

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.

LI-Fraumeni syndrome Cancer susceptibility MEH1, MSH2, MSH6, PMS2 Lynch syndrome (Hereditary Non-Polyposis Colorectal Cancer syndrome) MEH1, MSH2, MSH6, PMS2 Lynch syndrome (Hereditary Non-Polyposis Colorectal Cancer syndrome) MUTYH MUTYH MUTYH- associated polyposis Cancer susceptibility MEMPRIA, SMAD4 Juvenile polyposis Cancer susceptibility MEMPRIA, SMAD4 Juvenile polyposis Cancer susceptibility MEM1 Von Hippel-Lindau syndrome MEM1 Multiple endocrine neoplasia type 1 Multiple endocrine neoplasia type 1 Multiple endocrine neoplasia type 2: Familial medullary thyroid cancer Cancer susceptibility PTEN PTEN hamartoma tumor syndrome Cancer susceptibility PTEN PTEN hamartoma tumor syndrome Cancer susceptibility PTEN PTEN hamartoma tumor syndrome Cancer susceptibility PTEN PTEN PTEN hamartoma tumor syndrome Cancer susceptibility PTEN PTEN PTEN hamartoma tumor syndrome Cancer susceptibility PTEN PTEN PTEN hamartoma tumor syndrome (PGL1, PGL2, PGL3, PGL4) TSC1, TSC2 Tuberous sclerosis complex Cancer susceptibility WT1 WT1-related Wilms tumor Cancer susceptibility NPS2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Ehlers-Danlos syndrome, vascular type Connective Tissue COL3A1 Ehlers-Danlos syndrome MAFan syndrome Connective Tissue COL3A1 Ehlers-Danlos syndrome COL3A1 Ehlers-Danlos syndrome COnnective Tissue CACTA2 Familial thoracic aortic aneurysm COCHEVI Descriptione Connective Tissue CACTA2 Familial thoracic aortic aneurysm Cardiovascular Cardiovascular PSPAP, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular CACTA2 Romano-Ward long-QT syndrome types 1, 2, and 3 Gardiovascular CACTA2 Romano-Ward long-QT syndrome types 1, 2, and 3 Gardiovascular CACTA2 Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular CACTA2 Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndro	Gene	Medically Actionable Disease	Disease Category
STK11 Peutz-Jeghers syndrome Cancer susceptibility MLH1, MSH2, MSH6, PMS2 Lynch syndrome (Hereditary Non-Polyposis Colorectal Cancer syndrome) Cancer susceptibility MUTYH MUTYH—Associated polyposis Cancer susceptibility BMPRIA, SMAD4 Juvenile polyposis Cancer susceptibility Cancer susceptibility MEN1 Von Hippel-Lindau syndrome Cancer susceptibility MEN1 Multiple endocrine neoplasia type 1 Cancer susceptibility PTEN Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer Cancer susceptibility PTEN PTEN PTEN hamartoma tumor syndrome Cancer susceptibility PTEN PTEN PTEN hamartoma tumor syndrome Cancer susceptibility RB1 Retinoblastoma Cancer susceptibility RB1 Retinoblastoma Cancer susceptibility RB2, SDHA, SDHAF2, SDHC, SDHB Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) WT1 WT1-related Wilms tumor Cancer susceptibility WT2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Ehlers-Danlos syndrome, vascular type Connective Tissue COL3A1 Ehlers-Danlos syndrome Connective Tissue CACTA2 Familial thoracic aortic aneurysm	BRCA1, BRCA2	Hereditary breast and ovarian cancer syndrome	Cancer susceptibility
MLH1, MSH2, MSH6, PMS2 Lynch syndrome (Hereditary Non-Polyposis Colorectal Cancer syndrome) Familial adenomatous polyposis MUTYH MUTYH-associated polyposis Cancer susceptibility MUTYH MUTYH-associated polyposis Cancer susceptibility VHL Von Hippel-Lindau syndrome Maltiple endocrine neoplasia type 1 Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer susceptibility PTEN Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer PTEN hamartoma tumor syndrome Cancer susceptibility RBI Retinoblastoma Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) TSC1, TSC2 Tuberous sclerosis complex WT1 - related Wilms tumor Cancer susceptibility NV2 Neurofibromatosis type 2 Cancer susceptibility NV2 Neurofibromatosis type 2 Cancer susceptibility RBN1 Marfan syndrome Connective Tissue FBN1 Marfan syndrome Connective Tissue FBN1 Marfan syndrome Connective Tissue CACTA2 Familial thoracic aortic aneurysm Connective Tissue NMAD3 Loey-Dietz syndrome Connective Tissue RACTA2 Familial thoracic aortic aneurysm Connective Tissue RWR2 Catecholaminergic polymorphic ventricular tachycardia Cardiovascular MYBPC3, MYH7, TNN72, TNN13, Hypertrophic cardiomyopathy; Dilated cardiomyopathy WH2, LMNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular KCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular MYBPCB, CSK9 Familial hypercholesterolemia Wilson disease Metabolic Ornithine transcarbamylase deficiency Metabolic	TP53	Li-Fraumeni syndrome	Cancer susceptibility
