ThromboGenomics

Cambridge University Hospitals NHS
NHS Foundation Trust

# GENETIC DIAGNOSIS FOR PATIENTS WITH RARE INHERITED BLEEDING CLOTTING, THROMBOTIC AND PLATELET DISORDERS

ThromboGenomics is now available at the East Midlands and East of England Genomics Laboratory Hub using the GEMINI clinical exome panel

£650 for UK NHS patients — reporting time - 16 weeks

Carrier testing and testing for familial variants

£150 for UK NHS patients — reporting time - 4 weeks

An additional 50% surcharge is included for private and international patients

For further information and to download a referral form visit our website
http://thrombo.cambridgednadiagnosis.org.uk/

or email Kate Downes at
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| 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 |
| Familial thrombotic thrombocytopenic purpura | ADAMTS13 |
| Kininogen Deficiency | KNG1 |

| 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 XII deficiency, hereditary Angioedema type III | F12 |
| 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 |
| Prothrombin deficiency | F2 |

| Disorders with bleeding or platelet phenotypes | Genes |
|---|---|
| Hereditary hemorrhagic telangiectasia type 2 | ACVRL1; ENG; SMAD4 |
| Ehlers-Danlos syndrome | COL1A1; COL3A1; COL5A1; COL5A2 |
| Fletcher factor (prekallikrein) deficiency | KLKB1 |
| Noonan Syndrome | PTPN11 |
| Oculocutaneous albinism type IV | SLC45A2 |

| Platelet Disorders | Genes |
|---|---|
| ADP receptor defect | P2RY12 |
| Amegakaryocytic thrombocytopenia with radio-ulnar synostosis | HOXA11 |
| ARC syndrome | VPS33B; VIPAS39 |
| Autosomal dominant thrombocytopenia 2 | ANKRD26 |
| Autosomal dominant thrombocytopenia 4 | CYCS |
| Bernard-Soulier syndrome, Platelet type VWD | GP1BA; GP1BB; GP9 |
| Bleeding diathesis due to glycoprotein VI deficiency | GP6 |
| Chediak–Higashi syndrome | LYST |
| Congenital amegakaryocytic thrombocytopenia | MPL |
| Cyclic thrombocytopenia and thrombocythemia 1 | THPO |
| Deficiency of phospholipase A2, group IVA | PLA2G4A |
| Dense granule abnormalities | NBEA |
| Ehlers-Danlos syndrome, musculocontractural type | CHST14 |
| 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 |
| Hermansky–Pudlak syndrome | HPS1; AP3B1; AP3D1; HPS3; HPS4; HPS5; HPS6; DTNBP1; BLOC1S3; BLOC1S6 |
| Leukocyte integrin adhesion deficiency, type III | FERMT3 |
| Macrothrombocytopenia, Beta-tubulin 1 related | ACTN1; FLNA; TUBB1 |
| Macrothrombocytopenia and sensorineural hearing loss | DIAPH1 |
| May-Hegglin and other MYH9 disorders | MYH9 |
| Myopathy associated with thrombocytopenia | GNE |
| Paris-Trousseau thrombocytopenia and Jacobsen syndrome | PLAU |
| Platelet-type bleeding disorder 18 | RASGRP2 |
| Quebec platelet disorder | PLAU |
| Scott syndrome | ANO6 |
| Sitosterolemia & Thrombocytopenia | ABCG5; ABCG8 |
| Stormorken syndrome | STIM1 |
| Takenouchi-Kosaki syndrome | CDC42 |
| Thrombocytopenia and myelofibrosis | KDSR; SRC |
| Thrombocytopenia and susceptibility to cancer | ETV6 |
| Thrombocytopenia absent radius (TAR) syndrome | RBM8A |
| Thromboxane A2 receptor defect | TBXA2R |
| Radioulnar synostosis with amegakaryocytic thrombocytopenia 2 | MECOM |
| Wiskott-Aldrich syndrome | WAS |
| X-linked thrombocytopenia with dyserythropoiesis | GATA1 |