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
Email: rbh-tr.genomics@nhs.net or geneticslab@rbht.nhs.uk

UKAS MEDICAL 9295

RB&HH SPECIALIST CARE
Royal Brompton & Harefield Hospitals

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

Family name: ..... Gender: ☐ M ☐ F
First name(s): .....
Hospital Number: .....
Date of Birth: ../../.. Phone Number: .....
Email: .....
Postcode: ..... RBHT Family Number: .....
Interpreter required: ☐ Yes ☐ No Language: .....

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: .....

Payment Details

Payment Method: ☐ Insurance ☐ Embassy ☐ Self-Funding
Payment Provider: .....

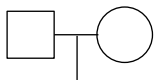
Referrer Details

Referrer: .....
Phone Number: .....
Named Consultant: .....
Hospital: .....
Department: .....
Address: .....
Email address: .....
CC reports to (name and address): .....
.....

Family History and Clinical Information

Please provide 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
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: / /
Clinician's name: ..... Clinician's signature: .....

PHLEBOTOMY/REFERRER: (Please take 2 x 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:

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

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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

Aortopathy/Vasculopathy and connective tissue disorders

- ☐ Alport syndrome, X-linked (*COL4A5*)
- ☐ Cutis laxa (~4 genes)
- ☐ Ehlers-Danlos syndrome (EDS) (~15 genes)
- ☐ Familial thoracic aortic aneurysm (FTAA) (large panel)
- ☐ Loeys-Dietz syndrome (LDS) (*5 genes*)
- ☐ Marfan syndrome (MFS) (*6 genes*)
- ☐ Weill-Marchesani syndrome (*ADAMTS10, ADAMTS17, LTBP2*)
- ☐ All Aortopathy and connective tissue genes (~62 genes)

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 (~38 genes)

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) (~8 genes)
- ☐ All Cardiomyopathy genes (~88 genes)
- ☐ Molecular autopsy (Sudden Cardiac Death) (large panel)
- ☐ Paediatric or syndromic cardiomyopathy (large panel)

Other cardiac conditions

- ☐ Alagille syndrome (*JAG1*)
- ☐ Barth syndrome (*TAZ*)
- ☐ Carney complex (*PRKAR1A*)
- ☐ (R131) Fabry disease (*GLA*)
- ☐ Familial Hypercholesterolemia (FH) (~4 genes)
- ☐ Holt-Oram syndrome (*TBX5*)
- ☐ *NKX2-5*-related disorders (*NKX2-5*)
- ☐ RASopathies/Noonan spectrum disorders (~11 genes)
- ☐ *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 (~12 genes)

Bronchiectasis/Cystic Fibrosis/Ciliopathies

- ☐ Cystic Fibrosis targeted analysis – 36 common Caucasian *CFTR* variants
- ☐ Cystic Fibrosis full gene including introns (*CFTR*)
- ☐ Primary Ciliary Dyskinesia (PCD) (~43 genes)
- ☐ Respiratory ciliopathies including non-CF bronchiectasis (large panel including PCD genes and *CFTR*)
- ☐ 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 conditions

- ☐ Alveolar capillary dysplasia (*FOXF1*)
- ☐ Ataxia telangiectasia (*ATM*)
- ☐ Central Congenital Hypoventilation syndrome (*PHOX2B ONLY*)
- ☐ Central Congenital 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

- ☐ 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)

- ☐ Surfacatant deficiency (childhood ILD) (small panel)
- ☐ 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)

Laterality Disorders and Isomerism

- ☐ Laterality disorders & isomerism (heterotaxy) (~30 genes)

Pulmonary Hypertension

- ☐ Pulmonary Arterial Hypertension (small panel)

- ☐ 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. *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)

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.