APC Familial adenomatous polyposis Cancer susceptibility MUTYH MUTYH-associated polyposis Cancer susceptibility MUTYH MUTYH-associated polyposis Cancer susceptibility WHL Von Hippel-Lindau syndrome Cancer susceptibility WHL Von Hippel-Lindau syndrome Cancer susceptibility RET Multiple endocrine neoplasia type 1 Cancer susceptibility RET Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer Cancer susceptibility PTEN PTEN PTEN hamartoma tumor syndrome Cancer susceptibility RBI Retinoblastoma Cancer susceptibility RBI Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) RTSC1, TSC2 Tuberous sclerosis complex Cancer susceptibility NF2 Neurofibromatosis type 2 Cancer susceptibility NF2 Neurofibromatosis type 2 Cancer susceptibility NF2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Eblers-Danlos syndrome, vascular type Connective Tissue FBN1 Marfan syndrome Connective Tissue FBN2 Loey-Dietz syndrome Connective Tissue FBN3 Loey-Dietz syndrome Connective Tissue RMAD3 Loey-Dietz syndrome RWR1 Cancer susceptibility RMATI. Familial thoracic aortic aneurysm Connective Tissue RWR2 Catcholaminergic polymorphic ventricular tachycardia Cardiovascular RWPPC3, MYH7, TNNT2, TNN13, RPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA RPCP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular RCRACA Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular RCRACA Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular RCRACA Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular RCRACA Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome	STK11	Peutz-Jeghers syndrome	Cancer susceptibility
MUTYH MUTYH-associated polyposis Cancer susceptibility BMPRIA, 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 SDHD, SDHAF2, SDHC, SDHB Retinoblastoma Cancer susceptibility RET Tuberous sclerosis complex Cancer susceptibility RET Tuberous sclerosis complex Cancer susceptibility RTSC1, TSC2 Tuberous sclerosis complex Cancer susceptibility RTSC1, TSC2 Tuberous sclerosis complex Cancer susceptibility RTSC1, TSC2 Tuberous sclerosis tomplex Cancer susceptibility RTSC1, TSC2 Tuberous sclerosis type 2 Cancer susceptibility RTSC1, TSC2 Tuberous sclerosis type 2 Cancer susceptibility RTSC1, TSC2 Tuberous sclerosis type 2 Cancer susceptibility RTSC1, TSC2 Tuberous sclerosis complex RTSC1, TSC2 Tuberous sclerosis complex COL3A1 Ehlers-Danlos syndrome, vascular type Connective Tissue RTSC1, TGEBR1, TGEBR2 Loey-Dietz syndrome Connective Tissue RTSC2 SMAD3 Loey-Dietz syndrome Connective Tissue RTSC2 CACTA2 Familial thoracic aortic aneurysm Connective Tissue RTSC3, MYH1, TNT2, TNN13, TPM1, MYL3, ACTC1, PRKAG2, SLA, MYL2, LAUNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular RCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular RTPB Wilson disease Metabolic OTC Ornithine transcarbamylase deficiency Metabolic	MLH1, MSH2, MSH6, PMS2	Lynch syndrome (Hereditary Non-Polyposis Colorectal Cancer syndrome)	Cancer susceptibility
BMPRIA, 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 RB1 Retinoblastoma Cancer susceptibility RB1 Retinoblastoma Cancer susceptibility RDHD, SDHAF2, SDHC, SDHB Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) WT1 Tuberous sclerosis complex Cancer susceptibility WT1 WT1-related Wilms tumor Cancer susceptibility WT2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Ehlers-Danlos syndrome, vascular type Connective Tissue FBN1 Marfan syndrome Connective Tissue FBN1 Marfan syndrome Connective Tissue COL3A1 Loey-Dietz syndrome Connective Tissue RGFBR1, TGFBR2 Loey-Dietz syndrome Connective Tissue RGFACA2 Familial thoracic aortic aneurysm Connective Tissue RACTA2 Familial thoracic aortic aneurysm Connective Tissue RYRP2 Catecholaminergic polymorphic ventricular tachycardia Cardiovascular RYRP2 Catecholaminergic polymorphic ventricular tachycardia Cardiovascular RYRP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular RCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular RCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular RCDLR, APOB, PCSK9 Familial hypercholesterolemia Cardiovascular RTP7B Wilson disease Metabolic	APC	Familial adenomatous polyposis	Cancer susceptibility
