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
£975 for private and international patients
Reporting time - 16 weeks

For further information see http://thrombo.cambridgednadiagnosis.org.uk/

Contact Kate Downes with any queries:
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ThromboGenomics_GEMINI_Version1

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