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 & Harefield NHS
NHS Foundation Trust

UKAS MEDICAL 9295

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 sometimes 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 ☐

OR

(b) Further diagnostic tests to be undertaken on the stored sample and to be told of any informative results ☐

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. ☐ **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**
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

Aortopathy and connective tissue genes
- ☐Alport syndrome, X-linked (*COL4A5*)
- ☐Cutis laxa (~4 genes)
- ☐Ehlers-Danlos syndrome (EDS) (~15 genes)
- ☐Familial thoracic aortic aneurysm (FTAA) (~26 genes)
- ☐Loeys-Dietz syndrome (LDS) (~5 genes)
- ☐Marfan syndrome (MFS) (*~5 genes*)
- ☐Weill-Marchesani syndrome (*ADAMTS10, ADAMTS17, LTBP2*)
- ☐All Aortopathy and connective tissue genes (~63 genes)

Arrhythmia genes
- ☐Andersen-Tawil syndrome (*KCNJ2*)
- ☐Brugada syndrome (BrS) (~13 genes)
- ☐Catecholaminergic polymorphic ventricular tachycardia (CPVT) (~4 genes)
- ☐Long QT syndrome (LQTS) (~14 genes)
- ☐Short QT syndrome (~6 genes)
- ☐All Arrhythmia genes (~38 genes)

Cardiomyopathy genes
- ☐Arrhythmogenic cardiomyopathy (ACM) (~14 genes)
- ☐Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) (~8 genes)
- ☐Dilated cardiomyopathy (DCM) (~38 genes)
- ☐Hypertrophic cardiomyopathy (HCM) (~29 genes)
- ☐Laminopathy (*LMNA*)
- ☐Noncompaction cardiomyopathy (LVNC) (~8 genes)
- ☐Fabry disease (*GLA*)
- ☐All Cardiomyopathy genes (~88 genes)

☐**Familial Hypercholesterolemia (FH) (~4 genes + 14 SNPs)**

Other cardiac conditions and genes
- ☐Alagille syndrome (*JAG1*)
- ☐Carney complex (*PRKAR1A*)
- ☐Heterotaxy/situs ambiguous (HTX) (~30 genes)
- ☐Holt-Oram syndrome (*TBX5*)
- ☐*NKX2-5*-related disorders
- ☐Noonan spectrum disorders (~11 genes)
- ☐*SALL4*-related disorders

Vasculopathy genes
- ☐Birt-Hogg-Dubé syndrome (Primary spontaneous pneumothorax) (*FLCN*)
- ☐Capillary malformation-arteriovenous malformation/Parkes-Weber syndrome (*RASA1*)
- ☐Hereditary Haemorrhagic Telangiectasia (HHT) (~4 genes)
- ☐Homocystinuria (*MTHFR*, *CBS*)
- ☐Microcephaly Capillary Malformation syndrome (*STAMBP*)
- ☐Venous Malformations (*GLMN, TEK*)
- ☐All Vasculopathy genes (~12 genes)

Bronchiectasis genes
- ☐Cystic Fibrosis targeted mutation analysis - 36 most common *CFTR* mutations in EU populations
- ☐Sequencing of the *CFTR* gene (exons)
- ☐Non-CF Bronchiectasis (4 x ENAC genes)
- ☐Primary Ciliary Dyskinesia (PCD) (~43 genes)
- ☐All Bronchiectasis genes (~48 genes including PCD genes and *CFTR*)

Ciliopathy genes
- ☐Joubert syndrome (JS) (~20 genes)
- ☐Orofaciodigital syndrome (OFD) (~6 genes)
- ☐Short rib thoracic dysplasia (Jeune syndrome) (SRTD) (~13 genes)
- ☐All Ciliopathy genes (including PCD) (~76 genes)

Congenital respiratory condition genes
- ☐Alveolar capillary dysplasia (*FOXF1*)
- ☐Ataxia telangiectasia (*ATM*)
- ☐Central Hypoventilation syndrome (~7 genes)
- ☐Periventricular nodular heterotopia and lung disease (*FLNA*)
- ☐Primary pulmonary hypoplasia (*ZFPM2*)
- ☐Pulmonary alveolar microlithiasis (PAM) (*SLC34A2*)
- ☐All Congenital respiratory condition genes (~12 genes)

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

Immunodeficiency genes
- ☐Agammaglobulinemia (*PIK3R1, BTK*)
- ☐Autoimmune lymphoproliferative syndrome (*CTLA4*)
- ☐Autoinflammation, antibody deficiency and immune dysregulation syndrome (*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) genes
- ☐Childhood ILD (ChILD) including surfactant genes (~7 genes)
- ☐Hermansky-Pudlak Syndrome (HPS) (~8 genes)
- ☐Pulmonary fibrosis, familial (FPF) (~26 genes)
- ☐Tuberous sclerosis (TS) (*TSC1, TSC2*)
- ☐All Interstitial Lung Disease (ILD) genes (~36 genes)

☐**Molecular autopsy (Sudden Cardiac Death, SCD) (~115 genes)**

☐**Pulmonary Hypertension (~6 genes)**

☐**All Inherited Cardiac Condition genes (~169 genes)**
Only available after discussion with the laboratory

☐**All Inherited Respiratory Condition genes (~171 genes)**
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)

☐ Family studies (for variant interpretation) **Variant details:**

☐ 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.