# Haemophilia & Thrombosis Genetics Request Form

South East Scotland Molecular Genetics Laboratory, Western General Hospital, Crewe Road, Edinburgh, EH4 2XU
0131 537 2010 HaemophiliaGenetics.RIE@nhslothian.scot.nhs.uk https://tinyurl.com/ScottishHaemophiliaGenetics

NHS Lothian

| Patient Details | |
|---|---|
| CHI number: | Consultant: |
| Surname: | |
| Forename: | Hospital & Department: |
| DOB: | |
| Gender: M / F | Email (nhs.net/ scot.nhs.uk): |
| Hospital Number: | |
| Family number (if relevant): | Report via email? Y / N |
| Is the patient or partner pregnant? Yes / No<br>Gestation: | Sample taken by:<br>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 Function ☐ Thrombocytopenia ☐ Coagulation/Fibrinolysis ☐ Thrombosis ☐

**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:**

**ISTH BAT score (if applicable):**

# Haemophilia & Thrombosis Genetics Request Form

South East Scotland Molecular Genetics Laboratory, Western General Hospital, Crewe Road, Edinburgh, EH4 2XU
0131 537 2010 HaemophiliaGenetics.RIE@nhslothian.scot.nhs.uk https://tinyurl.com/ScottishHaemophiliaGenetics

NHS Lothian

**(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 | | Collagen | *Normal / Impaired / Absent* |
| FXI | | VWF CBA | | High Dose Ristocetin | *Normal / Impaired / Absent* |
| FXIII | | Multimers | | Low Dose Ristocetin | *Normal / Abnormal* |
| Fibrinogen Ag | | VWF RIPA | *Normal / Abnormal* | Arachidonic acid | *Normal / Impaired / Absent* |
| Fibrinogen Clauss | | | | U46619 1.0 mu | *Normal / Impaired / Absent* |
| Please provide any other relevant results for this patient: | | | | ADP | *Normal / Impaired / Absent* |
| | | | | Adrenaline | *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 (calendar days)** | | | |
|---|---|---|---|
| Single gene tests 56 | Known variant test 28 | NGS Gene panel 84 | Prenatal / other urgent testing<br>Please contact the laboratory |