# HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 2.12 (133 genes)

| *Gene* | *Median* | *% covered > 10x* | *% covered > 20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|
| A2M | 119.2 | 99 | 98 | Alpha-2-macroglobulin deficiency, 614036<br>{Alzheimer disease, susceptibility to}, 104300 |
| ABCG5 | 145.4 | 100 | 99 | Sitosterolemia, 210250 |
| ABCG8 | 148.7 | 99 | 96 | Sitosterolemia, 210250<br>{Gallbladder disease 4}, 611465 |
| ACTN1 | 143.9 | 100 | 99 | Bleeding disorder, platelet-type, 15, 615193 |
| ACVRL1 | 123 | 99 | 98 | Telangiectasia, hereditary hemorrhagic, type 2, 600376 |
| ADAMTS13 | 100 | 96 | 91 | Thrombotic thrombocytopenic purpura, familial, 274150 |
| ANKRD26 | 79.7 | 89 | 77 | Thrombocytopenia 2, 188000 |
| ANO6 | 137.2 | 98 | 92 | Scott syndrome, 262890 |
| AP3B1 | 95 | 97 | 90 | Hermansky-Pudlak syndrome 2, 608233 |
| BLOC1S3 | 28.7 | 88 | 64 | Hermansky-Pudlak syndrome 8, 614077 |
| BLOC1S6 | 97.3 | 98 | 91 | Hermansky-pudlak syndrome 9, 614171 |
| BRAF | 74.3 | 87 | 77 | Adenocarcinoma of lung, somatic, 211980<br>Cardiofaciocutaneous syndrome, 115150<br>Colorectal cancer, somatic<br>LEOPARD syndrome 3, 613707<br>Melanoma, malignant, somatic<br>Nonsmall cell lung cancer, somatic<br>Noonan syndrome 7, 613706 |
| C3 | 145.8 | 100 | 99 | C3 deficiency, 613779<br>{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925<br>{Macular degeneration, age-related, 9}, 611378 |
| CALR | 113.6 | 99 | 97 | Myelofibrosis, somatic, 254450<br>Thrombocythemia, somatic, 187950 |
| CBL | 133.2 | 99 | 98 | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563<br>?Juvenile myelomonocytic leukemia,607785 |

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| CD36 | 123.2 | 99 | 95 | Platelet glycoprotein IV deficiency, 608404<br>[Macrothrombocytopenia]<br>{Coronary heart disease, susceptibility to, 7}, 610938<br>{Malaria, cerebral, reduced risk of}, 611162<br>{Malaria, cerebral, susceptibility to}, 611162 |
| CD46 | 115.2 | 97 | 93 | {Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 |
| CFB | 33.4 | 23 | 23 | ?Complement factor B deficiency, 615561<br>{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924<br>{Macular degeneration, age-related, 14, reduced risk of}, 615489 |
| CFH | 183.1 | 98 | 95 | Basal laminar drusen, 126700<br>Complement factor H deficiency, 609814<br>{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400<br>{Macular degeneration, age-related, 4}, 610698 |
| CFHR1 | 234.8 | 95 | 94 | {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400<br>{Macular degeneration, age-related, reduced risk of}, 603075 |
| CFHR3 | 103 | 92 | 87 | {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400<br>{Macular degeneration, age-related, reduced risk of}, 603075 |
| CFI | 152.5 | 98 | 96 | Complement factor I deficiency, 610984<br>{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923<br>{Macular degeneration, age-related, 13, susceptibility to}, 615439 |
| COL3A1 | 104.2 | 97 | 92 | Ehlers-Danlos syndrome, type IV, 130050 |
| CTLA4 | 194.1 | 100 | 100 | Autoimmune lymphoproliferative syndrome, type V, 616100<br>{Celiac disease, susceptibility to, 3}, 609755<br>{Diabetes mellitus, insulin-dependent, 12}, 601388<br>{Hashimoto thyroiditis}, 140300<br>{Systemic lupus erythematosus, susceptibility to}, 152700 |
| CYCS | 72.1 | 99 | 94 | Thrombocytopenia 4, 612004 |
| DGKE | 142.3 | 99 | 95 | Nephrotic syndrome, type 7, 615008<br>{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 |
| DIAPH1 | 120.5 | 99 | 97 | Deafness, autosomal dominant 1, 124900<br>Seizures, cortical blindness, microcephaly syndrome, 616632 |
| DNASE1 | 201.3 | 100 | 100 | {Systemic lupus erythematosus, susceptibility to}, 152700 |
| DTNBP1 | 113.9 | 99 | 95 | Hermansky-Pudlak syndrome 7, 614076 |
| ENG | 128.9 | 97 | 93 | Telangiectasia, hereditary hemorrhagic, type 1, 187300 |

