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Full name:	Trombosis - Hemostasis (107 genes) - KUL
Description:	https://firebasestorage.googleapis.com/v0/b/uz-laboboeken.appspot.com/o/GHB_CME%2FNGS%20-%20Trombo%20-%20full%20panel.pdf?alt=media&token=20656d63-ec60-43ca-852c-e30cb1a59fb9
Version number:	Trombose-Hemostase_v6
Laboratory:	Centrum Menselijke Erfelijkheid - KUL
Created:	03 Jul 2019 - 12:29
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Related Diseases

- Acute myeloid leukemia with t(8;21)(q22;q22) translocation
- Aggressive systemic mastocytosis
- Alpha delta granule deficiency
- Arthrogryposis-renal dysfunction-cholestasis syndrome
- Attenuated Chédiak-Higashi syndrome
- Atypical hemolytic uremic syndrome with complement gene abnormality
- Atypical hemolytic-uremic syndrome with thrombomodulin anomaly
- Autosomal dominant macrothrombocytopenia
- Autosomal dominant thrombocytopenia with platelet secretion defect
- Autosomal thrombocytopenia with normal platelets
- Baraitser-Winter cerebrofrontofacial syndrome
- Becker nevus syndrome
- Bernard-Soulier syndrome
- Beta-thalassemia-X-linked thrombocytopenia syndrome
- Bleeding diathesis due to glycoprotein VI deficiency
- Bleeding diathesis due to thromboxane synthesis deficiency
- Bleeding disorder due to CalDAG-GEFI deficiency
- Bleeding disorder due to P2Y12 defect
- Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency
- Cerebral sinovenous thrombosis

- Chronic myeloid leukemia
- Chédiak-Higashi syndrome
- Combined deficiency of factor V and factor VIII
- Congenital alpha2-antiplasmin deficiency
- Congenital amegakaryocytic thrombocytopenia
- Congenital autosomal recessive small-platelet thrombocytopenia
- Congenital erythropoietic porphyria
- Congenital factor II deficiency
- Congenital factor V deficiency
- Congenital factor VII deficiency
- Congenital factor X deficiency
- Congenital factor XI deficiency
- Congenital factor XII deficiency
- Congenital factor XIII deficiency
- Congenital high-molecular-weight kininogen deficiency
- Congenital plasminogen activator inhibitor type 1 deficiency
- Congenital prekallikrein deficiency
- Congenital thrombotic thrombocytopenic purpura
- Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder
- DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome
- Diamond-Blackfan anemia
- East Texas bleeding disorder
- Essential thrombocythemia
- F12-related hereditary angioedema with normal C1Inh
- Familial abdominal aortic aneurysm
- Familial afibrinogenemia
- Familial cerebral saccular aneurysm
- Familial dysfibrinogenemia
- Familial hemophagocytic lymphohistiocytosis
- Familial hypodysfibrinogenemia
- Familial hypofibrinogenemia
- Familial platelet disorder with associated myeloid malignancy
- Familial thrombocytosis
- Fetal and neonatal alloimmune thrombocytopenia
- Gaucher disease type 1
- Gaucher disease type 3

- Ghosal hematodiaphyseal dysplasia
- Glanzmann thrombasthenia
- Gray platelet syndrome
- Hereditary combined deficiency of vitamin K-dependent clotting factors
- Hereditary hemorrhagic telangiectasia
- Hereditary isolated aplastic anemia
- Hereditary thrombocytopenia with early-onset myelofibrosis
- Hereditary thrombophilia due to congenital antithrombin deficiency
- Heritable pulmonary arterial hypertension
- Hermansky-Pudlak syndrome due to AP-3 deficiency
- Hermansky-Pudlak syndrome due to BLOC-1 deficiency
- Hermansky-Pudlak syndrome due to BLOC-2 deficiency
- Hermansky-Pudlak syndrome due to BLOC-3 deficiency
- Hermansky-Pudlak syndrome type 8
- Hermansky-Pudlak syndrome type 9
- Homozygous familial hypercholesterolemia
- Hypoplasminogenemia
- Juvenile myelomonocytic leukemia
- Lethal hydranencephaly-diaphragmatic hernia syndrome
- Leukocyte adhesion deficiency type III
- MYH9-related disease
- Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome
- Mild hemophilia A
- Mild hemophilia B
- Moderate hemophilia A
- Moderate hemophilia B
- Myelodysplastic syndrome
- Neonatal-onset severe multisystemic autoinflammatory disease with increased IL18
- PLG-related hereditary angioedema with normal C1Inh
- Paris-Trousseau thrombocytopenia
- Paroxysmal nocturnal hemoglobinuria
- Polycythemia vera
- Primary hypereosinophilic syndrome
- Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome
- Pseudo-von Willebrand disease
- Quebec platelet disorder

- Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome
- Scott syndrome
- Severe autosomal recessive macrothrombocytopenia
- Severe hemophilia A
- Severe hemophilia B
- Severe hereditary thrombophilia due to congenital protein C deficiency
- Severe hereditary thrombophilia due to congenital protein S deficiency
- Sialuria
- Sitosterolemia
- Stormorken-Sjaastad-Langslet syndrome
- Thrombocythemia with distal limb defects
- Thrombocytopenia with congenital dyserythropoietic anemia
- Thrombocytopenia-absent radius syndrome
- Thrombomodulin-related bleeding disorder
- Vascular Ehlers-Danlos syndrome
- Von Willebrand disease type 1
- Von Willebrand disease type 2A
- Von Willebrand disease type 2B
- Von Willebrand disease type 2M
- Von Willebrand disease type 2N
- Von Willebrand disease type 3
- Wiskott-Aldrich syndrome
- X-linked dyserythropoietic anemia with abnormal platelets and neutropenia
- X-linked severe congenital neutropenia
- X-linked thrombocytopenia with normal platelets

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ABCC4	95.00	0	NM_005845.4/ interpretable range CS1>95%
ABCG5	95.00	0	NM_022436.2/ interpretable range CS1>95%
ABCG8	95.00	0	NM_022437.2/ interpretable range CS1>95%
ACTB	95.00	0	NM_001101.4/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ACTN1	95.00	0	NM_001130004.1/ interpretable range CS1>95%
ACVRL1	95.00	0	NM_000020.2/ interpretable range CS1>95%
ADAMTS13	95.00	0	NM_139025.4/ interpretable range CS1>95%
ANKRD26	95.00	0	NM_014915.2/ interpretable range CS1>95%
ANO6	95.00	0	NM_001025356.2/ interpretable range CS1>95%
AP3B1	95.00	0	NM_003664.4/ interpretable range CS1>95%
AP3D1	95.00	0	NM_001261826.1/ interpretable range CS1>95%
ARPC1B	95.00	0	NM_005720.3/ interpretable range CS1>95%
BLOC1S3	95.00	0	NM_212550.3/ interpretable range CS1>95%
BLOC1S5	95.00	0	NM_201280.2/ interpretable range CS1>95%
BLOC1S6	95.00	0	NM_012388.2/ interpretable range CS1>95%
CDC42	95.00	0	NM_001791.3/ interpretable range CS1>95%
CHST14	95.00	0	NM_130468.3/ interpretable range CS1>95%
COL1A1	95.00	0	NM_000088.3/ interpretable range CS1>95%
COL3A1	95.00	0	NM_000090.3/ interpretable range CS1>95%
COL4A1	95.00	0	NM_001845.5/ interpretable range CS1>95%
COL4A2	95.00	0	NM_001846.3/ interpretable range CS1>95%
COL5A1	95.00	0	NM_000093.4/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
COL5A2	95.00	0	NM_000393.3/ interpretable range CS1>95%
CYCS	95.00	0	NM_018947.5/ interpretable range CS1>95%
DIAPH1	95.00	0	NM_005219.4/ interpretable range CS1>95%
DTNBP1	95.00	0	NM_032122.4/ interpretable range CS1>95%
ENG	95.00	0	NM_000118.3/ interpretable range CS1>95%
ETV6	95.00	0	NM_001987.4/ interpretable range CS1>95%
F10	95.00	0	NM_000504.3/ interpretable range CS1>95%
F11	95.00	0	NM_000128.3/ interpretable range CS1>95%
F12	95.00	0	NM_000505.3/ interpretable range CS1>95%
F13A1	95.00	0	NM_000129.3/ interpretable range CS1>95%
F13B	95.00	0	NM_001994.2/ interpretable range CS1>95%
F2	95.00	0	NM_000506.3/ interpretable range CS1>95%
F5	95.00	0	NM_000130.4/ interpretable range CS1>95%
F7	95.00	0	NM_000131.4/ interpretable range CS1>95%
F8	95.00	0	NM_000132.3/ interpretable range CS1>95%
F9	95.00	0	NM_000133.3/ interpretable range CS1>95%
FERMT3	95.00	0	NM_031471.5/ interpretable range CS1>95%
FGA	95.00	0	NM_021871.2/ interpretable range CS1>95%
FGB	95.00	0	NM_005141.4/ interpretable range CS1>95%
FGG	95.00	0	NM_000509.4/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
FLII	95.00	0	NM_002017.4/ interpretable range CS1>95%
FLNA	95.00	0	NM_001456.3/ interpretable range CS1>95%