WHL 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 hamartoma tumor syndrome Cancer susceptibility RB1 Retinoblastoma Cancer susceptibility RB1 Retinoblastoma Cancer susceptibility RB1 Retinoblastoma Cancer susceptibility RB1 Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) TSC1, TSC2 Tuberous sclerosis complex Cancer susceptibility WT1 WT1-related Wilms tumor Cancer susceptibility NF2 Neurofibromatosis type 2 Cancer susceptibility NF2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Eblers-Danlos syndrome, vascular type Connective Tissue RBN1 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, ACTC1, PRKAC2, GLA, MYL2, LMNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular CACN3A Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome CACTONSA Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome CACTONSA Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome CACTONSA MYARDS CACTONSA MYARDS PCSK9 Familial hypercholesterolemia CACTONSCORA	MUTYH	MUTYH-associated polyposis	Cancer susceptibility
MENI Multiple endocrine neoplasia type 1 Cancer susceptibility RET Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer Cancer susceptibility PTEN hamartoma tumor syndrome Cancer susceptibility REBI Retinoblastoma Cancer susceptibility RECI, PGL2, PGL3, PGL4) RECI, PGL3, PGL4 RECI, PGL4, PGL4 RECI, PGL4 RECI, PGL4, PGL4 RECI, P	BMPR1A, SMAD4	Juvenile polyposis	Cancer susceptibility
Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer PTEN hamartoma tumor syndrome Cancer susceptibility PTEN hamartoma tumor syndrome Cancer susceptibility SDHD, SDHAF2, SDHC, SDHB Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) TSC1, TSC2 Tuberous sclerosis complex Cancer susceptibility WT1 WT1-related Wilms tumor Cancer susceptibility NE2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Ehlers-Danlos syndrome, vascular type Connective Tissue FBN1 Marfan syndrome Connective Tissue Loey-Dietz syndrome ACTA2 Familial thoracic aortic aneurysm Connective Tissue MYH11 Familial thoracic aortic aneurysm Connective Tissue ACTA2 Are Catecholaminergic polymorphic ventricular tachycardia Cardiovascular MYBPC3, MYH7, TNNT2, TNN13, FPM1, MYL3, ACTC1, PRKAG2, SLA, MYL2, LMNA Arrhythmogenic right ventricular tachycardia Cardiovascular Ca	VHL	Von Hippel-Lindau syndrome	Cancer susceptibility
PTEN hamartoma tumor syndrome Cancer susceptibility RB1 Retinoblastoma Cancer susceptibility SDHD, SDHAF2, SDHC, SDHB Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) TSC1, TSC2 Tuberous sclerosis complex Cancer susceptibility WT1 WT1-related Wilms tumor Cancer susceptibility WT2 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 MYBPC3, MYH7, TNNT2, TNN13, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular KCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3 Cardiovascular	MEN1	Multiple endocrine neoplasia type 1	Cancer susceptibility
Relinoblastoma Cancer susceptibility SDHD, SDHAF2, SDHC, SDHB Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) 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 SMAD3 Loey-Dietz syndrome Connective Tissue SMAD3 Loey-Dietz syndrome Connective Tissue MYH11 Familial thoracic aortic aneurysm Connective Tissue MYH11 Familial thoracic aortic aneurysm Connective Tissue MYH11 Familial thoracic aortic aneurysm Connective Tissue MYRPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GIA, MYL2, LMNA Hypertrophic cardiomyopathy; Dilated cardiomyopathy Cardiovascular KCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3 Cardiovascular	RET	Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer	Cancer susceptibility
Hereditary paraganglioma-pheochromocytoma syndrome (PGL1, PGL2, PGL3, PGL4) TSC1, TSC2 Tuberous sclerosis complex WT1 WT1-related Wilms tumor Cancer susceptibility WT2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Ehlers-Danlos syndrome, vascular type Connective Tissue FBN1 Marfan syndrome Connective Tissue Cardiovascular MYBPC3, MYH7, TNNT2, TNN13, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular KCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3 Cardiovascular	PTEN	PTEN hamartoma tumor syndrome	Cancer susceptibility
(PGL1, PGL2, PGL3, PGL4) TSC1, TSC2 Tuberous sclerosis complex WT1 WT1-related Wilms tumor Neurofibromatosis type 2 Cancer susceptibility NE2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Ehlers-Danlos syndrome, vascular type Connective Tissue FBN1 Marfan syndrome Connective Tissue Connective	RB1	Retinoblastoma	Cancer susceptibility
WT1 wT1-related Wilms tumor NE2 Neurofibromatosis type 2 Neurofibromatosis type 2 Neurofibromatosis type 2 Cancer susceptibility COL3A1 Ehlers-Danlos syndrome, vascular type Connective Tissue FBN1 Marfan syndrome Connective Tissue C	SDHD, SDHAF2, SDHC, SDHB		Cancer susceptibility
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, TNN13, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular	TSC1, TSC2	Tuberous sclerosis complex	Cancer susceptibility