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| ETV6 | 140.3 | 100 | 99 | Leukemia, acute myeloid, somatic, 601626<br>Thrombocytopenia 5, 616216 |
| F10 | 185.4 | 99 | 98 | Factor X deficiency, 227600 |
| F11 | 155 | 100 | 100 | Factor XI deficiency, autosomal dominant, 612416<br>Factor XI deficiency, autosomal recessive, 612416 |
| F12 | 111.6 | 100 | 99 | Angioedema, hereditary, type III, 610618<br>Factor XII deficiency, 234000 |
| F13A1 | 147.4 | 100 | 99 | Factor XIIIA deficiency, 613225<br>{Myocardial infarction, protection against}, 608446<br>{Venous thrombosis, protection against}, 188050 |
| F13B | 113.4 | 96 | 87 | Factor XIIIB deficiency, 613235 |
| F2 | 124.4 | 99 | 98 | Dysprothrombinemia, 613679<br>Hypoprothrombinemia, 613679<br>Thrombophilia due to thrombin defect, 188050<br>{Pregnancy loss, recurrent, susceptibility to, 2}, 614390<br>{Stroke, ischemic, susceptibility to}, 601367 |
| F2RL3 | 103 | 100 | 99 | No OMIM phenotype<br>Impaired thrombin-induced platelet response (Bianchi et al. (2016) Blood 127(10):1249-1259) |
| F5 | 173.6 | 99 | 97 | Factor V deficiency, 227400<br>Thrombophilia due to activated protein C resistance, 188055<br>{Budd-Chiari syndrome}, 600880<br>{Pregnancy loss, recurrent, susceptibility to, 1}, 614389<br>{Stroke, ischemic, susceptibility to}, 601367<br>{Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 |
| F7 | 164.4 | 100 | 98 | Factor VII deficiency, 227500<br>{Myocardial infarction, decreased susceptibility to}, 608446 |
| F8 | 118.8 | 99 | 98 | Hemophilia A, 306700 |
| F9 | 145.5 | 99 | 95 | Hemophilia B, 306900<br>Thrombophilia, X-linked, due to factor IX defect, 300807<br>{Deep venous thrombosis, protection against}, 300807<br>{Warfarin sensitivity}, 122700 |
| FCGR2A | 245.1 | 100 | 100 | {Lupus nephritis, susceptibility to}, 152700<br>{Malaria, severe, susceptibility to}, 611162<br>{Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 |

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| FCGR2B | 166.1 | 99 | 97 | {Malaria, resistance to}, 611162<br>{Systemic lupus erythematosus, susceptibility to}, 152700 |
| FCGR2C | 208 | 98 | 97 | Thrombocytopenic purpura, autoimmune, 188030 |
| FERMT3 | 122.6 | 100 | 98 | Leukocyte adhesion deficiency,type III,612840 |
| FGA | 157.2 | 99 | 96 | Afibrinogenemia, congenital, 202400<br>Amyloidosis, familial visceral, 105200<br>Dysfibrinogenemia, congenital, 616004<br>Hypodysfibrinogenemia, congenital, 616004 |
| FGB | 190.7 | 99 | 97 | Afibrinogenemia, congenital, 202400<br>Dysfibrinogenemia, congenital, 616004<br>Hypofibrinogenemia, congenital, 202400 |
| FGG | 136.9 | 99 | 96 | Afibrinogenemia, congenital, 202400<br>Dysfibrinogenemia, congenital, 616004<br>Hypodysfibrinogenemia, 616004<br>Hypofibrinogenemia, congenital, 202400 |
| FLI1 | 189.8 | 99 | 97 | No OMIM phenotype<br>Platelet dense granule secretion defect,excessive bleeding (Stockley (2013) Blood 122,4090) |
| FLNA | 139.2 | 100 | 99 | Cardiac valvular dysplasia, X-linked, 314400<br>Congenital short bowel syndrome, 300048<br>FG syndrome 2, 300321<br>Frontometaphyseal dysplasia, 305620<br>Heterotopia, periventricular, 300049<br>Heterotopia, periventricular, ED variant, 300537<br>Intestinal pseudoobstruction, neuronal, 300048<br>Melnick-Needles syndrome, 309350<br>Otopalatodigital syndrome, type I, 311300<br>Otopalatodigital syndrome, type II, 304120<br>Terminal osseous dysplasia, 300244 |
| GATA1 | 84.2 | 99 | 95 | Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835<br>Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685<br>Thrombocytopenia with beta-thalassemia, X-linked, 314050<br>Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 |
| GFI1B | 189.6 | 100 | 100 | Bleeding disorder, platelet-type, 17, 187900 |
| GGCX | 115.4 | 100 | 99 | Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842<br>Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 |

