# 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 |
**Website:** https://www.rbht.nhs.uk/our-services/clinical_support/laboratories/clinical-genetics-and-genomics-laboratory
**Email:** gstt.rbhh.genomics@nhs.net

RB&HH SPECIALIST CARE
Royal Brompton & Harefield Hospitals

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

Family name: ..... Sex: M☐ F☐
First name(s): .....
Hospital Number: .....
Date of Birth: ..... Phone Number: .....
Email: .....
Home Address: .....
Postcode: ..... Country: .....
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:
(not editable for online version)

Have other members of this family been tested by our lab? Y☐ N☐
Details:

**CONSENT STATEMENT:** The results of a genetic test may have implications both for the person being tested and for other members of that person's family. It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test, that the sample may be stored for future diagnostic testing, and that the sample may be used to inform appropriate healthcare of members of the patient's family.
In sending this form and sample for testing, the clinician has obtained consent for testing, storage and for the use of this sample and the information gathered from it to be shared with members of the patient's family through their health professionals (if appropriate). The patient should be advised that the sample may be used anonymously for quality assurance and training purposes. If the patient does not wish information to be shared, or does not wish the sample to be stored, or to be used for quality assurance and training purposes, please write this clearly in the clinical summary box.
In the course of genetic analysis, we generate sequence data on many genes. It is foreseeable, that in a small proportion of cases, that while not actively sought, we will identify "incidental" findings in genes unrelated to the initial presenting clinical phenotype. Incidental Pathogenic/Likely Pathogenic variants in genes listed in the ACMG SF v3.1 list of secondary findings may be reported, following discussion with the referring clinician.

I consent for any surplus diagnostic samples to be used in ethical research projects approved by the Trust's research office. Some research projects involve collaboration with commercial companies. Samples will not be used for any animal experiments, or any research that benefits non-healthcare industry. Clinical data will only be accessed by authorised staff in relation to approved research projects and will be anonymised 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 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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*Diagnostic testing is by Next Generation Sequencing (NGS) using custom panels. Data is generated and stored on all genes in each panel. Analysis, including CNV calling, will be reported on the genes of clinical relevance to the disease category requested below. Incidental findings may also be reported (see consent statement on page 1)*

## Inherited Cardiac and Respiratory Diseases

*For full details of the genes included on each subpanel please refer to our website (link above).* Please select a panel(s) for testing using tick boxes below

### Aortopathies
- ☐ Familial thoracic aortic aneurysm (FTAA)
- ☐ Marfan syndrome (MFS)

### Arrhythmias
- ☐ Andersen-Tawil syndrome (*KCNJ2*)
- ☐ Brugada syndrome (BrS) (*SCN5A*)
- ☐ Catecholaminergic polymorphic VT (CPVT)
- ☐ Long QT syndrome (LQTS)
- ☐ Short QT syndrome
- ☐ All Arrhythmia genes

### Cardiomyopathies
- ☐ Arrhythmogenic right ventricular cardiomyopathy (ARVC)
- ☐ Dilated/arrhythmogenic cardiomyopathy (DCM/ACM)
- ☐ Hypertrophic cardiomyopathy (HCM)
- ☐ Laminopathy (*LMNA*)
- ☐ Noncompaction cardiomyopathy (LVNC)
- ☐ Paediatric or syndromic cardiomyopathy
- ☐ Sudden unexplained death or survivors of a cardiac event (Molecular autopsy)
- ☐ All Cardiomyopathy genes

### Hypercholesterolemia
- ☐ Familial Hypercholesterolemia (FH)

Requested information from the referrer:
Score according to the Dutch Lipids Clinic Network Criteria
☐ Possible: 3-5 points ☐ Probable: 6-7 points ☐ Definite: ≥ points
Plasma levels of LDL-C
☐ mg/dL ☐ mmol/L
In the most recent analysis performed
☐ Treated ☐ Untreated

### Other cardiac conditions
- ☐ Generalised arterial calcification in infancy
- ☐ Alagille syndrome (*JAG1*)
- ☐ Barth syndrome (*TAZ*)
- ☐ Carney complex (*PRKAR1A*)
- ☐ Fabry disease (*GLA*)
- ☐ Holt-Oram syndrome (*TBX5*)
- ☐ *NKX2-5*-related disorders (*NKX2-5*)
- ☐ RASopathies/Noonan spectrum disorders
- ☐ *SALL4*-related disorders

### Primary Lymphoedema
- ☐ Primary Lymphoedema

### Bronchiectasis/Cystic Fibrosis/Ciliopathies
- ☐ Cystic Fibrosis full gene including introns (*CFTR*)
- ☐ Respiratory ciliopathies including non-CF bronchiectasis (includes PCD genes and *CFTR*)
- ☐ Orofaciodigital syndrome (OFD) (small panel)
- ☐ Schwann-diamond syndrome (*SBDS*)

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

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

### Interstitial Lung Disease (ILD)
- ☐ Surfactant deficiency (childhood ILD)
- ☐ Pulmonary fibrosis, familial (FPF) (medium panel)

### Laterality Disorders and Isomerism
- ☐ Laterality disorders & isomerism (heterotaxy)

### Pulmonary Hypertension
- ☐ Pulmonary Arterial Hypertension

### Vasculopathies
- ☐ Birt-Hogg-Dubé syndrome (*FLCN*)
- ☐ Capillary malformation-arteriovenous malformation (*RASA1*)
- ☐ Familial Pneumothorax (medium panel)
- ☐ Hereditary Haemorrhagic Telangiectasia (HHT)