Ehlers-Danlos syndrome, vascular type Connective Tissue FBN1 Marfan syndrome 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, TNN13, FPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular KCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3 Cardiovascular	WT1	WT1-related Wilms tumor	Cancer susceptibility
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 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	NF2	Neurofibromatosis type 2	Cancer susceptibility
TGFBR1, TGFBR2 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 PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular KCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3 Cardiovascular	COL3A1	Ehlers-Danlos syndrome, vascular type	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 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	FBN1	Marfan syndrome	Connective Tissue
Familial thoracic aortic aneurysm Connective Tissue RYR2 Catecholaminergic polymorphic ventricular tachycardia Cardiovascular Hypertrophic cardiomyopathy; Dilated cardiomyopathy Cardiovascular PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular RCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3 Cardiovascular Connective Tissue Cardiovascular	TGFBR1, TGFBR2	Loey-Dietz syndrome	Connective Tissue
Familial thoracic aortic aneurysm Cardiovascular MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular KCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3 Cardiovascular Connective Tissue Cardiovascular Connective Tissue	SMAD3	Loey-Dietz syndrome	Connective Tissue
Catecholaminergic polymorphic ventricular tachycardia Cardiovascular MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular Cardiovascular Cardiovascular Cardiovascular Cardiovascular Cardiovascular 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 Wilson disease Metabolic OTC Ornithine transcarbamylase deficiency Metabolic	ACTA2	Familial thoracic aortic aneurysm	Connective Tissue
Hypertrophic cardiomyopathy; Dilated cardiomyopathy Cardiovascular Hypertrophic cardiomyopathy; Dilated cardiomyopathy Cardiovascular PKP2, DSP, DSC2, TMEM43, DSG2 Arrhythmogenic right ventricular tachycardia Cardiovascular RCNQ1, KCNH2 Romano-Ward long-QT syndrome types 1, 2, and 3 Cardiovascular ROMANO-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular Cardiovascular Cardiovascular Cardiovascular Wilson disease Metabolic OTC Ornithine transcarbamylase deficiency Metabolic	MYH11	Familial thoracic aortic aneurysm	Connective Tissue
TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA 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 Metabolic OTC Ornithine transcarbamylase deficiency Metabolic	RYR2	Catecholaminergic polymorphic ventricular tachycardia	Cardiovascular
ROMANO-Ward long-QT syndrome types 1, 2, and 3 SCN5A Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome Cardiovascular LDLR, APOB, PCSK9 Familial hypercholesterolemia Cardiovascular Wilson disease Metabolic OTC Ornithine transcarbamylase deficiency Metabolic	MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Cardiovascular
Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome LDLR, APOB, PCSK9 Familial hypercholesterolemia Cardiovascular Wilson disease Metabolic OTC Ornithine transcarbamylase deficiency Metabolic	PKP2, DSP, DSC2, TMEM43, DSG2	Arrhythmogenic right ventricular tachycardia	Cardiovascular
LDLR, APOB, PCSK9 Familial hypercholesterolemia Cardiovascular Wilson disease Metabolic OTC Ornithine transcarbamylase deficiency Metabolic	KCNQ1, KCNH2	Romano-Ward long-QT syndrome types 1, 2, and 3	Cardiovascular
ATP7B Wilson disease Metabolic OTC Ornithine transcarbamylase deficiency Metabolic	SCN5A	Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome	Cardiovascular
OTC Ornithine transcarbamylase deficiency Metabolic	LDLR, APOB, PCSK9	Familial hypercholesterolemia	Cardiovascular
	ATP7B	Wilson disease	Metabolic
RYR1, CACNA1S Malignant hyperthermia susceptibility Other Actionable Dis-	OTC	Ornithine transcarbamylase deficiency	Metabolic
	RYR1, CACNA1S	Malignant hyperthermia susceptibility	Other Actionable Dise



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.

0	
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
ВСКДНВ	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
AYO7A	Deafness; Usher syndrome, type 1B
PAH	Phenylketonuria; Hyperphenylalaninemia
SMPD1	Niemann-Pick disease, types A & B
USH2A	Retinitis pigmentosa; Usher syndrome, type 2A

 $^{^{\}ast}$ 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. Additionally, for the carrier conditions listed, not all subtypes may be detectable. Please see the myGenome Technical Limitations for additional test information.

^{**} 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.