

# Molecular Genetic Testing Request and Consent Form



**Clinical Genetics & Genomics** | Level 2 Sydney Wing, Sydney Street, London SW3 6NP  
 Tel: 00 44 (0)20 7352 8121 extension:83009 | Fax: 0207 351 8143 | Website: [www.rbht.nhs.uk/ggl](http://www.rbht.nhs.uk/ggl)  
 Email: [rbh-tr.genomics@nhs.net](mailto:rbh-tr.genomics@nhs.net) or [geneticslab@rbht.nhs.uk](mailto:geneticslab@rbht.nhs.uk)



Royal Brompton & Harefield Hospitals

<p><b>Patient Details</b> (Affix sticker if available. A minimum of three identifiers are required)</p> <p>Family name: ..... Gender: <input type="checkbox"/> M <input type="checkbox"/> F</p> <p>First name(s): .....</p> <p>Hospital Number: .....</p> <p>Date of Birth: ____/____/____ Phone Number: .....</p> <p>Email: .....</p> <p>Home Address: .....</p> <p>Postcode: ..... Country: .....</p> <p>RBHT Family Number: .....</p> <p>Interpreter required: <input type="checkbox"/> Yes <input type="checkbox"/> No Language: .....</p>	<p><b>Payment Details</b></p> <p>Payment Method: <input type="checkbox"/> Insurance <input type="checkbox"/> Embassy <input type="checkbox"/> Self-Funding</p> <p>Payment Provider: .....</p>
<p><b>Ethnic Origin</b></p> <p><input type="checkbox"/> Caucasian <input type="checkbox"/> African/African American <input type="checkbox"/> Hispanic/Latino</p> <p><input type="checkbox"/> Middle Eastern <input type="checkbox"/> S Asian (inc. Bangladeshi, Indian &amp; Pakistani)</p> <p><input type="checkbox"/> E Asian (inc. Chinese &amp; Japanese) <input type="checkbox"/> Ashkenazi Jewish</p> <p><input type="checkbox"/> Mixed .....</p> <p><input type="checkbox"/> Other ..... Country: .....</p>	<p><b>Referrer Details</b></p> <p>Referrer: .....</p> <p>Phone Number: .....</p> <p>Named Consultant: .....</p> <p>Hospital: .....</p> <p>Department: .....</p> <p>Address: .....</p> <p>Email address: .....</p> <p>CC reports to (name and address): .....</p>
<p><b>Family History and Clinical Information</b></p> <p>Please provide as much clinical &amp; genetic information as possible.</p> <div style="text-align: right; margin-top: 20px;"> <p>For familial cases please include a pedigree with the patient clearly marked:</p>  </div> <p>Have other members of this family been tested by our lab? <input type="checkbox"/> Y <input type="checkbox"/> N</p> <p>Details: .....</p>	
<p><b>Record of discussion regarding testing and storage of genetic material</b></p> <p><i>Your clinician will offer you a copy of this consent form for your information.</i></p> <p>1. The results of a genetic test may have implications both for the person being tested and for other members of that person's family. I acknowledge that my results may sometimes be used to inform the appropriate healthcare of members of my family and give my permission for this.</p> <p>2. Occasionally leftover samples may be useful in validating and developing new laboratory techniques and assays; and my sample might also be used as a 'quality control' for other testing, for example, that of family members.</p> <p>3. In the course of our routine clinical sequencing, we may generate sequence data on many genes. This enables us to streamline and maximise the usefulness of the test. It is foreseeable, that in a small proportion of cases we will identify "incidental" or "secondary" findings. Current policy is for clinical interpretation and validation to be undertaken ONLY in those genes requested overleaf.</p> <p>4. Normal laboratory practice is to store the sample even after the current testing is complete. This is because further/new tests may become available. In such cases I would like:</p> <p><input type="checkbox"/> (a) To be contacted before further relevant tests are performed</p> <p style="text-align: center;"><b>OR</b></p> <p><input type="checkbox"/> (b) Further diagnostic tests to be undertaken on the stored sample and to be told of any informative results</p> <p>5 I consent for any surplus diagnostic samples that are taken during the course of my treatment to be used for the purposes of research in projects that are considered to be ethical and have been approved by the Trust's research office. Some research projects may originate from and be carried out in collaboration with commercial companies. Samples will not be used for any animal experiments, or any research that benefits the tobacco industry. Clinical data will only be accessed by authorised staff in relation to approved research projects and will be anonymised (no identifiable details included) to any person not involved my direct clinical care. <input type="checkbox"/> Yes <input type="checkbox"/> No</p> <p><b>I consent to genetic testing on my sample and understand the above information:</b></p> <p>Patient/parent's signature: ..... Date:    /    /</p> <p>Clinician's name: ..... Clinician's signature: .....</p>	
<p><b>PHLEBOTOMY/REFERRER:</b> (Please take 2 x 4ml EDTA blood)  <small>A minimum of 2x 1ml of EDTA Blood is acceptable for paediatric samples</small></p>	<p><b>LAB:</b> Sample(s) received:</p> <p>Aliquot checked:</p>

**Illegible, unclear or incomplete forms or incorrect blood containers will result in delayed processing or no tests being performed**  
 Note: Please ensure the latest version of this request form is used, this can be found on our website: [www.rbht.nhs.uk/ggl](http://www.rbht.nhs.uk/ggl)

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Email: [rbh-tr.genomics@nhs.net](mailto:rbh-tr.genomics@nhs.net) or [geneticslab@rbht.nhs.uk](mailto:geneticslab@rbht.nhs.uk)

Royal Brompton & Harefield Hospitals

Date of collection: / /

**NEXT GENERATION SEQUENCING (NGS)** - Testing for the conditions below NGS. Data will be generated and stored on all genes in each panel. Comprehensive bioinformatic analysis, including copy number variant analysis, clinical interpretation and variant confirmation will be reported only on the genes of clinical relevance to the disease category requested below.

