Panel de coagulación sanguínea

Nuestro **panel de coagulación sanguínea** contiene genes para el diagnóstico molecular de trombofilia, trombocitopenia, telangiectasia hemorrágica hereditaria, síndrome ARC, síndrome de Hermasky-Pudlak, trastornos del factor de coagulación y trastornos relacionados con las plaquetas.

Nº de genes:	112
Entrega:	25 días
Cobertura:	≥99,5% ≥20x Cobertura media con profundidad ≥ 150 x
Detalles:	Análisis CNV incluido

SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

Afibrinogenemia

Síndrome de artrogriposis-disfunción renal-colestasis

Trastornos del factor de coagulación

Hemofilia

Telangiectasia hemorrágica hereditaria

Síndrome de Hermasky-Pudlak

Trastornos relacionados con las plaquetas

Trombocitopenia

Trombofilia

Resumen de genes y enfermedades asociadas

Genes	OMIM (gen)	Enfermedades asociadas (OMIM)	Herencia
ABCG5	605459	Sitosterolemia 2	AR
ABCG8	605460	Gallbladder disease 4;Sitosterolemia 1	AR
ACTN1	102575	Bleeding disorder, platelet-type, 15	AD
ACVRL1	601284	Telangiectasia, hereditary hemorrhagic, type 2	AD
ADAMTS13	604134	Thrombotic thrombocytopenic purpura, hereditary	AR
ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ANKRD26	610855	Thrombocytopenia 2	AD
ANO6	608663	Scott syndrome	AR
AP3B1	603401	Hermansky-Pudlak syndrome 2	AR
ARPC1B	604223	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia	AR
BLOC1S3	609762	Hermansky-Pudlak syndrome 8	AR
BLOC1S6	604310	?Hermansky-pudlak syndrome 9	AR
C1R	613785	Ehlers-Danlos syndrome, periodontal type, 1	AD
CCM2	607929	Cerebral cavernous malformations-2	AD
CD36	173510	Malaria, cerebral, reduced risk of;Coronary heart disease, susceptibility to, 7;Platelet glycoprotein IV deficiency;Malaria, cerebral, susceptibility to	AR

CD40LG	300386	Immunodeficiency, XL, with hyper-IgM	XLR
CDC42	116952	Takenouchi-Kosaki syndrome	AD
CHST14	608429	Ehlers-Danlos syndrome, musculocontractural type 1	AR
COL1A2	120160	Osteoporosis, postmenopausal;Ehlers-Danlos syndrome, cardiac valvular type;Ehlers-Danlos syndrome, arthrochalasia type, 2;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2;Osteogenesis imperfecta, type II	AD, AR
COL3A1	120180	Ehlers-Danlos syndrome, vascular type;Polymicrogyria with or without vascular-type EDS	AD, AR
COL4A1	120130	?Retinal arteries, tortuosity of;Hemorrhage, intracerebral, susceptibility to;Microangiopathy and leukoencephalopathy, pontine, AD;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps;Brain small vessel disease with or without ocular anomalies	AD
COLGALT1	617531	Brain small vessel disease 3	AR
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts	AR
CYCS	123970	Thrombocytopenia 4	AD
DIAPH1	602121	Seizures, cortical blindness, microcephaly syndrome;Deafness, AD 1, with or without thrombocytopenia	AR, AD
DTNBP1	607145	Hermansky-Pudlak syndrome 7	AR
EFEMP2	604633	Cutis laxa, AR, type IB	AR