- ☐ 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.*

RB&HH
SPECIALIST CARE

# REGISTRATION / AGREEMENT TO PAY FORM

PLEASE COMPLETE PATIENT DETAILS IN CAPITAL LETTERS

***PLEASE INFORM A MEMBER OF STAFF IF YOU REQUIRE HELP OR SUPPORT WITH ANY ASPECT OF YOUR VISIT**

| | | | |
|---|---|---|---|
| **Hospital Number** | **Episode Date** | **Consultant** | |
| **Title** | **Surname** | **Forename** | **Date of Birth** |
| **Nationality** | **Ethnicity** | **Religion** | **Occupation** |
| **Address'**<br>**Permanent** | | **Postcode** | |
| **Temporary** | | **Postcode** | |
| **Home Telephone number** | **Work Telephone number** | **Mobile Phone number** | **Email Address** |
| **Happy to receive SMS text** | **Yes** **No** | **Marital Status** | **NHS number** |
| **Emergency contact:-** | **Name** | **Relationship** | **Telephone** |
| | **Address** | | |
| **GP Details** | | | |

**METHOD OF PAYMENT**

| ☐ **SELF-PAY ACCOUNT** | ☐ **MEDICAL INSURANCE** | ☐ **SPONSORED** |
|---|---|---|
| Deposit taken £ | INSURER | SPONSOR |
| | MEMBERSHIP NO | LETTER OF GUARANTEE |
| | AUTHORISATION | REFERENCE NO |
| *Payment in full is required for each completed out-patient consultation and or investigations. Patients being admitted need to have paid a deposit in cleared funds based on the estimated cost of the anticipated treatment on or prior to the admission date. Our tariff and estimated costs are available on request.* | *Patients covered by a Trust agreed insurer will be requested to provide the name of the insurer, membership number and authorisation details for this episode of care. In order for us to invoice and deal with your insurer directly.* | *Sponsored patients via Embassy or any other organisation with which the Trust has an agreement must provide an approved letter of guarantee.* |

*In order to provide you with the appropriate care required, we may need to share your medical records/images. These will be appropriately stored and shared with those involved in your care including other healthcare organisations if necessary.*

*Your personal information will also be used for administration, ethically approved clinical research and internal audit purposes. Additionally, the information you have provided on this form may be used to contact you for medical or financial matters.*

*I consent to the medical information of the above patient being submitted to, and validated by, official external agencies so that the quality of health care provided can be monitored independently and national health trends analysed. YES* ☐ *NO* ☐

*I understand that my consultant may charge in excess of the agreed insurance rate and that any outstanding amount will be my responsibility. I understand that the prices quoted for diagnostics are indicative only and can differ when final invoice is produced*

*I agree that a copy of my credit/debit card details will be taken to cover any outstanding costs.*

*I hereby undertake to pay Royal Brompton & Harefield NHS Foundation Trust for all services provided for private patient treatment or under such circumstances where medical insurance/sponsorship is declined or partially paid. I am aware that once discharge medication has been dispensed from the pharmacy it cannot be returned and that I am responsible for the costs of these items.*

| | |
|---|---|
| **Signature** | **Witnessed** |
| **Name Printed** | **Name Printed** |
| **Relationship to Patient** | **Position** |
| **Date** | **Date** |

*Consent Model wording (UK-wide)*

*Private Healthcare Information Network (PHIN)*

*In order to improve public's access to information on private healthcare quality/outcome we share some of your personal information (NHS number and postcode) with PHIN. PHIN then sends this Number to the relevant national information authority which then links it to the national hospital and mortality data. The linked information, with your personal data removed, is then provided to PHIN to measure care quality, check adverse events after hospital discharge (such as unplanned readmissions to hospital, emergency transfers or deaths following treatment).*

*Personal information is treated with high standards of confidentiality in accordance with data protection laws and the duty of confidentiality. Any information published is anonymous and only serves statistical purposes. This information will not be shared or analysed for any other purpose than those stated above.*

*Further detail is contained in PHIN's Privacy Notice, a copy of which is available on PHIN's website (phin.org.uk).*

*Your consent for your personal information to be processed in this way is entirely voluntary. You do not have to give consent and are free to withdraw consent at any time without giving any reason, and without your medical care or legal rights being affected.*

*I have read and understood how my personal data may be used, and* ***agree*** *to this purpose* ☐ ***disagree*** *to this purpose* ☐

**Royal Brompton & Harefield Hospitals Specialist Care**
Sydney Street, London SW3 6NP
T: +44 (0)20 3811 6871 W: www.rbhh-specialistcare.co.uk

Place Patient Label Here

Dear Patient,

Your doctor has recommended that you take a genetic test in order to aid the diagnosis of your condition. The Clinical Genetics and Genomics Laboratory offers genetic testing for inherited cardiac and respiratory conditions. This test may be able to detect whether your condition has a genetic cause or not and the results of this test may have implications for further family members, who may also wish to have genetic testing in the future.

For the test, we will require x2 5 ml EDTA blood, which will be taken in the private outpatient's department. You will be asked to read and sign a consent form, which is part of the test request form and will accompany the blood sample to the Clinical Genetics and Genomics Laboratory. The processing of such genetic tests is complex, so that you may not receive the result of your test for up to 3 months.

In the event of a positive genetic test, the Royal Brompton and Harefield hospitals offer Clinical Genetic services, including genetic counselling. Please ask your doctor to be referred to a Clinical Geneticist if you would like to discuss your test result or the possibility of testing of additional family members.

Since the private UK medical insurers do not routinely pay for all genetic tests, please note that you will have to pay for the cost of the test (around £1,000, but not more than £1,500) yourself before testing begins and then claim it back from your insurance company if they agree to pay for the test. In this regard, please check the box below and sign this form:

◯ I will pay for the test myself, please proceed with the test.

Signature:_____ Name Printed:_____ Date: ___/___/___