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BIOCHEMICAL GENETICS REQUEST FORM

Patient Information or Addressograph

First name: _____Surname: _____

Patient address: _____

_____

DOB::_____ Sex: _____

Ward/Clinic: _____ Hospital No._____

2 whole blood EDTA samples required

SJH Laboratory number

Referral Information:

Consultant's name: _____

Address of requesting consultant: _____ Hospital: _____

Name of referrer _____ Title/position: _____ Ext/Bleep: _____

Details of Test(s) Requested: **(include gene if known)**

Current Diagnosis (biochemical condition):

Clinical Information:

Family History: **(include details of name and DOB of index case & relationship, gene & familial variant if known)**

Informed Consent Information: ***Please retain original consent form in patient file.***

Patient/Guardian has signed consent form? (**Y/N**) _____ Patient/Guardian signature: _____

Specimen Information:

Specimen Type: (Whole Blood, Buccal Swab, DNA) _____

Date Taken: _____ (for internal use only: Date received: _____)

Requesting Clinician Signature: _____

MCRN no: _____

Date: _____

Please note that samples arriving without confirmed informed consent by signature will not be processed

P.T.O

Consent form for Diagnostic Genetic Testing on patient

1. I, _____, request that an attempt be made using genetic material (either DNA, RNA or both) to assess the probability that: I / my child (DELETE WHERE NOT APPLICABLE) might have inherited a disease-causing genetic variant in one or more of the genes listed in **Table 1**. Such genetic variants are associated with a susceptibility to a specific MEDICAL CONDITION ("disorder") as indicated also in **Table 1**.

Table 1: Please tick the genetic test required

MEDICAL CONDITION	Genes	Genetic test requested (tick)
Porphyrias		
ACUTE HEPATIC PORPHYRIAS [including acute intermittent porphyria (AIP), variegate porphyria (VP) and hereditary coproporphyria (HCP)]	HMBS, PPOX, CPOX	
Familial porphyria cutanea tarda (fPCT)	UROD, HFE	
Erythropoietic protoporphyria (EPP) and X-linked protopoprhyria (XLP)	FECH, ALAS2	
Congenital erythropoietic porphyria (CEP)	UROS	
Other Biochemical conditions		
Dysbetalipoproteinaemia (Type III Hyperlipidaemia)	APOE	
Gilbert's syndrome (Benign unconjugated hypberbilirubinaemia)	UGT1A1	
Familial hypocalciuric hypercalcaemia (FHH) Autosomal Dominant Hypocalcaemia (ADH)	CASR	
Hypophosphatasia	ALPL	
Butyrylcholine esterase deficiency (Succinylcholine sensitivity, Pseudocholinesterase deficiency)	BCHE	
Familial partial lipodystrophy (FPLD)	PPARG & LMNA	
Hereditary Transthyretin mediated (hATTR) Amyloidosis	TTR	
Other: (Please indicate condition/gene if known)		

2. In wishing to proceed with this test I have been fully informed about the test. I understand that the test will show **ONE** of the following:

a. That I do have the disorder or carry a strong genetic susceptibility for the disorder and that other family members may therefore be at risk of developing this condition.
b. That I do not have the disorder
c. That the test results are indeterminate or difficult to interpret.

3. Patient or Guardian:

I consent to be tested for the genetic test(s) and understand the implications of the test	YES / NO
I consent for the DNA from this sample to be stored	YES / NO
I consent for this sample to be used for quality assurance and audit purposes	YES / NO
I consent for the results of this test to be available to assist in testing other family members	YES / NO

Please note: *samples will be stored for a minimum of 5 years after which time they may be discarded, unless otherwise requested by patient/Guardian*

Signature of patient/parent/guardian: _____

Date: _____

For Medical Staff:
I have explained in detail to the above patient the principles and implications of genetic testing for the disorder. Given the clinical information available at this juncture I believe this test to be in the best interests of the patient.

Signature: _____ Date: _____

Name (Printed): _____

Medical Council registration number: _____