

HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL

DG 2.18 (156 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
<i>A2M</i>	99,90%	99,60%	100%	100%	No OMIM disease ID
<i>ABCG5</i>	100%	100%	100%	100%	Sitosterolemia 2, 618666
<i>ABCG8</i>	99,10%	97,30%	100%	100%	Sitosterolemia 1, 210250
<i>ACBD5</i>	100%	99,20%	100%	100%	Retinal dystrophy with leukodystrophy, 618863
<i>ACTB</i>	99,70%	96,10%	100%	100%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
<i>ACTN1</i>	100%	100%	100%	100%	Bleeding disorder, platelet-type, 15, 615193
<i>ACVRL1</i>	100%	98,90%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
<i>ADAMTS13</i>	97,10%	93,80%	99,90%	99,50%	Thrombotic thrombocytopenic purpura, hereditary, 274150
<i>ANKRD26</i>	95,00%	89,30%	97,20%	97,20%	Thrombocytopenia 2, 188000
<i>ANO6</i>	99,90%	98,70%	100%	100%	Scott syndrome, 262890
<i>AP3B1</i>	99,20%	95,80%	100%	100%	Hermansky-Pudlak syndrome 2, 608233
<i>AP3D1</i>	99,80%	98,60%	100%	100%	?Hermansky-Pudlak syndrome 10, 617050
<i>ARPC1B</i>	100%	100%	100%	100%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
<i>BLOC1S3</i>	98,50%	81,30%	100%	100%	Hermansky-Pudlak syndrome 8, 614077
<i>BLOC1S6</i>	99,90%	97,10%	94,90%	94,90%	?Hermansky-pudlak syndrome 9, 614171
<i>BRAF</i>	95,60%	85,10%	100%	100%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
<i>C3</i>	99,90%	99,20%	100%	100%	C3 deficiency, 613779
<i>CALR</i>	94,80%	89,10%	100%	100%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950

<i>CBL</i>	97,30%	97,10%	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
<i>CD36</i>	99,70%	98,70%	100%	100%	Platelet glycoprotein IV deficiency, 608404
<i>CD46</i>	99,90%	99,40%	100%	100%	No OMIM disease ID
<i>CDC42</i>	97,80%	90,70%	100%	100%	Takenouchi-Kosaki syndrome, 616737
<i>CFB</i>	100%	100%	100%	100%	?Complement factor B deficiency, 615561
<i>CFH</i>	99,90%	99,00%	100%	99,90%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
<i>CFHR1</i>	96,40%	94,90%	95,40%	93,80%	No OMIM disease ID
<i>CFHR3</i>	94,00%	92,20%	96,00%	95,20%	No OMIM disease ID
<i>CFI</i>	99,20%	96,80%	100%	100%	Complement factor I deficiency, 610984
<i>CHST14</i>	99,90%	98,90%	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
<i>COL1A1</i>	99,90%	98,60%	100%	100%	Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
<i>COL3A1</i>	99,60%	97,60%	100%	100%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
<i>COL5A1</i>	98,80%	98,00%	100%	99,90%	Ehlers-Danlos syndrome, classic type, 1, 130000
<i>COL5A2</i>	100%	99,50%	100%	100%	Ehlers-Danlos syndrome, classic type, 2, 130010
<i>CTLA4</i>	100%	100%	100%	100%	Autoimmune lymphoproliferative syndrome, type V, 616100
<i>CYCS</i>	99,10%	94,90%	100%	100%	Thrombocytopenia 4, 612004
<i>DGKE</i>	99,80%	98,10%	100%	100%	Nephrotic syndrome, type 7, 615008
<i>DIAPH1</i>	99,80%	99,00%	99,50%	97,90%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
<i>DNASE1</i>	100%	99,90%	100%	100%	No OMIM disease ID
<i>DTNBP1</i>	99,80%	98,70%	100%	100%	Hermansky-Pudlak syndrome 7, 614076
<i>ENG</i>	99,60%	96,00%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
<i>EPHB2</i>	98,10%	98,10%	99,80%	98,80%	?Bleeding disorder, platelet-type, 22, 618462
<i>ETV6</i>	100%	99,90%	100%	100%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
<i>F10</i>	99,80%	99,10%	100%	100%	Factor X deficiency, 227600
<i>F11</i>	100%	100%	100%	100%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
<i>F12</i>	99,90%	98,80%	100%	100%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618

