ThromboGenomics

Cambridge University Hospitals NHS
NHS Foundation Trust

REGIONAL GENETICS LABORATORIES TEST REQUEST

All tests requested will be reviewed against departmental criteria. If testing is not arranged, the samples will be stored and the referring clinicians informed. After testing, samples may be used anonymously for the development of new tests and for quality monitoring.

Surname	**Date of Birth**	**Age at Presentation**
First Names		**Sex**
NHS Number		**Ethnicity**
Hospital Number	**Family Number**	

Home Address

Postcode

Patient email address

GP Name (Printed)

GP Address

Postcode

GP email address (nhs.net preferred)

Consultant (Print)	**Hospital**

Speciality/Dept/Ward

Contact telephone number

Email address (nhs.net preferred)

Results to (if different from above) **inc email address (nhs.net preferred)**

Venous blood samples: Adult: 5ml; Children: 1-5ml

☐ DNA test: EDTA tube

☐ Chromosomes: Lithium Heparin tube

☐ Microarray: Lithium Heparin and EDTA tubes

Other samples:

☐ Cord/Placenta/insertion site/skin

☐ Products of Conception (whole specimen in sterile pot)

☐ Amnio sample

☐ CVS

☐ Other (please contact the laboratory)

Sample obtained by

Signature ……

Printed Name ……..……..

Date ……

Billing to:
Department of Haematology
University of Cambridge
c/o Sophie Stock
Private Patient: ☐

In Submitting this sample, the clinician confirms that consent has been obtained for:

a) Testing and Storage
☐ Yes ☐No

b) The use of this sample and the information generated from it to be shared with members of the patient's family and their health professionals (if appropriate)
☐Yes ☐No

Clinical Synopsis

Please provide clinical synopsis, results from laboratory testing and relevant family history to aid the MDT in diagnosis.

Tests Required:

Storage Only: ☐

Billing to:

Private Patient: ☐

Gestation in weeks (If pregnant):

Partners Name and DOB:

Index Case (if not this patient):

The Laboratory does NOT report results via the telephone

ThromboGenomics

Cambridge University Hospitals NHS
NHS Foundation Trust

All samples MUST be labelled with FULL name, date of birth and NHS number
Processing of samples will be delayed if information is incomplete

Send samples at room temperature by 1st class post or courier to:
East Anglian Medical Genetics Service,
Genetics Laboratories, Box 143, ATC Level 6, Addenbrooke's Hospital,
Hills Road, Cambridge, CB2 0QQ

Laboratory opening hours: 8.30am - 5.30pm Monday to Friday
Telephone: 01223 348866
Fax: 01223 348712
Email: geneticslaboratories@nhs.net

For further information about sample requirements and tests available see:
www.cuh.org.uk/genetics-labs

Patients with a heritable bleeding, thrombotic and/or platelet disorder, with a suspected mutation in one of the genes listed below, can be submitted for the diagnostic ThromboGenomics test.

Patients without a candidate gene can be enrolled to a different project called BRIDGE BPD by contacting:
Dr Sofia Papadia, sp605@medschl.cam.ac.uk

Please select one or more relevant genes below. This will help the Multi Disciplinary Team (MDT) to generate a report. If you do not select a particular gene you will be contacted prior to sequencing. The sample will be tested for all the genes on the platform using the next generation sequencing.

For further information about the diagnostic ThromboGenomics test:
please visit www.thrombogenomics.org.uk or contact Kate Downes, PhD info@thrombogenomics.org.uk

Platelet Disorders	Genes	
ADP receptor defect	P2RY12	☐
Amegakaryocytic thrombocytopenia with radio-ulnar synostosis	HOXA11	☐
ARC syndrome	VPS33B; VIPAS3	☐
Autosomal dominant thrombocytopenia 2	ANKRD26	☐
Autosomal dominant thrombocytopenia 4	CYCS	☐
Bernard-Soulier syndrome	GP1BA; GP1BB; GP9	☐
Bleeding diathesis due to glycoprotein VI deficiency	GP6	☐
Chediak–Higashi syndrome	LYST	☐
Congenital amegakaryocytic thrombocytopenia (CAMT)	MPL	☐
Cyclic thrombocytopenia and thrombocythemia 1	THPO	☐
Deficiency of phospholipase A2, group IVA	PLA2G4A	☐
Dense granule abnormalities	NBEA	☐
Familial haemophagocytic lymphohistiocytosis, type 5	STXBP2	☐
Familial platelet disorder with predisposition to AML	RUNX1	☐
Ghosal syndrome	TBXAS1	☐
Glanzmann thrombasthenia	ITGA2B; ITGB3	☐
Gray platelet syndrome	NBEAL2	☐
Gray platelet-like syndrome	GFI1B	☐

Platelet Disorders	Genes	
Hermansky–Pudlak syndrome	HPS1; AP3B1; HPS3; HPS4; HPS5; HPS6; DTNBP1; BLOC1S3; BLOC1S6	☐
Leukocyte integrin adhesion deficiency, type III	FERMT3	☐
Macrothrombocytopenia	ACTN1; FLNA	☐
May-Hegglin and other MYH9 disorders	MYH9	☐
Myopathy associated with thrombocytopenia	GNE	☐
Paris-Trousseau thrombocytopenia and Jacobson syndrome	FLI1	☐
Platelet-type von Willebrand disease	GP1BA	☐
Platelet-type bleeding disorder 18	RASGRP2	☐
Quebec platelet disorder	PLAU	☐
Scott syndrome	ANO6	☐
Stormorken syndrome	STIM1; ORAI1	☐
Thrombocytopenia and susceptibility to cancer	ETV6	☐
Thrombocytopenia absent radius (TAR) syndrome	RBM8A	☐
Thromboxane A2 receptor defect	TBXA2R	☐
Wiskott-Aldrich syndrome	WAS	☐
X-linked thrombocytopenia with dyserythropoiesis	GATA1	☐

Coagulation Factor Disorders	Genes	
Alpha 2 anti-plasmin deficiency	SERPINF2	☐
Combined V and VIII deficiency	LMAN1; MCFD2	☐
Factor V deficiency	F5	☐
Factor VII deficiency	F7	☐
Factor X deficiency	F10	☐
Factor XI deficiency	F11	☐
Factor XIII deficiency	F13A1; F13B	☐
Fibrinogen deficiency	FGA; FGB; FGG	☐
Haemophilia A	F8	☐
Haemophilia B	F9	☐
Multiple coagulation factor deficiency type 3	GGCX	☐
Multiple coagulation factor deficiency type 2	VKORC1	☐
Plasminogen Activator Inhibitor 1 deficiency	SERPINE1	☐
Prothrombin deficiency	F2	☐
von Willebrand disease types 2 or 3	VWF	☐

Thrombotic Disorders	Genes	
Anti-thrombin deficiency	SERPINC1	☐
Heparin co-factor 2 deficiency	SERPIND1	☐
Histidine-rich glycoprotein deficiency	HRG	☐
Plasminogen deficiency	PLG	☐
Protein C deficiency	PROC	☐
Protein S deficiency	PROS1	☐
Thrombomodulin deficiency	THBD	☐
Tissue plasminogen activator deficiency	PLAT	☐

CUH Laboratory Use Only:

Receipt date and time:		**Other Information:**
Volume:	**No of tubes:**	
Tube type:		