## Inherited Cardiac and Respiratory Diseases

For full details of the genes included on each subpanel please refer to our website: [www.rbht.nhs.uk/ggl](http://www.rbht.nhs.uk/ggl)

### Aortopathies

- Familial thoracic aortic aneurysm (FTAA) (medium panel)
- Loeys-Dietz syndrome (LDS) (small panel)
- Marfan syndrome (MFS) (small panel)
- Weill-Marchesani syndrome (small panel)
- All Aortopathy genes (medium panel)

### Arrhythmias

- Andersen-Tawil syndrome (*KCNJ2*)
- Brugada syndrome (BrS) (*SCN5A*)
- Catecholaminergic polymorphic VT (CPVT) (small panel)
- Long QT syndrome (LQTS) (small panel)
- Short QT syndrome (small panel)
- All Arrhythmia genes (medium panel)

### Cardiomyopathies

- Arrhythmogenic right ventricular cardiomyopathy (ARVC) (small panel)
- Dilated/arrhythmogenic cardiomyopathy (DCM/ACM) (large panel)
- Hypertrophic cardiomyopathy (HCM) (large panel)
- Laminopathy (*LMNA*)
- Noncompaction cardiomyopathy (LVNC) (small panel)
- All Cardiomyopathy genes (medium panel)
- Molecular autopsy (Sudden Cardiac Death) (medium panel)
- Paediatric or syndromic cardiomyopathy (large panel)

### Hypercholesterolemia

- Familial Hypercholesterolemia (FH) (small panel)

Requested information from the referrer:

Score according to the Dutch Lipids Clinic Network Criteria

- Possible: 3-5 points
- Probable: 6-7 points
- Definite:  $\geq$  points

Plasma levels of LDL-C

- mg/dL
- mmol/L

In the most recent analysis performed

- Treated
- Untreated

### Other cardiac conditions

- Alagille syndrome (*JAG1*)
- Barth syndrome (*TAZ*)
- Carney complex (*PRKAR1A*)
- (R131) Fabry disease (*GLA*)
- Holt-Oram syndrome (*TBX5*)
- NKX2-5-related disorders (*NKX2-5*)
- RASopathies/Noonan spectrum disorders (small panel)
- SALL4-related disorders

### Primary Lymphoedema

- R136 Primary Lymphoedema (medium panel)

### Bronchiectasis/Cystic Fibrosis/Ciliopathies

- Cystic Fibrosis targeted analysis – 36 common Caucasian *CFTR* variants
- Cystic Fibrosis full gene including introns (*CFTR*)
- Primary Ciliary Dyskinesia (PCD) (medium panel)
- Respiratory ciliopathies including non-CF bronchiectasis (large panel including PCD genes and *CFTR*)
- Joubert syndrome (JS) (small panel)
- Orofaciodigital syndrome (OFD) (small panel)
- Short rib thoracic dysplasia (Jeune syndrome) (SRTD) (small panel)
- All Ciliopathy genes (including PCD) (medium panel)

### Congenital respiratory conditions

- Alveolar capillary dysplasia (*FOXF1*)
- Central Congenital Hypoventilation syndrome (small panel)
- Periventricular nodular heterotopia and lung disease (*FLNA*)
- Primary pulmonary hypoplasia (*ZFPM2*)
- Pulmonary alveolar microlithiasis (PAM) (*SLC34A2*)
- All Congenital respiratory condition genes (small panel)

### Emphysema

- Alpha-1-Antitrypsin deficiency (AAT) (*SERPINA1*)
- All Emphysema genes (small panel)

### Interstitial Lung Disease (ILD)

- Surfactant deficiency (childhood ILD) (small panel)
- Hermansky-Pudlak Syndrome (HPS) (small panel)
- Pulmonary fibrosis, familial (FPF) (medium panel)
- Tuberous sclerosis (TS) (small panel)
- All Interstitial Lung Disease (ILD) genes (medium panel)

### Laterality Disorders and Isomerism

- Laterality disorders & isomerism (heterotaxy) (medium panel)

### Pulmonary Hypertension

- Pulmonary Arterial Hypertension (small panel)

### Vasculopathies

- Birt-Hogg-Dubé syndrome (*FLCN*)
- Capillary malformation-arteriovenous malformation (*RASA1*)
- R190 Familial Pneumothorax (large panel)
- R186 Hereditary Haemorrhagic Telangiectasia (HHT) (small panel)
- Homocystinuria (small panel)
- Venous Malformations (small panel)
- Alport syndrome, X-linked (*COL4A5*)
- Cutis laxa (small panel)
- All Vasculopathy genes (small panel)

- All Inherited Cardiac Condition genes (large panel)

*Only available after discussion with the laboratory*

- All Inherited Respiratory Condition genes (large panel)

*Only available after discussion with the laboratory*

## TESTING FOR A KNOWN FAMILIAL VARIANT:

*Please provide a copy of the familial report or full details of the proband if tested at RBH*

- Diagnostic/confirmatory testing (has phenotype consistent with familial disease-causing variant)
- Predictive/pre-symptomatic testing (has no or unknown phenotype. *Available for pathogenic or likely pathogenic variants only*)
- Family studies (for variant interpretation)

Variant details:

Extract and store DNA (no test will be performed until requested)

Variant re-analysis and reporting

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