**University of Washington, New Exome Technology in (NEXT) Medicine Study**

**ACTIONABLE IF GENES = 117**

**AUTOSOMAL DOMINANT**

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| **Symbol** | **Disorder(s)** |
| [*ACTA2*](http://ncgenes.org/w/index.php?title=ACTA2&action=edit&redlink=1) | Aortic aneurysm, familial thoracic |
| *ACTC1* | Cardiomyopathy, dilated; Cardiomyopathy, familial hypertrophic; Left ventricular noncompaction |
| [*ACVRL1*](http://ncgenes.org/w/index.php?title=ACVRL1&action=edit&redlink=1) | Telangiectasia, hereditary hemorrhagic |
| [*APC*](http://ncgenes.org/w/index.php?title=APC&action=edit&redlink=1) | Familial adenomatous polyposis |
| *BMPR1A* | Juvenile polyposis syndrome |
| [*BRCA1*](http://ncgenes.org/w/index.php?title=BRCA1&action=edit&redlink=1) | Hereditary breast and ovarian cancer |
| [*BRCA2*](http://ncgenes.org/w/index.php?title=BRCA2&action=edit&redlink=1) | Hereditary breast and ovarian cancer |
| *CACNA1C* | SQTS-4 |
| *CACNA1S* | Malignant hyperthermia susceptibility |
| *CACNB2* | SQTS-5 |
| *CDC73* | Hyperparathyroidism-jaw tumor syndrome |
| [*CDH1*](http://ncgenes.org/w/index.php?title=CDH1&action=edit&redlink=1) | Hereditary diffuse gastric cancer |
| *CDKN2A* | Melanoma and pancreatic cancer (mild/moderate genetic risk gene) |
| *CNBPx* | Myotonic dystrophy 2 |
| *COL3A1* | Ehlers-Danlos syndrome |
| *DMPKx* | Myotonic dystrophy 1 |
| *DSC2* | Arrhythmogenic right ventricular dysplasia |
| *DSG2* | Arrhythmogenic right ventricular dysplasia; Cardiomyopathy, dilated |
| *DSP* | Arrhythmogenic right ventricular dysplasia |
| *ENG* | Hereditary Hemorrhagic Telangiectasia |
| *EPCAM* | Hereditary nonpolyposis colorectal cancer/Lynch syndrome |
| [*FBN1*](http://ncgenes.org/w/index.php?title=FBN1&action=edit&redlink=1) | Marfan syndrome |
| [*FH*](http://ncgenes.org/w/index.php?title=FH&action=edit&redlink=1) | Leiomyomatosis and renal cell cancer |
| *FLCN* | *Birt-Hogg-Dube syndrome* |
| *GCH1* | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia |
| *GREM1* | Hereditary Mixed Polyposis |
| *HMBS* | Porphyria, acute intermittent |
| *KCNE1* | LQTS-5 |
| *KCNE2* | LQTS-6 |
| *KCNH2* | LQTS-2, SQTS-1 |
| *KCNJ2* | SQTS-3 |
| *KCNQ1* | LQTS-1, SQTS-2 |
| *KIT* | Gastrointestinal stromal tumor |
| *LDLR* | Hypercholesterolemia, familial |
| *LMNA* | Cardiomyopathy |
| *MAX* | Susceptibility to pheochromocytoma |
| [*MEN1*](http://ncgenes.org/w/index.php?title=MEN1&action=edit&redlink=1) | Multiple endocrine neoplasia, type 1 |
| *MET* | Renal cell carcinoma, papillary, familial |
| [*MLH1*](http://ncgenes.org/w/index.php?title=MLH1&action=edit&redlink=1) | Hereditary nonpolyposis colorectal cancer/Lynch syndrome |
| [*MSH2*](http://ncgenes.org/w/index.php?title=MSH2&action=edit&redlink=1) | Hereditary nonpolyposis colorectal cancer/Lynch syndrome |
| [*MSH6*](http://ncgenes.org/w/index.php?title=MSH6&action=edit&redlink=1) | Hereditary nonpolyposis colorectal cancer/Lynch syndrome |
| *MYBPC3* | ?Cardiomyopathy, dilated; Cardiomyopathy, familial hypertrophic |
| [*MYH11*](http://ncgenes.org/w/index.php?title=MYH11&action=edit&redlink=1) | Aortic aneurysm, familial thoracic |
| *MYH7* | Cardiomyopathy, dilated; Cardiomyopathy, familial hypertrophic; Left ventricular noncompaction |
| *MYL2* | Cardiomyopathy, familial hypertrophic |
| *MYL3* | Cardiomyopathy, familial hypertrophic |
| *MYLK* | Aortic aneurysm, familial thoracic |
| *NF2* | Neurofibromatosis, type 2 |
| *PALB2* | Breast cancer, susceptibility; Pancreatic cancer, susceptibility |
| *PDGFRA* | Gastrointestinal stromal tumor |
| *PKP2* | Arrhythmogenic right ventricular dysplasia |
| *PLN* | Cardiomyopathy, dilated; Cardiomyopathy, familial hypertrophic |
| [*PMS2*](http://ncgenes.org/w/index.php?title=PMS2&action=edit&redlink=1) | Hereditary nonpolyposis colorectal cancer/Lynch syndrome |
| *POLD1* | Colorectal adenomas and carcinomas *\*only exonuclease domain* |
| *POLE* | Colorectal adenomas and carcinomas *\*only exonuclease domain* |
| [*PRKAG2*](http://ncgenes.org/w/index.php?title=PRKAG2&action=edit&redlink=1) | Wolff-Parkinson-White syndrome; Cardiomyopathy, hypertrophic 6 |
| *PRKAR1A* | **Carney** **complex**, type 1 |
| *PRKG1* | Familial thoracic aortic aneurysm *\*1 variant only c.530C>A, p.Arg177Gln* |
| *PROC* | Thrombophilia due to protein C deficiency |
| *PROS1* | Thrombophilia due to protein S deficiency |
| *PTCH1* | Basal cell nevus syndrome |
| [*PTEN*](http://ncgenes.org/w/index.php?title=PTEN&action=edit&redlink=1) | Cowden syndrome |
| *RBM20* | Cardiomyopathy, dilated |
| [*RET*](http://ncgenes.org/w/index.php?title=RET&action=edit&redlink=1) | Multiple endocrine neoplasia Type 2 |
| *RYR1* | Malignant hyperthermia susceptibility 1 |
| *RYR2* | Arrhythmogenic right ventricular dysplasia |
| *SCG5* | Hereditary Mixed Polyposis |
| *SCN5A* | LQTS-3; BRGDA 1 |
| [*SDHAF2*](http://ncgenes.org/w/index.php?title=SDHAF2&action=edit&redlink=1) | Hereditary paragangliomas and pheochromocytomas |
| [*SDHB*](http://ncgenes.org/w/index.php?title=SDHB&action=edit&redlink=1) | Hereditary paragangliomas and pheochromocytomas |
| [*SDHC*](http://ncgenes.org/w/index.php?title=SDHC&action=edit&redlink=1) | Hereditary paragangliomas and pheochromocytomas |
| [*SDHD*](http://ncgenes.org/w/index.php?title=SDHD&action=edit&redlink=1) | Hereditary paragangliomas and pheochromocytomas |
| *SERPINC1* | Thrombophilia due to antithrombin III deficiency |
| *SGCD* | Cardiomyopathy, dilated |
| *SMAD3* | Loeys-Dietz syndrome 1C |
| [*SMAD4*](http://ncgenes.org/w/index.php?title=SMAD4&action=edit&redlink=1) | Juvenile polyposis syndrome |
| [*STK11*](http://ncgenes.org/w/index.php?title=STK11&action=edit&redlink=1) | Peutz-Jeghers syndrome |
| *TGFB2* | Loeys-Dietz syndrome, type 4 |
| *TGFB3* | Arrhythmogenic right ventricular dysplasia 1 |
| [*TGFBR1*](http://ncgenes.org/w/index.php?title=TGFBR1&action=edit&redlink=1) | Loeys-Dietz syndrome, type 1A and type 2A |
| [*TGFBR2*](http://ncgenes.org/w/index.php?title=TGFBR2&action=edit&redlink=1) | Loeys-Dietz syndrome |
| *TMEM127* | Susceptibility to pheochromocytoma |
| *TMEM43* | Arrhythmogenic right ventricular dysplasia 5 |
| *TNNI3* | Cardiomyopathy, dilated; Cardiomyopathy, familial hypertrophic |
| *TNNT2* | Cardiomyopathy, dilated; Cardiomyopathy, familial hypertrophic |
| [*TP53*](http://ncgenes.org/w/index.php?title=TP53&action=edit&redlink=1) | Li-Fraumeni syndrome |
| *TPM1* | Cardiomyopathy, dilated; Cardiomyopathy, familial hypertrophic |
| *TSC1* | Tuberous sclerosis complex |
| *TSC2* | Tuberous sclerosis complex |
| [*VHL*](http://ncgenes.org/w/index.php?title=VHL&action=edit&redlink=1) | von Hippel-Lindau syndrome |

**AUTOSOMAL RECESSIVE** (*Homozygotes/compound heterozygotes)*

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| **Symbol** | **Disorder(s)** |
| [*ATP7B*](http://ncgenes.org/w/index.php?title=ATP7B&action=edit&redlink=1) | Wilson disease |
| *BCHE* | Pseudocholinesterase deficiency *(homozygotes for null alleles only)* |
| *BLM* | Bloom syndrome |
| *CASQ2* | Ventricular tachycardia, catecholaminergic polymorphic |
| [*CFTR*](http://ncgenes.org/w/index.php?title=CFTR&action=edit&redlink=1) | Cystic fibrosis |
| *CPT2* | CPT deficiency, hepatic, type II |
| [*F5*](http://ncgenes.org/w/index.php?title=F5&action=edit&redlink=1) | Factor V deficiency |
| *GAA* | Glycogen storage disease II |
| [*HAMP*](http://ncgenes.org/w/index.php?title=HAMP&action=edit&redlink=1) | Hemochromatosis, type 2B |
| [*HFE*](http://ncgenes.org/w/index.php?title=HFE&action=edit&redlink=1) | Hemochromatosis *\*C282Y only* (mild/moderate genetic risk gene) |
| [*HFE2*](http://ncgenes.org/w/index.php?title=HFE2&action=edit&redlink=1) | Hemochromatosis, type 2A |
| *IDUA* | Mucopolysaccharidosis |
| [*LDLRAP1*](http://ncgenes.org/w/index.php?title=LDLRAP1&action=edit&redlink=1) | Hypercholesterolemia, familial |
| [*PAH*](http://ncgenes.org/w/index.php?title=PAH&action=edit&redlink=1) | Phenylketonuria |
| [*PCBD1*](http://ncgenes.org/w/index.php?title=PCBD1&action=edit&redlink=1) | Hyperphenylalaninemia, BH4-deficient, D |
| [*PTS*](http://ncgenes.org/w/index.php?title=PTS&action=edit&redlink=1) | Hyperphenylalaninemia, BH4-deficient, A |
| [*QDPR*](http://ncgenes.org/w/index.php?title=QDPR&action=edit&redlink=1) | Hyperphenylalaninemia, BH4-deficient, C |
| *SERPINA1* | Emphysema due to AAT deficiency |
| *SLC25A13* | Citrullinemia, adult-onset type II |
| [*SLC37A4*](http://ncgenes.org/w/index.php?title=SLC37A4&action=edit&redlink=1) | Glycogen storage disease Ib; Glycogen storage disease Ic |
| [*SLC7A9*](http://ncgenes.org/w/index.php?title=SLC7A9&action=edit&redlink=1) | Cystinuria |
| *SLC3A1* | Cystinuria |
| [*MUTYH*](http://ncgenes.org/w/index.php?title=MUTYH&action=edit&redlink=1) | MYH Associated polyposis |

**X-LINKED**

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| **Symbol** | **Disorder(s)** |
| *DMD* | Becker muscular dystrophy; Cardiomyopathy, dilated; Duchenne muscular dystrophy |
| *EMD* | Emery-Dreifuss muscular dystrophy 1 |
| [*GLA*](http://ncgenes.org/w/index.php?title=GLA&action=edit&redlink=1) | Fabry disease |
| *OTC* | Ornithine transcarbamylase deficiency |