<i>F13A1</i>	100%	100%	100%	100%	Factor XIII A deficiency, 613225
<i>F13B</i>	98,70%	93,50%	100%	100%	Factor XIII B deficiency, 613235
<i>F2</i>	99,90%	97,90%	100%	100%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050
<i>F2RL3</i>	100%	100%	100%	100%	No OMIM disease ID
<i>F5</i>	99,40%	98,50%	100%	100%	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055
<i>F7</i>	100%	100%	100%	100%	Factor VII deficiency, 227500
<i>F8</i>	99,50%	98,30%	100%	99,90%	Hemophilia A, 306700
<i>F9</i>	99,60%	97,60%	99,90%	98,80%	Thrombophilia, X-linked, due to factor IX defect, 300807 Hemophilia B, 306900
<i>FBN1</i>	100%	99,90%	100%	100%	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
<i>FCGR2A</i>	100%	100%	100%	100%	No OMIM disease ID
<i>FCGR2B</i>	99,50%	95,40%	100%	100%	No OMIM disease ID
<i>FCGR2C</i>	98,90%	98,60%	98,10%	97,20%	No OMIM disease ID
<i>FERMT3</i>	100%	100%	100%	100%	Leukocyte adhesion deficiency, type III, 612840
<i>FGA</i>	99,10%	97,20%	100%	100%	Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Hypodysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
<i>FGB</i>	99,80%	99,10%	100%	100%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
<i>FGG</i>	99,70%	98,20%	100%	100%	Hypofibrinogenemia, congenital, 202400 Hypodysfibrinogenemia, 616004 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
<i>FLI1</i>	99,50%	98,20%	100%	100%	Bleeding disorder, platelet-type, 21, 617443
<i>FLNA</i>	100%	99,90%	100%	100%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048

					Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
<i>FYB1</i>	99,40%	97,00%	100%	100%	Thrombocytopenia 3, 273900
<i>GATA1</i>	99,80%	98,40%	100%	100%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Thrombocytopenia with beta-thalassemia, X-linked, 314050
<i>GATA2</i>	100%	98,30%	100%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
<i>GBA</i>	100%	100%	100%	100%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
<i>GDF2</i>	100%	100%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
<i>GFI1B</i>	98,50%	96,70%	100%	100%	Bleeding disorder, platelet-type, 17, 187900
<i>GGCX</i>	100%	99,90%	100%	100%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
<i>GNE</i>	100%	99,70%	100%	100%	Sialuria, 269921 Nonaka myopathy, 605820
<i>GP1BA</i>	98,60%	95,90%	100%	100%	Bernard-Soulier syndrome, type A1 (recessive), 231200 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
<i>GP1BB</i>	72,90%	59,60%	99,50%	95,00%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
<i>GP6</i>	100%	100%	94,90%	91,70%	Bleeding disorder, platelet-type, 11, 614201
<i>GP9</i>	96,50%	89,30%	100%	100%	Bernard-Soulier syndrome, type C, 231200
<i>HABP2</i>	100%	99,90%	100%	100%	No OMIM disease ID
<i>HOXA11</i>	97,10%	87,50%	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
<i>HPS1</i>	100%	100%	100%	100%	Hermansky-Pudlak syndrome 1, 203300
<i>HPS3</i>	99,70%	97,50%	100%	100%	Hermansky-Pudlak syndrome 3, 614072
<i>HPS4</i>	100%	100%	100%	100%	Hermansky-Pudlak syndrome 4, 614073

