Veritas
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myGenome Gene/Disease List: Consumer

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, PIK3CA, 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 (18 genes)	*COL3A1,* *ADAMTS2, B3GALT6, B4GALT7, CHST14, COL1A1, COL1A2, COL5A1, COL5A2, COL7A1, 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; Factor XI deficiency; 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; Hyperornithinemia-hyperammonemia-homocitrullinemia 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; Mitochondrial complex IV deficiency; 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; Transient infantile liver failure; Tyrosinemia deficiency; Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency; Weyers acrofacial dysostosis; Wolman disease; Zellweger spectrum disorder **(89 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, MMUT, 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: The 59 genes and associated diseases/conditions considered medically actionable by the American College of Medical Genetics and Genomics are included in both myGenome Standard and myGenome Premium.

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

DISCLAIMERS: Please note that many of the genes listed here may be associated with more than one condition/disease. For the purposes of this list, the predominant gene-disease association was used. For carrier conditions listed, not all subtypes may be detectable with myGenome Standard or Premium.

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