type="checkbox"/> Ectopia lentis | <input type="checkbox"/> Bicuspid aortic valve |
| <input type="checkbox"/> Myopia | <input type="checkbox"/> Arterial tortuosity |
| <input type="checkbox"/> Disproportionate tall stature | <input type="checkbox"/> Aneurysm-osteoarthritis syndrome |
| <input type="checkbox"/> Ventricular fibrillation | <input type="checkbox"/> Bruising susceptibility |
| <input type="checkbox"/> Atrial fibrillation | <input type="checkbox"/> Tachycardia |
| <input type="checkbox"/> Atrial flutter | <input type="checkbox"/> Bradycardia |
| <input type="checkbox"/> Prolonged QTc interval | <input type="checkbox"/> Syncope |
| <input type="checkbox"/> Shortened QT interval | <input type="checkbox"/> Palpitations |
| <input type="checkbox"/> Left bundle branch block | <input type="checkbox"/> Right bundle branch block |
| <input type="checkbox"/> ST segment elevation | <input type="checkbox"/> Impaired myocardial contractility |
| <input type="checkbox"/> Atrioventricular block | <input type="checkbox"/> Sudden cardiac death |
| <input type="checkbox"/> Subvalvular aortic stenosis | <input type="checkbox"/> Severely reduced left ventricular ejection fraction |
| <input type="checkbox"/> Hypertrophic cardiomyopathy | <input type="checkbox"/> Increased left ventricular end-diastolic volume |
| <input type="checkbox"/> Asymmetric septal hypertrophy | <input type="checkbox"/> Sensorineural hearing impairment |
| <input type="checkbox"/> Congestive heart failure | <input type="checkbox"/> Generalized arterial calcification |
| <input type="checkbox"/> Arrhythmia | <input type="checkbox"/> Premature arteriosclerosis |
| <input type="checkbox"/> Ventricular arrhythmia | <input type="checkbox"/> Precocious atherosclerosis |
| <input type="checkbox"/> Sinus bradycardia | <input type="checkbox"/> Hypertension |
| <input type="checkbox"/> Dilated cardiomyopathy | <input type="checkbox"/> Angina pectoris |
| <input type="checkbox"/> Cardiomegaly | <input type="checkbox"/> Myocardial infarction |
| <input type="checkbox"/> Arterial stenosis | <input type="checkbox"/> Coronary artery atherosclerosis |
| <input type="checkbox"/> Pulmonary artery stenosis | <input type="checkbox"/> Abnormality of the lymphatic system |
| <input type="checkbox"/> Abnormal left ventricular function | <input type="checkbox"/> Short stature |
| <input type="checkbox"/> Heart murmur | |

Other (state)

Respiratory related

- | | |
|---|--|
| <input type="checkbox"/> Bronchiectasis | <input type="checkbox"/> Failure to thrive |
| <input type="checkbox"/> Chronic bronchitis | <input type="checkbox"/> Exocrine pancreatic insufficiency |
| <input type="checkbox"/> Chronic rhinitis | <input type="checkbox"/> Situs inversus totalis |
| <input type="checkbox"/> Chronic sinusitis | <input type="checkbox"/> Ciliary dyskinesia |
| <input type="checkbox"/> Recurrent respiratory infections | <input type="checkbox"/> Immotile cilia |
| <input type="checkbox"/> Nasal polyposis | <input type="checkbox"/> Absent outer dynein arms |
| <input type="checkbox"/> Chronic otitis media | <input type="checkbox"/> Absent inner dynein arms |
| <input type="checkbox"/> Elevated sweat chloride | <input type="checkbox"/> Male infertility |
| <input type="checkbox"/> Abnormal lung lobation | <input type="checkbox"/> Hypoventilation |
| <input type="checkbox"/> Alveolar capillary dysplasia | <input type="checkbox"/> Hypoxemia |
| <input type="checkbox"/> Neonatal respiratory distress | <input type="checkbox"/> Apnea |
| <input type="checkbox"/> Progressive pulmonary function impairment | <input type="checkbox"/> Intra-alveolar nodular calcifications |
| <input type="checkbox"/> Emphysema | <input type="checkbox"/> Absent surfactant-protein |
| <input type="checkbox"/> Desquamative interstitial pneumonitis | <input type="checkbox"/> Interstitial pneumonitis |
| <input type="checkbox"/> Respiratory distress | <input type="checkbox"/> Respiratory insufficiency |
| <input type="checkbox"/> Respiratory failure | <input type="checkbox"/> Pulmonary fibrosis |
| <input type="checkbox"/> Ground-glass opacification | <input type="checkbox"/> Cough |
| <input type="checkbox"/> Crazy paving pattern | <input type="checkbox"/> Exertional dyspnea |
| <input type="checkbox"/> Abnormal pulmonary interstitial morphology | <input type="checkbox"/> Elevated pulmonary artery pressure |
| <input type="checkbox"/> Pulmonary arterial hypertension | <input type="checkbox"/> Increased pulmonary vascular resistance |
| <input type="checkbox"/> Abnormal pleura morphology | <input type="checkbox"/> Telangiectasia of the skin |
| <input type="checkbox"/> Pneumothorax | <input type="checkbox"/> Mucosal telangiectasiae |
| <input type="checkbox"/> Epistaxis | <input type="checkbox"/> Spontaneous hematomas |
| <input type="checkbox"/> Arteriovenous malformation | |

Other (state)

ADDITIONAL TESTING/ ☐ R442 Variant Reinterpretation (requests for variants previously classified by our lab are only considered if classification is >2 years old)
VARIANT REINTERPRETATION ☐ R387 Re-analysis of existing data (ie: analysis of another gene panel following diagnostic testing)

TESTING FOR A KNOWN FAMILIAL VARIANT: *A COPY OF PROBAND REPORT AND A POSITIVE CONTROL SAMPLE MUST BE SUPPLIED, OR FULL DETAILS OF WHERE THE PROBAND WAS TESTED MUST BE INDICATED*

- ☐ R240.1 Diagnostic/confirmatory testing (patient has phenotype consistent with familial disease-causing variant)
☐ R242.1 Predictive/pre-symptomatic testing (no or unknown phenotype; available for pathogenic or likely pathogenic variants only)
☐ R244.1 Family studies (carrier testing or segregation analysis for variant interpretation)

Variant/previous testing details:

- ☐ R346.1 DNA STORAGE ONLY (no test will be performed until requested)