EFL1	617538	Shwachman-Diamond syndrome 2	AR
ENG	131195	Telangiectasia, hereditary hemorrhagic, type 1	AD
ETV6	600618	Leukemia, acute myeloid, somatic;Thrombocytopenia 5	AD
F10	613872	Factor X deficiency	AR
F11	264900	Factor XI deficiency, AD;Factor XI deficiency, AR	
F12	610619	Factor XII deficiency;Angioedema, hereditary, type III	AR, AD
F13A1	134570	Factor XIIIA deficiency;Venous thrombosis, protection against;Myocardial infarction, protection against	AR, AD
F13B	134580	Factor XIIIB deficiency	AR
F2	176930	Dysprothrombinemia;Stroke, ischemic, susceptibility to;Pregnancy loss, recurrent, susceptibility to, 2;Hypoprothrombinemia;Thrombophilia due to thrombin defect	AR, mi, AD
F5	612309	Pregnancy loss, recurrent, susceptibility to, 1;Factor V deficiency;Stroke, ischemic, susceptibility to;Budd-Chiari syndrome;Thrombophilia, susceptibility to, due to factor V Leiden;Thrombophilia due to activated protein C resistance	AD, AR, mi
F7	613878	Myocardial infarction, decreased susceptibility to;Factor VII deficiency	AR
F8	300841	Hemophilia A	XLR
F9	300746	Warfarin sensitivity;Thrombophilia, XL, due to factor IX defect;Hemophilia B;Deep venous thrombosis, protection against	XL, XLR
FANCA	607139	Fanconi anemia, complementation group A	AR
FCGR2C	612169		

FERMT3	607901	Leukocyte adhesion deficiency, type III	AR
FGA	134820	Hypodysfibrinogenemia, congenital;Afibrinogenemia, congenital;Amyloidosis, familial visceral;Dysfibrinogenemia, congenital	AR, AD
FGB	134830	Afibrinogenemia, congenital;Dysfibrinogenemia, congenital;Hypofibrinogenemia, congenital	AR
FGG	134850	Dysfibrinogenemia, congenital;Afibrinogenemia, congenital;Hypofibrinogenemia, congenital;Hypodysfibrinogenemia	AR
FLI1	193067	Bleeding disorder, platelet-type, 21	AD, AR
FLNA	300017	Frontometaphyseal dysplasia 1;Heterotopia, periventricular, 1;Terminal osseous dysplasia;Congenital short bowel syndrome;Otopalatodigital syndrome, type II;Melnick-Needles syndrome;Cardiac valvular dysplasia, XL;Intestinal pseudoobstruction, neuronal;?FG syndrome 2;Otopalatodigital syndrome, type I	XLR, XLD, XL
FYB1	602731	Thrombocytopenia 3	AR
GATA1	305371	Thrombocytopenia with beta-thalassemia, XL;Thrombocytopenia, XL, with or without dyserythropoietic anemia;Anemia, XL, with/without neutropenia and/or platelet abnormalities;Leukemia, megakaryoblastic, with or without Down syndrome, somatic	XLR
GFI1B	604383	Bleeding disorder, platelet-type, 17	AD, AR

GGCX	137167	Vitamin K-dependent clotting factors, combined deficiency of, 1;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	AR
GNE	603824	Nonaka myopathy;Sialuria	AR, AD
GP1BA	606672	Bernard-Soulier syndrome, type A1 (recessive);von Willebrand disease, platelet-type;Bernard-Soulier syndrome, type A2 (dominant);Nonarteritic anterior ischemic optic neuropathy, susceptibility to	AR, AD
GP1BB	138720	Giant platelet disorder, isolated;Bernard-Soulier syndrome, type B	AR
GP6	605546	Bleeding disorder, platelet-type, 11	AR
GP9	173515	Bernard-Soulier syndrome, type C	AR
GUCY1A1	139396	Moyamoya 6 with achalasia	AR
HOXA11	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1	AD
HPS1	604982	Hermansky-Pudlak syndrome 1	AR
HPS3	606118	Hermansky-Pudlak syndrome 3	AR
HPS4	606682	Hermansky-Pudlak syndrome 4	AR
HPS5	607521	Hermansky-Pudlak syndrome 5	AR
HPS6	607522	Hermansky-Pudlak syndrome 6	AR
HRG	142640	Thrombophilia due to HRG deficiency	AD
IGFBP7	602867	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	AR
IL2RG	308380	Severe combined immunodeficiency, XL;Combined immunodeficiency, XL, moderate	XLR
ITGA2B	607759	Glanzmann thrombasthenia;Bleeding disorder, platelet-type, 16, AD	AR, AD