FYB1	95.00	0	NM_001465.5/ interpretable range CS1>95%
GATA1	95.00	0	NM_002049.3/ interpretable range CS1>95%
GDF2	95.00	0	NM_016204.3/ interpretable range CS1>95%
GFI1B	95.00	0	NM_004188.5/ interpretable range CS1>95%
GGCX	95.00	0	NM_000821.5/ interpretable range CS1>95%
GNE	95.00	0	NM_001128227.2/ interpretable range CS1>95%
GP1BA	95.00	0	NM_000173.6/ interpretable range CS1>95%
GP1BB	95.00	0	NM_000407.4/ interpretable range CS1>95%
GP6	95.00	0	NM_001083899.2/ interpretable range CS1>95%
GP9	95.00	0	NM_000174.4/ interpretable range CS1>95%
HOXA11	95.00	0	NM_005523.5/ interpretable range CS1>95%
HPS1	95.00	0	NM_000195.3/ interpretable range CS1>95%
HPS3	95.00	0	NM_032383.3/ interpretable range CS1>95%
HPS4	95.00	0	NM_022081.5/ interpretable range CS1>95%
HPS5	95.00	0	NM_181507.1/ interpretable range CS1>95%
HPS6	95.00	0	NM_024747.5/ interpretable range CS1>95%
HRG	95.00	0	NM_000412.3/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
IKZF5	95.00	0	NM_001271840.1/ interpretable range CS1>95%
ITGA2B	95.00	0	NM_000419.3/ interpretable range CS1>95%
ITGB3	95.00	0	NM_000212.2/ interpretable range CS1>95%
KDSR	95.00	0	NM_002035.2/ interpretable range CS1>95%
KNG1	95.00	0	NM_001102416.2/ interpretable range CS1>95%
LMAN1	95.00	0	NM_005570.3/ interpretable range CS1>95%
LYST	95.00	0	NM_000081.3/ interpretable range CS1>95%
MCFD2	95.00	0	NM_139279.5/ interpretable range CS1>95%
MECOM	95.00	0	NM_001105078.3/ interpretable range CS1>95%
MPIG6B	95.00	0	NM_025260.3/ interpretable range CS1>95%
MPL	95.00	0	NM_005373.2/ interpretable range CS1>95%
MYH9	95.00	0	NM_002473.5/ interpretable range CS1>95%
NBEA	95.00	0	NM_015678.4/ interpretable range CS1>95%
NBEAL2	95.00	0	NM_015175.2/ interpretable range CS1>95%
P2RY12	95.00	0	NM_022788.4/ interpretable range CS1>95%
PIGA	95.00	0	NM_002641.3/ interpretable range CS1>95%
PLA2G4A	95.00	0	NM_024420.2/ interpretable range CS1>95%
PLAU	95.00	0	NM_002658.3/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
PLG	95.00	0	NM_000301.3/ interpretable range CS1>95%
PROC	95.00	0	NM_000312.3/ interpretable range CS1>95%
PROS1	95.00	0	NM_000313.3/ interpretable range CS1>95%
PTGS1	95.00	0	NM_000962.3/ interpretable range CS1>95%
RAP1B	95.00	-2	NM_015646.6/ interpretable range CS1>95%
RASGRP2	95.00	0	NM_153819.1/ interpretable range CS1>95%
RBM8A	95.00	0	NM_005105.4/ interpretable range CS1>95%
RUNX1	95.00	0	NM_001754.4/ interpretable range CS1>95%
SERPINC1	95.00	0	NM_000488.3/ interpretable range CS1>95%
SERPIND1	95.00	0	NM_000185.3/ interpretable range CS1>95%
SERPINE1	95.00	0	NM_000602.4/ interpretable range CS1>95%
SERPINF2	95.00	0	NM_000934.3/ interpretable range CS1>95%
SLFN14	95.00	0	NM_001129820.1/ interpretable range CS1>95%
SMAD4	95.00	0	NM_005359.5/ interpretable range CS1>95%
SRC	95.00	0	NM_005417.4/ interpretable range CS1>95%
STIM1	95.00	0	NM_003156.3/ interpretable range CS1>95%
STXBP2	95.00	0	NM_006949.3/ interpretable range CS1>95%
TBXA2R	95.00	0	NM_001060.5/ interpretable range CS1>95%
TBXAS1	95.00	0	NM_001061.4/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
THBD	95.00	0	NM_000361.2/ interpretable range CS1>95%
THPO	95.00	0	NM_000460.3/ interpretable range CS1>95%
TPM4	95.00	0	NM_001145160.1/ interpretable range CS1>95%
TUBB1	95.00	0	NM_030773.3/ interpretable range CS1>95%
VIPAS39	95.00	0	NM_022067.3/ interpretable range CS1>95%
VKORC1	95.00	0	NM_024006.4/ interpretable range CS1>95%
VPS33B	95.00	0	NM_018668.4/ interpretable range CS1>95%
VWF	95.00	0	NM_000552.3/ interpretable range CS1>95%
WAS	95.00	0	NM_000377.2/ interpretable range CS1>95%