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| GP1BA | 153.1 | 97 | 94 | Bernard-Soulier syndrome, type A1 (recessive), 231200<br>Bernard-Soulier syndrome, type A2 (dominant), 153670<br>von Willebrand disease, platelet-type, 177820<br>{Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 |
| GP1BB | 34.5 | 74 | 64 | Bernard-Soulier syndrome, type B, 231200<br>Giant platelet disorder, isolated, 231200 |
| GP6 | 136.8 | 100 | 100 | Bleeding disorder, platelet-type, 11, 614201 |
| GP9 | 123.1 | 96 | 89 | Bernard-Soulier syndrome, type C, 231200 |
| HABP2 | 136.3 | 100 | 99 | {?Thyroid cancer, nonmedullary, 5}, 616535<br>{Venous thromboembolism, susceptibility to}, 188050 |
| HOXA11 | 86.4 | 87 | 78 | Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 |
| HPS1 | 118 | 100 | 99 | Hermansky-Pudlak syndrome 1, 203300 |
| HPS3 | 135.2 | 99 | 96 | Hermansky-Pudlak syndrome 3, 614072 |
| HPS4 | 144.5 | 100 | 100 | Hermansky-Pudlak syndrome 4, 614073 |
| HPS5 | 133 | 99 | 98 | Hermansky-Pudlak syndrome 5, 614074 |
| HPS6 | 139.2 | 91 | 84 | Hermansky-Pudlak syndrome 6, 614075 |
| HRG | 178.5 | 95 | 94 | Thrombophilia due to elevated HRG, 613116<br>Thrombophilia due to HRG deficiency, 613116 |
| ITGA2 | 136.3 | 97 | 95 | ?Glycoprotein Ia deficiency, 614200 |
| ITGA2B | 119.9 | 99 | 97 | Bleeding disorder, platelet-type, 16, autosomal dominant, 187800<br>Glanzmann thrombasthenia, 273800<br>Thrombocytopenia, neonatal alloimmune, BAK antigen related |
| ITGB3 | 138.9 | 99 | 97 | Bleeding disorder, platelet-type, 16, autosomal dominant, 187800<br>Glanzmann thrombasthenia, 273800<br>Purpura, posttransfusion<br>Thrombocytopenia, neonatal alloimmune<br>{Myocardial infarction, susceptibility to}, 608446 |
| JAK2 | 90.5 | 95 | 94 | Erythrocytosis, somatic, 133100<br>Leukemia, acute myeloid, somatic, 601626<br>Myelofibrosis, somatic, 254450<br>Polycythemia vera, somatic, 263300<br>Thrombocythemia 3, 614521<br>{Budd-Chiari syndrome, somatic}, 600800 |
| KLKB1 | 143.5 | 99 | 96 | Fletcher factor (prekallikrein) deficiency, 612423 |

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| KNG1 | 193.7 | 100 | 100 | [High molecular weight kininogen deficiency], 228960<br>[Kininogen deficiency], 228960 |
| KRAS | 64.6 | 99 | 98 | Bladder cancer, somatic, 109800<br>Breast cancer, somatic, 114480<br>Cardiofaciocutaneous syndrome 2, 615278<br>Gastric cancer, somatic, 137215<br>Leukemia, acute myeloid, 601626<br>Lung cancer, somatic, 211980<br>Noonan syndrome 3, 609942<br>Pancreatic carcinoma, somatic, 260350<br>RAS-associated autoimmune leukoproliferative disorder, 614470<br>Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 |
| LMAN1 | 129.9 | 99 | 94 | Combined factor V and VIII deficiency, 227300 |
| LYST | 134.6 | 97 | 93 | Chediak-Higashi syndrome, 214500 |
| LZTR1 | 134.3 | 100 | 99 | Noonan syndrome 10, 616564<br>{Schwannomatosis-2, susceptibility to}, 615670 |
| MASTL | 130.5 | 100 | 99 | ?Thrombocytopenia-2, 188000 |
| MCFD2 | 101.5 | 99 | 98 | Factor V and factor VIII, combined deficiency of, 613625 |
| MECOM | 143.3 | 100 | 99 | Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 |
| MLPH | 95.2 | 99 | 95 | Griscelli syndrome, type 3, 609227 |
| MPL | 135.2 | 99 | 97 | Myelofibrosis with myeloid metaplasia, somatic, 254450<br>Thrombocythemia 2, 601977<br>Thrombocytopenia, congenital amegakaryocytic, 604498 |
| MTHFR | 131.3 | 100 | 100 | Homocystinuria due to MTHFR deficiency, 236250<br>{Neural tube defects, susceptibility to}, 601634<br>{Schizophrenia, susceptibility to}, 181500<br>{Thromboembolism, susceptibility to}, 188050<br>{Vascular disease, susceptibility to} |
| MYH9 | 130.8 | 99 | 98 | Deafness, autosomal dominant 17, 603622<br>Epstein syndrome, 153650<br>Fechtner syndrome, 153640<br>Macrothrombocytopenia and progressive sensorineural deafness, 600208<br>May-Hegglin anomaly, 155100<br>Sebastian syndrome, 605249 |
| MYO5A | 124.8 | 99 | 97 | Griscelli syndrome, type 1, 214450 |

