myGenome Gene-Disease Reference List for Providers

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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.

Clinically Actionable Disease Group	Gene	Mode of Inheritance	Clinically Actionable Disease by Gene	Additional Disease Associations (Sources: OMIM, GeneReviews, and/or Genetics Home Reference)
Cancer Susceptibility				
Hereditary Breast & Reproductive Cancers (4 genes) • Hereditary Breast and Ovarian Cancer syndrome • *PTEN* Hamartoma Tumor syndrome • Li-Fraumeni syndrome	*BRCA1*	AD	Hereditary Breast and Ovarian Cancer syndrome (female and male breast cancer, ovarian/fallopian tube, prostate, pancreatic)	N/A
	BRCA2	AD, AR	Hereditary Breast and Ovarian Cancer syndrome (female and male breast cancer, ovarian/fallopian tube, prostate, pancreatic, melanoma) (AD)	Fanconi anemia type D1 (AR); Glioblastoma; medulloblastoma; Wilms tumor
	PTEN	AD	PTEN Hamartoma tumor syndrome; Cowden syndrome; Banayan-Riley-Ruvalcaba syndrome	Lhermitte-Duclos syndrome; macrocephaly/autism syndrome; VATER association with macrocephaly and ventriculomegaly; Glioma susceptibility; meningioma
	TP53	AD	Li-Fraumeni syndrome	Bone marrow failure syndrome; Adrenocortical carcinoma (pediatric); Basal cell carcinoma; Choroid plexus papilloma; Colorectal cancer; glioma susceptibility; osteosarcoma
Hereditary Gastrointestinal Polyposis and Cancer (9 genes) • Lynch syndrome • Familial Adenomatous Polyposis • *MUTYH*-Associated Polyposis • Juvenile Polyposis • Peutz-Jeghers syndrome	*APC*	AD	Familial Adenomatous Polyposis (FAP); Attenuated FAP; Gardner syndrome; Turcot syndrome	N/A
	BMPR1A	AD	Juvenile Polyposis syndrome	Hereditary Mixed Polyposis syndrome
	MLH1	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD); also known as Lynch syndrome and/or Muir-Torre syndrome (AD)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR)
	MSH2	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD); also known as Lynch syndrome and/or Muir-Torre syndrome (AD)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR)
	MSH6	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD); also known as Lynch syndrome and/or Muir-Torre syndrome (AD)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR)
	MUTYH	AR, AD	MUTYH-Associated Polyposis (AR; aka also known as MYH-Associated Polyposis); Adenomas, multiple colorectal, FAP type 2; Autosomal Recessive Colorectal adenomatous polyposis with pilomatricomas	Susceptibility to colorectal cancer (AD)
	PMS2	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD); also known as Lynch syndrome and/or Muir-Torre syndrome (AD)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR)
	SMAD4	AD	Juvenile Polyposis syndrome	Hereditary Hemorrhagic Telangiectasia; Myrhe syndrome
	STK11	AD	Peutz-Jeghers syndrome	N/A
Neurocutaneous and Ocular (4 genes) • Neurofibromatosis Types 1 & 2 • Tuberous Sclerosis Complex Types 1 & 2 • Retinoblastoma	*NF2*	AD	Neurofibromatosis type 2	N/A
	RB1	AD	Retinoblastoma	N/A
	TSC1	AD, Unknown	Tuberous Sclerosis Complex type 1 (AD)	Lymphangioleiomyomatosis (Unknown)
	TSC2	AD	Tuberous Sclerosis Complex type 2	N/A
Neuro/endocrine and Renal (8 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*	AD, Unknown	Multiple Endocrine Neoplasia type 1 (AD)	Lung carcinoid (Unknown)
	RET	AD	Familial Medullary Thyroid Cancer (FMTC)	Multiple Endocrine Neoplasia Type IIA and IIB; Pheochromocytoma; Central hypoventilation syndrome, congenital; Susceptibility to and protection against Hirschsprung disease
	SDHAF2	AD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	N/A
	SDHB	AD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	Cowden syndrome; Gastrointestinal stromal tumor (GIST)
	SDHC	AD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	Cowden syndrome; Gastrointestinal stromal tumor (GIST)
	SDHD	AD, AR	Hereditary Paraganglioma-Pheochromocytoma Syndrome (AD)	Gastrointestinal stromal tumor (GIST; AD); Cowden syndrome (AD); mitochondrial complex II deficiency (AR)
	VHL	AD, AR	von Hippel-Lindau Syndrome (AD)	Familial erythrocytosis (AR)
	WT1	AD, Unknown, AR	WT1-related Wilm's tumor (AD)	WAGR syndrome (Wilms tumor, aniridia, genital anomalies, retardation; AD); Denys-Drash syndrome (AD); Frasier syndrome (AD); Congenital nephrotic syndrome (AD); Meacham syndrome (AR, Unknown)
Cardiac Disease				
Cardiomyopathy (16 genes) • Hypertrophic Cardiomyopathy • Dilated Cardiomyopathy • Arrhythmogenic Right Ventricular Cardiomyopathy • Left Ventricular Non-Compaction	*ACTC1*	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	Atrial septal defect; restrictive cardiomyopathy
	DSC2	AD, AR	Arrythmogenic right ventricular cardiomyopathy (AD, AR)	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair (AR, AD)
	DSG2	AD, AR, Unknown	Dilated cardiomyopathy (Unknown)	Arrhythmogenic right ventricular dysplasia 10 (AD) Arrthythmogenic right ventriuclar cardiomyopathy (AR)
	DSP	AD, AR, Unknown	Arrythmogenic right ventricular cardiomyopathy (AD); Dilated cardiomyopathy (Unknown)	Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis (AD); Dilated cardiomyopathy with woolly hair and keratoderma (AR); Lethal acantholytic epidermolysis bullosa (AR); Skin fragility-woolly hair syndrome (AR); Keratosis palmoplantaris striata II (Unknown)
	GLA	XL	Hypertrophic cardiomyopathy (XL)	Fabry disease (XL)
	LMNA	AD, AR	Dilated cardiomyopathy (AD)	Congenital muscular dystrophy (AD); Limb-girdle muscular dystrophy, type 1B (AD); Heart-hand syndrome, Slovenian type (AD); Lipodystrophy, familial partial, type 2 (AD); Malouf syndrome (AD); Emery-Dreifuss muscular dystrophy (AD, AR); Hutchinson-Gilford progeria (AD, AR); Charcot-Marie-Tooth disease, type 2B1 (AR); Mandibuloacral dysplasia (AR); Restrictive dermopathy, lethal (AR)
	MYBPC3	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	N/A
	MYH7	AD, AR	Dilated cardiomyopathy (AD); Hypertrophic cardiomyopathy (AD); Left ventricular noncompaction (AD)	Laing distal myopathy (AD); Scapuloperoneal syndrome, myopathic type (AD); Myosin storage myopathy (AD,AR)
	MYL2	AD	Hypertrophic cardiomyopathy	N/A
	MYL3	AD	Hypertrophic cardiomyopathy	N/A
	PKP2	AD	Arrythmogenic right ventricular cardiomyopathy	N/A
	PRKAG2	AD	Hypertrophic cardiomyopathy	Glycogen storage disease of heart, lethal congenital; Wolff-Parkinson-White syndrome
	TMEM43	AD	Arrhythmogenic right ventricular cardiomyopathy	Emery-Dreifuss muscular dystrophy
	TNNI3	AD, AR, Unknown	Dilated cardiomyopathy (AR, Unknown); Hypertrophic cardiomyopathy (AD)	Familial restrictive cardiomyopathy (AD)
	TNNT2	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	Familial restrictive cardiomyopathy (AD)
	TPM1	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	N/A

Clinically Actionable Disease Group*	Gene	Mode of Inheritance	Clinically Actionable Disease by Gene	Additional Disease Associations (Sources: OMIM, GeneReviews, and/or Genetics Home Reference)
Channelopathy and Arrhythmia (4 genes) • Romano-Ward Long QT syndromes Types 1, 2, and 3 • Brugada syndrome • Catecholaminergic Polymorphic Ventricular Tachycardia	*KCNQ1*	AD, AR	Long QT syndrome (AD); Atrial fibrillation, familial (AD); Short QT syndrome (AD); Jervell and Lange-Nielsen syndrome (AR)	N/A
	SCN5A	AD, AR, Unknown	Long QT syndrome (AD); Brugada syndrome (AD); Atrial fibrillation, familial (AD); Dilated cardiomyopathy (AD)	Heart block (progressive and nonprogressive), type IA (AD); Sick sinus syndrome (AR); Susceptibility to Sudden Infant Death Syndrome (AR): Ventricular fibrillation, familial (Unknown)
	KCNH2	AD, Unknown	Long QT syndrome (AD); Short QT syndrome (Unknown)	N/A
	RYR2	AD	Catecholaminergic polymorphic ventricular tachycardia; Arrythmogenic right ventricular cardiomyopathy	N/A
Familial Hypercholesterolemia (3 genes)	*APOB*	AD, AR	Familial hypercholesterolemia (AD)	Hypobetalipoproteinemia (AR)
	LDLR	AD	Familial hypercholesterolemia	N/A
	PCSK9	AD	Familial hypercholesterolemia	N/A
Connective Tissue				
Marfan syndrome, Loeys-Dietz syndromes, and Familial Thoracic Aortic Aneurysms and Dissections (6 genes)	*ACTA2*	AD	Familial thoracic aortic aneurysm	Moyamoya disease; Multisystemic smooth muscle dysfunction syndrome
	FBN1	AD, Unknown	Marfan syndrome (AD)	Acromicric dysplasia (AD); familial ectopia lentis (AD); geleophysic dysplasia (AD); Marfan lipodystrophy syndrome (AD); stiff skin syndrome (AD); Weill-Marchesani syndrome (AD); MASS syndrome (Unknown)
	MYH11	AD	Familial thoracic aortic aneurysm	N/A
	SMAD3	AD	Loeys-Dietz syndrome	N/A
	TGFBR1	AD	Loeys-Dietz syndrome	Susceptibility to multiple self-healing squamous epithelioma
	TGFBR2	AD	Loeys-Dietz syndrome	Colorectal cancer, hereditary nonpolyposis
Ehlers-Danlos syndrome, vascular type	*COL3A1*	AD	Ehlers-Danlos syndrome, vascular type	N/A
Other Actionable Diseases				
Malignant Hyperthermia Susceptibility (2 genes)	*CACNA1S*	AD	Malignant hyperthermia susceptibility	Hypokalemic periodic paralysis
	RYR1	AD, AR	Malignant hyperthermia susceptibility (AD)	King-Denborough syndrome (AD); Central core disease (AD, AR); Neuromuscular disease, congenital, with uniform type 1 fiber (AD, AR); Minicore myopathy with external ophthalmoplegia (AR)
Ornithine Transcarbamylase Deficiency	*OTC*	XL	Ornithine transcarbamylase deficiency	N/A
Wilson's Disease	*ATP7B*	AR	Wilson Disease	N/A