<i>HPS5</i>	100%	99,70%	100%	100%	Hermansky-Pudlak syndrome 5, 614074
<i>HPS6</i>	97,10%	88,90%	100%	100%	Hermansky-Pudlak syndrome 6, 614075
<i>HRG</i>	95,00%	94,20%	100%	100%	Thrombophilia due to HRG deficiency, 613116
<i>IKZF5</i>	100%	100%	100%	100%	No OMIM disease ID
<i>ITGA2</i>	99,60%	98,10%	100%	100%	No OMIM disease ID
<i>ITGA2B</i>	99,70%	97,80%	100%	100%	Glanzmann thrombasthenia, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
<i>ITGB3</i>	100%	99,40%	100%	100%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0
<i>JAK2</i>	98,10%	95,80%	100%	100%	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
<i>KDSR</i>	100%	99,50%	100%	100%	Erythrokeratoderma variabilis et progressiva 4, 617526
<i>KLKB1</i>	100%	99,50%	100%	100%	Fletcher factor (prekallikrein) deficiency, 612423
<i>KNG1</i>	100%	100%	100%	100%	No OMIM disease ID
<i>KRAS</i>	99,50%	96,90%	100%	100%	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
<i>LMAN1</i>	99,80%	99,20%	100%	100%	Combined factor V and VIII deficiency, 227300
<i>LYST</i>	99,60%	98,30%	100%	100%	Chediak-Higashi syndrome, 214500
<i>LZTR1</i>	100%	99,90%	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
<i>MASTL</i>	100%	100%	100%	100%	No OMIM disease ID
<i>MCFD2</i>	99,50%	96,90%	100%	100%	Factor V and factor VIII, combined deficiency of, 613625

<i>MECOM</i>	100%	99,90%	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
<i>MLPH</i>	100%	98,80%	100%	100%	Griscelli syndrome, type 3, 609227
<i>MPIG6B</i>	100%	100%	100%	100%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
<i>MPL</i>	100%	99,50%	100%	100%	Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498 Myelofibrosis with myeloid metaplasia, somatic, 254450
<i>MTHFR</i>	97,30%	96,00%	100%	100%	Homocystinuria due to MTHFR deficiency, 236250
<i>MYH9</i>	100%	99,30%	100%	100%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
<i>MYO5A</i>	99,80%	98,90%	100%	100%	Griscelli syndrome, type 1, 214450
<i>NBEA</i>	92,00%	90,60%	100%	100%	No OMIM disease ID
<i>NBEAL2</i>	99,40%	99,30%	100%	100%	Gray platelet syndrome, 139090
<i>NRAS</i>	100%	100%	100%	100%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
<i>ORAI1</i>	95,80%	92,80%	97,20%	92,40%	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
<i>P2RY12</i>	100%	100%	100%	100%	Bleeding disorder, platelet-type, 8, 609821
<i>TALDO1</i>	100%	97,90%	100%	100%	Transaldolase deficiency, 606003
<i>PIGA</i>	93,80%	86,70%	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
<i>PLA2G4A</i>	99,90%	99,40%	100%	100%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
<i>PLA2G7</i>	99,90%	99,00%	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278
<i>PLAT</i>	100%	99,10%	100%	100%	No OMIM disease ID
<i>PLAU</i>	100%	99,80%	100%	100%	Quebec platelet disorder, 601709
<i>PLG</i>	87,80%	87,50%	100%	100%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
<i>PRKACG</i>	100%	99,90%	100%	100%	?Bleeding disorder, platelet-type, 19, 616176
<i>PROC</i>	100%	100%	100%	100%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
<i>PROS1</i>	98,80%	96,10%	100%	100%	Thrombophilia due to protein S deficiency, autosomal recessive, 614514 Thrombophilia due to protein S deficiency, autosomal dominant, 612336