ITGB3	173470	Glanzmann thrombasthenia 2;Glanzmann thrombasthenia;Bleeding disorder, platelet-type, 16, AD;Myocardial infarction, susceptibility to;Bleeding disorder, platelet-type, 24, AD	AR, AD
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	AR
KDSR	136440	Erythrokeratodermia variabilis et progressiva 4	AR
KRIT1	604214	Cavernous malformations of CNS and retina;Cerebral cavernous malformations-1;Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations	AD
LMAN1	601567	Combined factor V and VIII deficiency	AR
LYST	606897	Chediak-Higashi syndrome	AR
LYZ	153450	Amyloidosis, renal	AD
MCFD2	607788	Factor V and factor VIII, combined deficiency of	
MECOM	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2	AD
MPL	159530	Myelofibrosis with myeloid metaplasia, somatic;Thrombocytopenia, congenital amegakaryocytic;Thrombocythemia 2	AR, AD, SM
MYH9	160775	Deafness, AD 17;Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	AD
NBEAL2	614169	Gray platelet syndrome	AR
P2RY12	600515	Bleeding disorder, platelet-type, 8	AR
PDCD10	609118	Cerebral cavernous malformations 3	AD
PLA2G4A	600522	Gastrointestinal ulceration, recurrent, with dysfunctional platelets	AR

PLAT	173370	Hyperfibrinolysis, familial, due to increased release of PLAT;Thrombophilia, familial, due to decreased release of PLAT	
PLAU	191840	Quebec platelet disorder;Alzheimer disease, late-onset, susceptibility to	AD
PROC	612283	Thrombophilia due to protein C deficiency, AD;Thrombophilia due to protein C deficiency, AR	AD, AR
PROS1	176880	Thrombophilia due to protein S deficiency, AR;Thrombophilia due to protein S deficiency, AD	AR, AD
PTPN11	176876	Leukemia, juvenile myelomonocytic, somatic;LEOPARD syndrome 1;Metachondromatosis;Noonan syndrome 1	AD
RBM8A	605313	Thrombocytopenia-absent radius syndrome	AR
RUNX1	151385	Leukemia, acute myeloid;Platelet disorder, familial, with associated myeloid malignancy	AD, SM, AD
SBDS	607444	Aplastic anemia, susceptibility to;Shwachman-Diamond syndrome	AR
SERPINC1	107300	Thrombophilia due to antithrombin III deficiency	AD, AR
SERPIND1	142360	Thrombophilia due to heparin cofactor II deficiency	AD
SERPINE1	173360	Plasminogen activator inhibitor-1 deficiency	AD, AR
SERPINF2	613168	Alpha-2-plasmin inhibitor deficiency	AR
SLC35A1	605634	Congenital disorder of glycosylation, type IIf	AR
SLC7A7	603593	Lysinuric protein intolerance	AR
SLFN14	614958	Bleeding disorder, platelet-type, 20	AD

SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;Pancreatic cancer, somatic;Myhre syndrome;Polyposis, juvenile intestinal	AD
SRC	190090	Colon cancer, advanced, somatic;?Thrombocytopenia 6	AD
STIM1	605921	Myopathy, tubular aggregate, 1;Stormorken syndrome;Immunodeficiency 10	AD, AR
STN1	613128	Cerebroretinal microangiopathy with calcifications and cysts 2	AR
TBXA2R	188070	Bleeding disorder, platelet-type, 13, susceptibility to	AD
THBD	188040	Thrombophilia due to thrombomodulin defect;Hemolytic uremic syndrome, atypical, susceptibility to, 6	AD
THPO	600044	Thrombocythemia 1	AD
TUBB1	612901	Macrothrombocytopenia, AD, TUBB1-related	AD
VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2	AR
VKORC1	608547	Vitamin K-dependent clotting factors, combined deficiency of, 2;Warfarin resistance	AR, AD
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1	AR
VWF	613160	von Willebrand disease, types 2A, 2B, 2M, and 2N;von Willebrand disease, type 1;von Willebrand disease, type 3	AD, AR, AD, AR
WAS	300392	Wiskott-Aldrich syndrome;Thrombocytopenia, XL;Neutropenia, severe congenital, XL;Thrombocytopenia, XL, intermittent	XLR
WIPF1	602357	Wiskott-Aldrich syndrome 2	AR
F8	300841	Hemophilia A	XLR