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

DISCLAIMER: Many genes are associated with additional diseases/conditions, however, myGenome is NOT designed to cover them comprehensively. Please see the myGenome Technical Limitations for additional test information.

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myGenome Standard/Premium Carrier Gene-Disease Reference List for Providers

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	Disease	Other Associated Diseases
ABCC8	Familial hyperinsulinism; Neonatal diabetes mellitus	Familial hyperinsulinism (AD); Hypoglycemia of infancy, leucine-sensitive (AD); Neonatal diabetes mellitus (AD)
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	
ASPA	Canavan disease	
BCKDHA	Maple syrup urine disease, type Ia	
BCKDHB	Maple syrup urine disease, type Ib	
BLM	Bloom syndrome	
CFTR	Congenital bilateral absence of vas deferens; Cystic fibrosis	Hereditary Pancreatitis (AD); Bronchiectasis with or without elevated sweat chloride (modifier of) (AD)
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	Delta-beta thalassemia (AD); Heinz body anemia (AD); Hereditary persistence of fetal hemoglobin (AD)
HEXA	Tay-Sachs disease	
MCOLN1	Mucolipidosis IV	
MYO7A	Deafness; Usher syndrome, type 1B	Deafness (AD)
PAH	Phenylketonuria; Hyperphenylalaninemia (non-PKU mild)	
SMPD1	Niemann-Pick disease, types A & B	
USH2A	Retinitis pigmentosa; Usher syndrome, type 2A	

** 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.