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myGenome Gene-Disease List

The Clinical Section of the myGenome test is designed to examine genes associated with a set of actionable genetic disorders. The list below shows all interpreted genes and their associated conditions/diseases. This test includes interpretation of 59 genes deemed medically actionable by the American College of Medical Genetics and Genomics*.

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

Gene	Medically Actionable Disease	Disease Category
BRCA1, BRCA2	Hereditary breast and ovarian cancer syndrome	Cancer susceptibility
TP53	Li-Fraumeni syndrome	Cancer susceptibility
STK11	Peutz-Jeghers syndrome	Cancer susceptibility
MLH1, MSH2, MSH6, PMS2	Lynch syndrome (Hereditary Non-Polyposis Colorectal Cancer syndrome)	Cancer susceptibility
APC	Familial adenomatous polyposis	Cancer susceptibility
MUTYH	MUTYH-associated polyposis	Cancer susceptibility
BMPR1A, SMAD4	Juvenile polyposis	Cancer susceptibility
VHL	Von Hippel-Lindau syndrome	Cancer susceptibility
MEN1	Multiple endocrine neoplasia type 1	Cancer susceptibility
RET	Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer	Cancer susceptibility
PTEN	PTEN hamartoma tumor syndrome	Cancer susceptibility
RB1	Retinoblastoma	Cancer susceptibility
SDHD, SDHAF2, SDHC, SDHB	Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4)	Cancer susceptibility
TSC1, TSC2	Tuberous sclerosis complex	Cancer susceptibility
WT1	WT1-related Wilms tumor	Cancer susceptibility
NF2	Neurofibromatosis type 2	Cancer susceptibility
COL3A1	Ehlers-Danlos syndrome, vascular type	Connective Tissue
FBN1	Marfan syndrome	Connective Tissue
TGFBR1, TGFBR2	Loey-Dietz syndrome	Connective Tissue
SMAD3	Loey-Dietz syndrome	Connective Tissue
ACTA2	Familial thoracic aortic aneurysm	Connective Tissue
MYH11	Familial thoracic aortic aneurysm	Connective Tissue
RYR2	Catecholaminergic polymorphic ventricular tachycardia	Cardiovascular
MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Cardiovascular
PKP2, DSP, DSC2, TMEM43, DSG2	Arrhythmogenic right ventricular tachycardia	Cardiovascular
KCNQ1, KCNH2	Romano-Ward long-QT syndrome types 1, 2, and 3	Cardiovascular
SCN5A	Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome	Cardiovascular
LDLR, APOB, PCSK9	Familial hypercholesterolemia	Cardiovascular
ATP7B	Wilson disease	Metabolic
OTC	Ornithine transcarbamylase deficiency	Metabolic
RYR1, CACNA1S	Malignant hyperthermia susceptibility	Other Actionable Disease

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The Carrier Section of the myGenome test is designed to examine genes associated with common recessive carrier conditions as informed by the American College of Obstetrics and Gynecology**.

The list below shows the 24 interpreted genes and their associated conditions/diseases.

Gene	Autosomal Recessive Disease
ABCC8	Familial hyperinsulinism; Neonatal diabetes mellitus
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
ASPA	Canavan disease
BCKDHA	Maple syrup urine disease type 1a
BCKDHB	Maple syrup urine disease type 1b
BLM	Bloom syndrome
CFTR	Congenital bilateral absence of vas deferens; Cystic fibrosis
CLRN1	Retinitis pigmentosa; Usher syndrome, type 3A
DHCR7	Smith-Lemli-Opitz syndrome
ELP1	Familial dysautonomia
FANCA	Fanconi anemia, complementation group A
FANCC	Fanconi anemia, complementation group C
FANCG	Fanconi anemia, complementation group G
G6PC	Glycogen storage disease Ia
GALK1	Galactokinase deficiency with cataracts
GALT	Galactosemia
GLB1	GM1-gangliosidosis, type I, type II, type III; Mucopolysaccharidosis type IVB (Morquio)
HBB	Sickle cell anemia; Beta-thalassemia
HEXA	Tay-Sachs disease
MCOLN1	Mucolipidosis IV
MYO7A	Deafness; Usher syndrome, type 1B
PAH	Phenylketonuria; Hyperphenylalaninemia
SMPD1	Niemann-Pick disease, types A & B
USH2A	Retinitis pigmentosa; Usher syndrome, type 2A

* Informed by the American College of Medical Genetics and Genomics (PMIDs: 23788249, 28383573)

** Informed by American College of Obstetrics and Gynecology (ACOG) Policies for expanded carrier screening recommendations (genes associated with a carrier frequency of 1 in 100 or greater were selected). Not intended to replace routine carrier screening.

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.