<i>PROZ</i>	100%	99,80%	100%	100%	No OMIM disease ID
<i>PTGS1</i>	100%	99,80%	100%	100%	No OMIM disease ID
<i>PTPN11</i>	99,10%	93,70%	100%	100%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
<i>PTPN22</i>	99,50%	97,10%	100%	100%	No OMIM disease ID
<i>PTPRJ</i>	97,70%	97,20%	99,90%	99,60%	Colon cancer, somatic, 114500
<i>RAB27A</i>	100%	100%	100%	100%	Griscelli syndrome, type 2, 607624
<i>RAF1</i>	100%	100%	100%	100%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
<i>RASGRP2</i>	99,70%	97,30%	100%	100%	?Bleeding disorder, platelet-type, 18, 615888
<i>RBM8A</i>	99,80%	97,90%	100%	100%	Thrombocytopenia-absent radius syndrome, 274000
<i>RIT1</i>	100%	100%	100%	100%	Noonan syndrome 8, 615355
<i>RNU4ATAC</i>	NC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
<i>RUNX1</i>	99,30%	94,90%	100%	100%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
<i>SERPINC1</i>	100%	100%	100%	100%	Thrombophilia due to antithrombin III deficiency, 613118
<i>SERPIND1</i>	100%	100%	100%	100%	Thrombophilia due to heparin cofactor II deficiency, 612356
<i>SERPINE1</i>	100%	100%	100%	100%	Plasminogen activator inhibitor-1 deficiency, 613329
<i>SERPINF2</i>	100%	99,80%	100%	100%	Alpha-2-plasmin inhibitor deficiency, 262850
<i>SH2B3</i>	99,40%	95,10%	100%	99,90%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
<i>SLFN14</i>	100%	100%	100%	100%	Bleeding disorder, platelet-type, 20, 616913
<i>SOS1</i>	99,80%	98,40%	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
<i>SOS2</i>	100%	99,20%	100%	100%	Noonan syndrome 9, 616559
<i>SRC</i>	100%	99,80%	100%	100%	Colon cancer, advanced, somatic, 114500 ?Thrombocytopenia 6, 616937
<i>STIM1</i>	99,80%	98,00%	100%	100%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
<i>STXBP2</i>	82,10%	79,70%	99,30%	97,10%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
<i>TBX1</i>	86,90%	79,50%	94,10%	90,80%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400

					Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
<i>TBXA2R</i>	97,60%	93,80%	99,80%	98,00%	No OMIM disease ID
<i>TBXAS1</i>	100%	100%	100%	100%	Ghosal hematodiaphyseal syndrome, 231095
<i>THBD</i>	100%	99,70%	100%	100%	Thrombophilia due to thrombomodulin defect, 614486
<i>THPO</i>	100%	99,50%	100%	100%	Thrombocythemia 1, 187950
<i>TNXB</i>	99,60%	95,10%	100%	100%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
<i>TPM4</i>	94,10%	79,90%	100%	100%	No OMIM disease ID
<i>TREX1</i>	100%	100%	100%	100%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
<i>TUBB1</i>	100%	100%	100%	100%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
<i>VIPAS39</i>	100%	100%	100%	100%	Arthrogyriposis, renal dysfunction, and cholestasis 2, 613404
<i>VKORC1</i>	100%	100%	93,00%	93,00%	Warfarin resistance, 122700 Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473
<i>VPS33B</i>	100%	100%	100%	100%	Arthrogyriposis, renal dysfunction, and cholestasis 1, 208085
<i>VWF</i>	99,80%	98,60%	100%	100%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
<i>WAS</i>	95,90%	85,30%	100%	99,80%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
<i>WIPF1</i>	100%	99,90%	100%	100%	?Wiskott-Aldrich syndrome 2, 614493

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. *Nucleic Acids Res.* 2015 Jan 43(Database issue):D1079-85.

Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.

TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with coverage denoting NC are non-DNA coding genes.

non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.

This list is accurate for panel version DG 2.18

Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors