Veritas
The Genome Company

myGenome Standard/Premium Gene-Disease List

We aim to inform and educate our customers and partners with clear, concise and convenient resources. The Clinical Section of the myGenome test is designed to examine genes associated with a set of actionable genetic disorders as well as genes associated with common carrier conditions.

The list below shows all interpreted genes and their associated conditions/diseases. Both myGenome test options (Standard and Premium) include interpretation of 59 genes deemed medically actionable by the American College of Medical Genetics and Genomics and 24 genes associated with recessive carrier conditions.

If you have any questions, please contact us at 1 (888) 507-6619 or email clinical@veritasgenetics.com.

Clinically Actionable Disease*	Genes
Cancer Susceptibility	
Hereditary Breast Cancer (45 genes) • Hereditary Breast and Ovarian Cancer syndrome • PTEN Hamartoma tumor syndrome • Li-Fraumeni syndrome • Reproductive cancers (gynecological, prostate)	*BRCA1, BRCA2, PTEN, TP53,* *ABRAXAS1, AKT1, AR, ATM, BARD1, BRIP1, CD82, CHEK2, CYP19A1, DICER1, ESR1, GEN1, HIP1, HMMR, HOXB13, KLLN, LSP1, MAP3K1, MRE11, MSR1, MXI1, NBN, NQO2, OPCML, PALB2, PHB, PIKC3A, PPM1D, RBBP8, RAD50, RAD51, RAD51C, RAD51D, RAD54L, RECQL, RECQL4, RINT1, SEC23B, SMARCA4, TOX3, XRCC2*
Hereditary Gastrointestinal Polyposis and Cancer (37 genes) • Lynch syndrome • Familial adenomatous polyposis • MUTYH-Associated Polyposis • Juvenile Polyposis • Peutz-Jeghers syndrome • Hereditary Pancreatitis	*APC, BMPR1A, MLH1, MSH2, MSH6, MUTYH, PMS2, SMAD4, STK11,* *AURKA, AXIN2, BUB1B, CCND1, CDH1, CTNNA1, CTNNB1, CTRC, ENG, EPCAM, FAN1, GALNT12, GREM1, KIT, MLH3, MSH3, NTHL1, ODC1, PALLD, PDGFRA, PLA2G2A, PMS1, POLD1, POLE, PRSS1, RPS20, SPINK1, TLR2*
Neurocutaneous and Ocular (6 genes) • Neurofibromatosis Types 1 & 2 • Tuberous Sclerosis Complex Types 1 & 2 • Retinoblastoma	*NF2, RB1, TSC1, TSC2,* *NF1, SMARCB1*
Neuro/endocrine and Renal (31 genes) • Familial Medullary Thyroid Cancer • Hereditary Paraganglioma-Pheochromocytoma syndrome • Multiple Endocrine Neoplasia types 1 & 2 • von Hippel-Lindau syndrome • WT1-related Wilm's tumor	*MEN1, RET, SDHAF2, SDHB, SDHC, SDHD, VHL, WT1,* *AIP, BAP1, CDC73,CDKN1B, CDKN1C, DIS3L2, EGLN1, EGLN2, EPAS1, FH, FLCN, H19, HABP2, KIF1B, MAX, MET, MITF, PRKAR1A, SDHA, SRGAP1, TMEM127, TRIM37, TSHR*
Other cancers (115 genes) • Skin • Hemotological • Solid Tumors	*ACD, ALK, ARID5B, ATR, CASP8, CBX8, CDK4, CDKN2A, CEBPA,CEBPE, CEP57, CREBBP, CTC1, CTLA4, CYLD, CYP19A1, DDB2, DDR2, DDX41, DKC1, DOCK8, EGFR, ELANE, EP300, ERBB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXO1, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FAT1, FGFR2, G6PC3, GATA1, GATA2, GPC3, HAX1, HMBS, HNF1A, IKZF1, ITK, JAK2, MC1R, MGMT, MPL, MTAP, NHP2, NOP10, NSD1, PARN, PAX5, PHOX2B, PICALM, POLH, POT1, PRF1, PRKDC, PTCH1, PTCH2, RAD51B, RBM15, RHBDF2, ROBO2, ROS1, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, RUNX1, SBDS, SH2D1A, SLX4, SMARCE1, STAT3, SUFU, TERC, TERT, TINF2, TYR, USB1, UROD, WAS, WRAP53, WRN, XPA, XPC, XRCC3*
Cardiovascular	
Cardiomyopathy (78 genes) • Hypertrophic cardiomyopathy • Dilated cardiomyopathy • Arrhythmogenic right ventricular cardiomyopathy	*ACTC1, DSC2, DSG2, DSP, GLA, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PRKAG2, TMEM43, TNNI3, TNNT2, TPM1,* *ACTA1, ACTN2, ALMS1, ANKRD1, BAG3, CALR3, CAV3, CAVIN4, CHRM2, COX15, CRYAB, CSRP3, CTF1, DES, DMD, DOLK, DPP6, DTNA, EMD, EYA4, FHL1, FHL2, GATAD1, HADHA, ILK, JPH2, JUP, KLF10, LAMA4, LAMP2, LDB3, MIB1, MYH6, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEBL, NEXN, PDLIM3, PLN, PRDM16, PSEN1, PSEN2, RBM20, SCO2, SELENON, SGCB, SGCD, SLC25A13, SLC25A4, TAZ, TCAP, TMPO, TNNC1, TRIM63, TTN, TTR, TXNRD2, VCL, ZBTB17*
RASopathy (13 genes) • Noonan syndrome • Costello syndrome • Cardiofaciocutaneous syndrome	*BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1*
Channelopathy and Arrhythmia (34 genes) • Romano-Ward Long QT Syndromes Types 1, 2, and 3 • Brugada syndrome • Catecholaminergic polymorphic ventricular tachycardia	*KCNH2, KCNQ1, RYR2, SCN5A,* *ABCC9, AKAP9, ANK2, CACNA1C, CACNB2, CALM1, CALM2, CASQ2, GJA5, GPD1L, HCN4, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNJ2, KCNJ5, KCNJ8, NPPA, RANGRF, SCN1B, SCN2B, SCN3B, SCN4B, SLMAP, SNTA1, TRDN, TRPM4*
Familial Hypercholesterolemia (14 genes)	*APOB, LDLR, PCSK9,* *ABCG5, ABCG8, APOA5, APOC2, APOE, CETP, CREB3L3, GPIHBP1, LDLRAP1, LMF1, SREBF2*

Connective Tissue	
Marfan and Related Conditions (21 genes) • Marfan Syndrome • Loeys-Dietz Syndromes • Familial Thoracic Aortic Aneurysms and Dissections	*ACTA2, FBN1, MYH11, SMAD3, TGFBR1, TGFBR2,* *CCM2, ELN, FBN2, LTBP2, MYLK, NKX2-5, NODAL, NOTCH1, NOTCH3, PDCD10, PRKG1, PRNP, SLC2A10, TGFB2, TGFB3*
Ehlers-Danlos syndrome, vascular and non-vascular types (17 genes)	*COL3A1,* *ADAMTS2, B3GALT6, B4GALT7, CHST14, COL1A1, COL1A2, COL5A1, COL5A2, DSE, EFEMP2, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469*
Other actionable diseases	
Malignant hyperthermia susceptibility (3 genes)	*CACNA1S, RYR1,* *CACNA2D1*
Ornithine transcarbamylase deficiency (1 gene)	*OTC*
Wilson's disease (1 gene)	*ATP7B*
Other (29 genes)	*ABCD1, ACVRL1, APOA4, CASR, CRELD1, EGR2, F5, F8, F9, FKRP, FXN, GCKR, GDF2, HSPB8, JAG1, LITAF, MPZ, MYCN, NEFL, PKD2, PMP22, PRKN, SALL4, SERPINA1, TBX20, TBX3, TBX5, ZHX3, ZIC3*
CARRIER	**Genes**
Autosomal recessive conditions	
Beta-thalassemia; Bloom syndrome; Canavan disease; Congenital bilateral absence of vas deferens; Cystic fibrosis; Deafness; Diabetes mellitus (neonatal); Familial dysautonomia; Familial hyperinsulinism; Fanconi anemia; Galactosemia; Glycogen storage disease; GM1-gangliosidosis; Maple syrup urine disease; Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency; Mucopolysaccharidosis type IVB (Morquio); Mucolipidosis IV; Niemann-Pick disease; Phenylketonuria/Hyperphenylalaninemia; Retinitis pigmentosa; Sickle cell anemia; Smith-Lemli-Opitz syndrome; Tay-Sachs disease; Usher syndrome **(24 diseases)**	*ABCC8, ACADM, ASPA, BCKDHA, BCKDHB, BLM, CFTR, CLRN1, DHCR7, ELP1, G6PC, GALK1, GALT, GLB1, HBB, HEXA, MCOLN1, MYO7A, PAH, SMPD1, USH2A*
3-methylglutaconic aciduria; Achondrogenesis; Achromatopsia; Amyotrophic lateral sclerosis (juvenile); Anauxetic dysplasia; Andermann syndrome; Arthrogryposis (lethal, with anterior horn cell disease); Asparagine synthetase deficiency; Aspartylglucosaminuria; Autoimmune polyendocrinopathy syndrome (ACEPED); Bardet-Biedl syndrome; Biotinidase deficiency; Carnitine palmitoyl transferase deficiency; Cartilage-hair hypoplasia; Cerebrotendinous xanthomatosis; Ceroid neuronal lipofuscinosis disease (CLN1, CLN2, and CLN5); Charcot-Marie-Tooth disease; Cholesteryl ester storage disease; Chronic granulomatous disease; Cockayne syndrome; Combined oxidative phosphorylation deficiency; Cone-rod dystrophy; Congenital adrenal hyperplasia (11-beta-hydroxylase deficiency & 21-hydroxylase deficiency); Congenital disorder of glycosylation; Congenital glaucoma; Congenital myasthenic syndrome; Cystinosis; Dihydropyrimidine dehydrogenase deficiency; Dilated cardiomyopathy; Dystonia-1 (torsion); Ellis-van Creveld syndrome; Familial Mediterranean fever; Gaucher disease; Gitelman syndrome; Glucose-6-phosphate dehydrogenase deficiency; Glutaric acidemia IIC; Glutaricaciduria (type I); Hemochromatosis; Hereditary fructose intolerance; Hermansky-Pudlak syndrome; Holocarboxylase synthetase deficiency; Homocystinuria; Hypoaldosteronism (congenital); Hydrolethalus syndrome; Hypophosphatasia; Isovaleric academia; Joubert sydrome; Juvenile macular degeneration; Krabbe disease; Leber congenital amaurosis; Leigh syndrome; Lethal congenital contracture syndrome; Limb-girdle muscular dystrophy; Lipoprotein lipase deficiency; Malonic and Methylmalonic Aciduria (Combined); McArdle disease; Meckel syndrome; Megalencephalic leukoencephalopathy; Metachromatic Leukodystrophy; Metaphyseal dysplasia; Methylmalonic aciduria; Microcephaly, postnatal progressive); Mitochondrial complex I deficiency; Mitochondrial DNA depletion syndrome (Mitochondrial recessive ataxia syndrome; Muscular dystrophy (Miyoshi, dystroglycanopathy, merosin-deficient); Nephrotic syndrome; Nonaka myopathy; Omenn syndrome; Ornithine translocase deficiency (HHH syndrome); Osteopetrosis; Pendred syndrome; Polycystic kidney disease; Pontocerebellar hypoplasia; Primary carnitine deficiency; Progressive external ophthalmoplegia; Pseudocholinesterase deficiency; Pyruvate carboxylase deficiency; Sandhoff disease; Short/branched chain acyl-CoA dehydrogenase (SCAD) deficiency; Sialic acid storage disorder; Sialuria; Spastic ataxia; Spastic paraplegia; Spinocerebellar ataxia; Spondylocostal dysostosis; Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency; Weyers acrofacial dysostosis; Wolman disease; Zellweger spectrum disorder **(84 diseases)**	*ACADSB, ACADVL, ACSF3, AGA, AGL, AIRE, ALDOB, ALPL, ARSA, ASNS, BBS1, BBS2, BCHE, BTD, CBS, CERKL, CHRNE, CLN5, CNGB3, CPT1A, CPT2, CTNS, CYBA, CYP11B1, CYP11B2, CYP1B1, CYP21A2, CYP27A1, DCLRE1C, DNAJC19, DPYD, DYSF, ERCC6, ETFDH, EVC, EYS, F11, FAH, FAM161A, FKTN, G6PD, GAA, GALC, GBA, GBE1, GCDH, GJB2, GJB6, GLE1, GNE, HEXB, HFE, HLCS, HPS1, HYLS1, IVD, LAMA2, LIPA, LPL, LRPPRC, MED17, MEFV, MESP2, MKS1, MLC1, MPV17, MTHFR, MUT, NDRG1, NDUFS6, NPHS1, OPA3, PC, PCDH15, PEX12, PEX6, PKHD1, PMM2, POLG, PPT1, PYGM, RMRP, RPE65, SACS, SEPSECS, SGCG, SLC12A3, SLC12A6, SLC17A5, SLC22A5, SLC25A15, SLC26A2, SLC26A4, SPG11, TCIRG1, TECPR2, TOR1A,TPP1, TRMU, TSFM*

Red text: *Denotes the 59 genes and associated diseases/conditions considered medically actionable by the American College of Medical Genetics and Genomics included in both myGenome Standard and Premium.*

Blue text: *Denotes carrier genes and associated conditions included in both myGenome Standard and Premium.*

Black text: *Denotes genes and associated diseases/conditions included in myGenome Premium ONLY.*

**** Informed by the American College of Medical Genetics and Genomics (PMID: 23788249)***

DISCLAIMER: Many genes are associated with additional diseases/conditions, however, myGenome is NOT designed to cover them comprehensively. Additionally, for the carrier conditions listed, not all subtypes may be detectable. Please see the myGenome Technical Limitations for additional test information.

