Haemophilia & Thrombosis Genetics Request Form

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Patient Details	
CHI number:	Consultant:
Surname:	
Forename:	Hospital & Department:
DOB:	
Sex: M / F	NHS Email:
Hospital Number:	
Family number (if relevant):	Report via email? Y / N
Is the patient or partner pregnant? Yes / No Gestation:	Sample taken by: Date taken:

Request type:

Index case ☐ - Complete sections A & C

Known familial variant ☐ - Complete sections B & C

(A) Index case testing

Test requested

Gene panel:

Platelet Disorders ☐ Coagulation/Fibrinolysis ☐ Thrombosis ☐

For a full list of genes included in panels see **tinyurl.com/HaemGenePanels**

Specific gene(s):

Haemophilia A (*F8*) ☐	FVII (*F7*) ☐	Antithrombin (*SERPINC1*) ☐
Haemophilia B (*F9*) ☐	FX (*F10*) ☐	Protein S (*PROS1*) ☐
von Willebrand (*VWF*) ☐	FXI (*F11*) ☐	Protein C (*PROC*) ☐
Fibrinogen (*FGA, FGB, FGG*) ☐	FV (*F5*) ☐	Combined FV & FVIII (*LMAN1* & *MCFD2*) ☐
Platelet type VWD (*GP1BA*) ☐	FXIII (*F13A1* & *F13B*) ☐	Glanzmann Thrombasthenia (*ITGA2B & ITGB3*) ☐
Bernard Soulier Syndrome (*GP1BA, GP1BB, GP9*) ☐		Other:

Clinical Details

Suspected diagnosis:

Age of bleeding/thrombotic onset: **ISTH BAT score (if applicable):**

Clinical synopsis:

Family history: (Please attach a copy of family tree if available)

(B) Familial variant testing

Index case (full name & CHI/DOB):

Relationship to index case (please attach a copy of family tree if available):

Gene & Variant(s):

Clinical synopsis of individual being tested:

(C) Laboratory results: Please complete relevant results

Please include units & at least 2 levels from separate occasions

Coagulation				**Platelet**	
FV		Antithrombin		Platelet count	
FVII		Protein C		MPV	
FVIII		Protein S		Blood film	
FIX		VWF:Ag		**Platelet function test results**	
FX		VWF RCo		ADP	*Normal / Impaired / Absent*
FXI		VWF CBA		Adrenaline	*Normal / Impaired / Absent*
FXIII		Multimers		Collagen	*Normal / Impaired / Absent*
Fibrinogen Ag		VWF RIPA	*Normal / Abnormal*	Ristocetin	*Normal / Impaired / Absent*
Fibrinogen Clauss				Arachidonic acid	*Normal / Impaired / Absent*
Please provide any other relevant results for this patient:				U46619	*Normal / Impaired / Absent*
				Nucleotides Ratio	

CONSENT

- **Signed copy of the UKHCDO consent form attached? (required)** ☐
- **Has the patient consented for their genetic test result to be added to their National Haemophilia Database record? Yes / No**

Requested by:	**Date:**

Further Information

- **Sample requirements** Minimum 2 x 3ml blood in EDTA or citrate anticoagulant OR 100µl DNA
- In complying with the Human Tissue Act 2004 all surplus tissue samples are discarded once DNA has been extracted
- All samples must be labelled with a minimum dataset of full name, DOB and CHI number
- All samples should be kept at room temperature but can be refrigerated overnight prior to dispatch if required. DO NOT FREEZE BLOOD SAMPLES
- Send samples by post or courier in a rigid crushproof container according to current Post Office regulations

Turnaround Times			
Single gene tests 8 weeks	Known variant test 2 weeks	Gene panel (>10 genes) 12 weeks	Prenatal 5 days