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| NBEA | 128.3 | 90 | 89 | No OMIM phenotype<br>Autism, idiopathic (Castermans (2003) J Med Genet 40, 352)<br>?Schizophrenia (Fromer (2014) Nature 506, 179)<br>?Obesity, extreme (Mariman (2015) Physiol Genomics 47,225)<br>?Tetralogy of Fallot (Silversides (2012) PloS Genet 8) |
| NBEAL2 | 173 | 99 | 99 | Gray platelet syndrome, 139090 |
| NRAS | 188.2 | 100 | 100 | Colorectal cancer, somatic, 114500<br>Epidermal nevus, somatic, 162900<br>Melanocytic nevus syndrome, congenital, somatic, 137550<br>Neurocutaneous melanosis, somatic, 249400<br>Noonan syndrome 6, 613224<br>Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200<br>Thyroid carcinoma, follicular, somatic, 188470<br>?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 |
| P2RX1 | 115.3 | 100 | 99 | Bleeding disorder due to P2RX1 defect, somatic, 609821 |
| P2RY12 | 185.8 | 100 | 100 | Bleeding disorder, platelet-type, 8, 609821 |
| PLA2G4A | 134.2 | 99 | 98 | Phospholipase A2, group IV A, deficiency of |
| PLA2G7 | 124.4 | 99 | 97 | Platelet-activating factor acetylhydrolase deficiency, 614278<br>{Asthma, susceptibility to}, 600807<br>{Atopy, susceptibility to}, 147050 |
| PLAT | 98 | 100 | 99 | Hyperfibrinolysis, familial, due to increased release of PLAT, 612348<br>Thrombophilia, familial, due to decreased release of PLAT, 612348 |
| PLAU | 111.5 | 99 | 98 | Quebec platelet disorder, 601709<br>{Alzheimer disease, late-onset, susceptibility to}, 104300 |
| PLG | 115.4 | 87 | 86 | Dysplasminogenemia, 217090<br>Plasminogen deficiency, type I, 217090 |
| PRKACG | 218.1 | 100 | 99 | ?Bleeding disorder, platelet-type, 19, 616176 |
| PROC | 140.4 | 99 | 97 | Thrombophilia due to protein C deficiency, autosomal dominant, 176860<br>Thrombophilia due to protein C deficiency, autosomal recessive, 612304 |
| PROS1 | 101.1 | 96 | 91 | Thrombophilia due to protein S deficiency, autosomal dominant, 612336<br>Thrombophilia due to protein S deficiency, autosomal recessive, 614514 |
| PROZ | 131.4 | 99 | 98 | [Protein Z deficiency], 614024 |
| PTGS1 | 153.1 | 99 | 98 | No OMIM phenotype |

