Illegible, unclear or incomplete forms or incorrect blood containers will result in delayed processing or no tests being performed

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Royal Brompton and Harefield hospitals
Part of Guy's and St Thomas' NHS Foundation Trust

Molecular Genetic Testing Request and Consent Form

For detailed lab and referral information please see our website: www.rbht.nhs.uk/ggl

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: Ext:

Named Consultant:

Hospital:

Department:

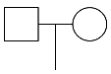
NHS email address:

CC reports to (name and address):

Clinical information and family history

Please give as much clinical & genetic information as possible

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:

Record of discussion regarding testing and storage of genetic material - *Your clinician will offer you a copy of this consent form for your information.*

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 be used to inform the appropriate healthcare of members of my family and give my permission for this.

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.

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.

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: (a) To be contacted before further relevant tests are performed ☐ **Yes** ☐ **No**

OR (b) Further diagnostic tests to be undertaken on the stored sample and to be told of any informative results ☐ **Yes** ☐ **No**

5. I consent for any surplus diagnostic samples that are taken during my treatment to be used for the purposes of research in projects that are 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. ☐ **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: Sample(s) received:

Aliquot checked:

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NEXT GENERATION SEQUENCING - *Testing for the conditions below utilises Next Generation Sequencing (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
*National Genomic Test Directory codes ('R' number) are included for cardiac and respiratory specialist test groups (**in bold**) only (brackets indicate sub-panel of main panel). Small panels defined as ≤10 genes; Large panels defined as >10 genes. **NOTE:** for NHS commissioned testing, requests MUST be for one of the Test directory coded panels*

Aortopathy/Vasculopathy and connective tissue disorders

- ☐ **R125 Familial thoracic aortic aneurysm (FTAA) (large panel)**
- ☐ Loeys-Dietz syndrome (LDS) (small panel) (R125)
- ☐ Marfan syndrome (MFS) (small panel) (R125)
- ☐ Alport syndrome, X-linked (COL4A5) (R125)
- ☐ Cutis laxa (small panel) (R125)
- ☐ Vascular Ehlers-Danlos syndrome (EDS) (small panel) (R125)
- ☐ Weill-Marchesani syndrome (ADAMTS10, ADAMTS17, LTBP2) (R125)
- ☐ All Aortopathy and connective tissue genes (large panel)

Arrhythmias

- ☐ **R127 Long QT syndrome (LQTS) (small panel)**
- ☐ (R127) Andersen-Tawil syndrome (KCNJ2)
- ☐ **R128 Brugada syndrome (BrS) (*SCN5A*)**
- ☐ **R129 Catecholaminergic polymorphic VT (CPVT) (small panel)**
- ☐ **R130 Short QT syndrome small panel)**
- ☐ All Arrhythmia genes (~38 genes)

Cardiomyopathies

- ☐ **R131 Hypertrophic cardiomyopathy (HCM) (large panel)**
- ☐ **R132 Dilated/arrhythmogenic cardiomyopathy (DCM/ACM) (large panel)**
- ☐ (R132) Laminopathy (*LMNA*)
- ☐ **R133 Arrhythmogenic right ventricular cardiomyopathy (ARVC) (small panel)**
- ☐ Noncompaction cardiomyopathy (LVNC) (~8 genes)
- ☐ **R135 Paediatric or syndromic cardiomyopathy (large panel)**
- ☐ All Cardiomyopathy genes (~88 genes)

Additional cardiac conditions

- ☐ **R138 Molecular autopsy (Sudden Cardiac Death) (large panel)**
- ☐ Alagille syndrome (*JAG1*)
- ☐ **R391 Barth syndrome (*TAZ*)**
- ☐ Carney complex (*PRKAR1A*)
- ☐ Fabry disease (*GLA*) (R131)
- ☐ Familial Hypercholesterolemia (FH) (small panel)
- ☐ Holt-Oram syndrome (*TBX5*)
- ☐ *NKX2-5*-related disorders (*NKX2-5*)
- ☐ RASopathies/Noonan spectrum disorders (large panel)
- ☐ *SALL4*-related disorders

Primary Lymphoedema

- ☐ **R136 Primary Lymphoedema (large 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 (*MTHFR, CBS*)
- ☐ Microcephaly Capillary Malformation syndrome (*STAMBP*)
- ☐ Venous Malformations (*GLMN, TEK*)
- ☐ All Vasculopathy genes (large panel)

Bronchiectasis/Cystic Fibrosis/Ciliopathies

- ☐ **R184 Cystic Fibrosis targeted analysis – 36 common Caucasian *CFTR* variants**
- ☐ **Cystic Fibrosis full gene including introns (*CFTR*)**(R184.1 & R184.2)
- ☐ **R189 Respiratory ciliopathies including non-CF bronchiectasis (large panel including PCD genes and *CFTR*)**
- ☐ Primary Ciliary Dyskinesia (PCD) (large panel) (R189)
- ☐ Joubert syndrome (JS) (large panel) (R189)
- ☐ Orofaciodigital syndrome (OFD) (small panel)
- ☐ Short rib thoracic dysplasia (Jeune syndrome) (SRTD) (large panel) (R189)
- ☐ All Ciliopathy genes (including PCD) (large panel) (R189)

Congenital respiratory conditions

- ☐ **R330 Alveolar capillary dysplasia (*FOXF1*)**
- ☐ Ataxia telangiectasia (*ATM*)
- ☐ **R333 Central Congenital Hypoventilation syndrome (*PHOX2B ONLY*)**
- ☐ 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 (~12 genes)

Emphysema

- ☐ **R191 Alpha-1-Antitrypsin deficiency (AAT) (*SERPINA1*)**
- ☐ All Emphysema genes (~5 genes)

Immunodeficiencies

- ☐ Agammaglobulinemia (*PIK3R1, BTK*)
- ☐ Autoimmune lymphoproliferative syndrome (*CTLA4*)
- ☐ Autoinflammation, antibody deficiency, immune dysregulation (*PLCG2*)
- ☐Candidiasis, familial (*CARD9, IL17R, IL17F*)
- ☐Hyper-IgE recurrent infection (*STAT3, DOCK8*)
- ☐Immunodeficiency, common variable (~20 genes)
- ☐Immunodysregulation, polyendocrinopathy & enteropathy (*FOXP3*)
- ☐Susceptibility to Aspergillosis (*CLEC7A*)
- ☐All Immunodeficiency genes (~31 genes)

Interstitial Lung Disease (ILD)

- ☐ **R192 Surfacatant deficiency (childhood ILD) (small panel)**
- ☐ Hermansky-Pudlak Syndrome (HPS) (small panel)
- ☐ Pulmonary fibrosis, familial (FPF) (large panel)
- ☐ Tuberous sclerosis (TS) (*TSC1, TSC2*)
- ☐ All Interstitial Lung Disease (ILD) genes (large panel)

Laterality Disorders and Isomerism

- ☐ **R139 Laterality disorders & isomerism (heterotaxy) (large panel)**

Pulmonary Hypertension

- ☐ **R188 Pulmonary Arterial Hypertension (small panel)**

☐ All Inherited Cardiac 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

☐ **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 details:**

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

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