The Genetic Testing and Your Health Study Gene List

Below is a list of the genes and conditions that are included in the Genetic Testing and Your Health Study results report. If you are interested in learning more about a particular gene or condition, we have provided links to websites with reliable health information when available. Please note that the genetic testing performed in this study has limitations. Talk to your doctor or a genetic counselor to see if additional testing is recommended for you based on your personal and/or family history.

CONDITION*	GENE
Cancer/Tumor	
Hereditary breast and ovarian cancer	*BRCA1* *BRCA2*
Breast cancer susceptibility	*CHEK2* *PALB2* *ATM*
Li-Fraumeni syndrome	*TP53*
Peutz-Jeghers syndrome	*STK11*
Lynch syndrome	*MLH1* *MSH2* *MSH6* *PMS2*
Familial adenomatous polyposis	*APC*
MYH-associated polyposis	*MUTYH*
Juvenile polyposis syndrome	*BMPR1A* *SMAD4*
Colorectal cancer susceptibility^	*POLE* *POLD1*
Von Hippel-Lindau syndrome	*VHL*
Multiple endocrine neoplasia type1	*MEN1*
Multiple endocrine neoplasia type 2/Familial medullary thyroid cancer	*RET*
PTEN hamartoma tumor syndrome	*PTEN*
Retinoblastoma	*RB1*
Hereditary paraganglioma-pheochromocytoma syndrome	*SDHD* *SDHAF2* *SDHC* *SDHB*
Tuberous sclerosis complex	*TSC1* *TSC2*
WT1-related Wilm's tumor	*WT1*
Neurofibromatosis type 2	*NF2*
Cardiac	
Ehlers-Danlos syndrome, vascular type	*COL3A1*
Marfan syndrome	*FBN1*
Loeys-Dietz syndrome	*TGFBR1*
Familial thoracic aortic aneurysms and dissections	*TGFBR2* *SMAD3* *ACTA2* *MYLK* *MYH11*

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^No online patient resources available

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CONDITION*	GENE
Cardiac (cont.)	
Arterial tortuosity syndrome	*SLC2A10*
Hypertrophic cardiomyopathy	*MYBPC3*
Dilated cardiomyopathy	*MYH7*
	TNNT2
	TNNI3
	TPM1
	MYL3
	ACTC1
	PRKAG2
	MYL2
	LMNA
	GLA
Catecholaminergic polymorphic ventricular tachycardia	*RYR2*
Arrhythmogenic right-ventricular cardiomyopathy	*PKP2*
	DSP
	DSC2
	TMEM43
	DSG2
Romano-Ward Long QT syndrome types 1,2 and 3	*ANK2*
Timothy syndrome	*CACNA1C*
Brugada syndrome	*KCNE1*
Jervell and Lange-Nielsen syndrome	*KCNQ1*
Andersen-Tawil syndrome	*KCNH2*
	KCNJ2
	SCN5A
Familial hypercholesterolemia	*LDLR*
	APOB
	PCSK9
Neurologic	
Hemiplegic migraine	*ATP1A2*
Episodic ataxia type 2	*CACNA1A*
Hemiplegic migraine	
Spinocerebellar ataxia 6	
Congenital insensitivity to pain with anhidrosis	*NTRK1*
Seizure disorders	*SCN1A*
Hemiplegic migraine	
Congenital insensitivity to pain	*SCN9A*
Inherited erythromelalgia	
Drugs	
Clopidogrel/Voriconazole^/Citalopram^/Escitalopram^/Amitriptyline^	*CYP2C19*
Capecitabine^/Fluorouracil^/Tegafur^	*DPYD*
Peginterferon alfa-2a^/Peginterferon alfa-2b^/Ribavirin^	*IFNL3*
Simvastatin	*SLCO1B1*
Azathioprine/Mercaptopurine/Thioguanine	*TPMT1*
Warfarin	*CYP2C9*
	VKORC1

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CONDITION*	GENE
Other	
OTC deficiency	*OTC*
Malignant hyperthermia susceptibility	*RYR1* *CACNA1S*
Hemolytic uremic syndrome	*CFH*
Cystic fibrosis	*CFTR*
Ehlers-Danlos syndrome, classic type	*COL5A1*
Atopic dermatitis	*FLG*
Maturity onset diabetes of the young (MODY)	*HNF1A* *HNF1B*
Obesity^	*MC4R*
Alpha-1 antitrypsin deficiency	*SERPINA1*
Pitt-Hopkins syndrome	*TCF4*
Autosomal recessive osteopetrosis	*TCIRG1*
Amyloidosis	*TTR*
Immunodeficiency^	*TYK2*
Uromodulin kidney disease	*UMOD*
Vitamin D-dependent Rickets, Type II	*VDR*

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