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| PTPN11 | 103.4 | 97 | 92 | LEOPARD syndrome 1, 151100<br>Leukemia, juvenile myelomonocytic, somatic, 607785<br>Metachondromatosis, 156250<br>Noonan syndrome 1, 163950 |
| PTPN22 | 134.4 | 98 | 91 | {Diabetes, type 1, susceptibility to}, 222100<br>{Rheumatoid arthritis, susceptibility to}, 180300<br>{Systemic lupus erythematosus susceptibility to}, 152700 |
| RAB27A | 144.1 | 100 | 99 | Griscelli syndrome, type 2, 607624 |
| RAF1 | 127.4 | 100 | 99 | Cardiomyopathy, dilated, 1NN, 615916<br>LEOPARD syndrome 2, 611554<br>Noonan syndrome 5, 611553 |
| RASGRP2 | 97.5 | 99 | 98 | ?Bleeding disorder, platelet-type, 18, 615888 |
| RBM8A | 106.6 | 100 | 99 | Thrombocytopenia-absent radius syndrome, 274000 |
| RIT1 | 165.5 | 100 | 100 | Noonan syndrome 8, 615355 |
| RUNX1 | 91.6 | 94 | 86 | Leukemia, acute myeloid, 601626<br>Platelet disorder, familial, with associated myeloid malignancy, 601399 |
| SELP | 125.8 | 100 | 99 | {Atopy, susceptibility to}, 147050 |
| SERPINC1 | 143.2 | 100 | 100 | Thrombophilia due to antithrombin III deficiency, 613118 |
| SERPIND1 | 181.9 | 100 | 100 | Thrombophilia due to heparin cofactor II deficiency,612356 |
| SERPINE1 | 155.2 | 100 | 100 | Plasminogen activator inhibitor-1 deficiency, 613329<br>{Transcription of plasminogen activator inhibitor, modulator of} |
| SERPINF2 | 144.1 | 99 | 99 | Alpha-2-plasmin inhibitor deficiency,262850 |
| SH2B3 | 97.5 | 90 | 79 | Erythrocytosis, somatic, 133100<br>Myelofibrosis, somatic, 254450<br>Thrombocythemia, somatic, 187950 |
| SLFN14 | 193 | 100 | 100 | Bleeding disorder,platelet-type,20,616913 |
| SOS1 | 95.5 | 96 | 90 | Noonan syndrome 4, 610733<br>?Fibromatosis, gingival, 1, 135300 |
| SOS2 | 97 | 98 | 92 | Noonan syndrome 9, 616559 |
| SRC | 105.5 | 99 | 97 | Colon cancer, advanced, somatic<br>?Thrombocytopenia 6,616937 |
| STIM1 | 127.2 | 100 | 99 | Immunodeficiency 10, 612783<br>Myopathy, tubular aggregate, 1 160565<br>Stormorken syndrome, 185070 |

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| STXBP2 | 125.3 | 98 | 97 | Hemophagocytic lymphohistiocytosis, familial, 5, 613101 |
| TALDO1 | 130.6 | 100 | 99 | Transaldolase deficiency, 606003 |
| TBX1 | 75.5 | 77 | 67 | Conotruncal anomaly face syndrome, 217095<br>DiGeorge syndrome, 188400<br>Tetralogy of Fallot, 187500<br>Velocardiofacial syndrome, 192430 |
| TBXA2R | 83.9 | 97 | 93 | {Bleeding disorder, platelet-type, 13, susceptibility to}, 614009 |
| TBXAS1 | 140.3 | 100 | 100 | Ghosal hematodiaphyseal syndrome, 231095<br>?Thromboxane synthase deficiency, 614158 |
| THBD | 108.4 | 99 | 97 | Thrombophilia due to thrombomodulin defect, 614486<br>{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 |
| THPO | 88.1 | 100 | 100 | Thrombocythemia 1, 187950 |
| TREX1 | 242.7 | 100 | 100 | Aicardi-Goutieres syndrome 1, dominant and recessive, 225750<br>Chilblain lupus, 610448<br>Vasculopathy, retinal, with cerebral leukodystrophy, 192315<br>{Systemic lupus erythematosus, susceptibility to}, 152700 |
| TUBB1 | 186.5 | 100 | 100 | Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112 |
| VIPAS39 | 144.7 | 100 | 100 | Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 |
| VKORC1 | 162.4 | 100 | 100 | Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473<br>Warfarin resistance, 122700 |
| VPS33B | 138.4 | 100 | 100 | Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 |
| VWF | 121.2 | 100 | 99 | von Willebrand disease, type 1, 193400<br>von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554<br>von Willibrand disease, type 3, 277480 |
| WAS | 66.6 | 88 | 78 | Neutropenia, severe congenital, X-linked, 300299<br>Thrombocytopenia, X-linked, 313900<br>Thrombocytopenia, X-linked, intermittent, 313900<br>Wiskott-Aldrich syndrome, 301000 |
| WIPF1 | 77.5 | 100 | 99 | ?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.*
*Median Coverage describes the average number of reads seen across 50 exomes.*
*% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions : April 14th, 2017.*
*This list is accurate for panel version DG 2.12*
*Ad 1. “No OMIM phenotype” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*