South East Genomic Laboratory Hub

Royal Brompton and Harefield hospitals NHS

Clinical Genetics & Genomics Laboratory, Level 2 Sydney Wing, Royal Brompton Hospital, Sydney Street, London, SW3 6NP, UK

tel: 0207 352 8121 ext. 83009
email: rbh-tr.genomics@nhs.net or geneticslab@rbht.nhs.uk

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: M / F **Billing:** NHS / PP

First name(s): Hospital Number:

Date of Birth: NHS number:

Postcode: CGGL Family Number:

Ethnic origin: ☐Caucasian ☐African/African American ☐Hispanic/Latino ☐Middle Eastern

☐S Asian (inc. Bangladeshi, Indian & Pakistani) ☐E Asian (inc. Chinese & Japanese) ☐Ashkenazi Jewish

☐Mixed_____ ☐Other_____ Country: _____

Referrer Details

Referrer: Tel:

Named Consultant:

Hospital:

Department:

NHS email address:

CC reports to (name and address):

Clinical information and family history

Please give as much clinical & genetic information as possible,

Please ensure this referral complies with current eligibility criteria: https://www.england.nhs.uk/publication/national-genomic-test-directories/

For familial cases please include a pedigree with the patient clearly marked:

Have other members of this family been tested by our lab? Y/N Please provide details:

Is this sample urgent Y/N 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 will 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:

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 Paediatric or syndromic cardiomyopathy**
- ☐ (R135) RASopathies/Noonan syndrome

Other cardiac conditions

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

Primary Lymphoedema

- ☐ **R136 Primary Lymphoedema**

RESPIRATORY

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**
- ☐ Periventricular nodular heterotopia and lung disease (*FLNA*)
- ☐ **R421 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

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

Other (state)

Respiratory related

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

Other (state)

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)
- ☐ **R242.1Predictive/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)**

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.