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: 3009 | **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: .....

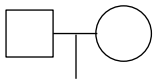
Referrer Details

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

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 agree that residual DNA samples may be stored for use in future ethically approved research projects conducted by the Clinical Genetics & Genomics Laboratory and their research collaborators, in order to help improve our understanding of human disease. ☐ **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)
Date of collection: / /

LAB: Sample(s) received: .....
Aliquot checked: ..... Form checked: .....

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

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☐ DNA STORAGE ONLY (no test will be performed until requested)

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:** .....

Inherited Cardiac and Respiratory Diseases

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) (*FBN1, FBN2, SLC2A10*)
☐ 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 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*)
☐ Megalencephaly Capillary Malformation Syndrome (*PIK3CA*)
☐ Microcephaly Capillary Malformation syndrome (MCAP) (*STAMBP*)
☐ Venous Malformations (*GLMN, TEK*)
☐ All Vasculopathy genes (13 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 genes)
☐ Primary Ciliary Dyskinesia (PCD) (43 genes)
☐ All Bronchiectasis genes (48 genes)

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) (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

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

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