MyCode®

Results reported

5,100 patient-participants have received results* from the Genomic Screening and Counseling Program

For the latest results, see **geisinger.org/MyCode-results.**

Geisinger

February 1, 2025

350,000+ participants have made the success of MyCode possible

Risk Condition	Patients per condition	Gene	Patients per gene
CDC tier 1 conditions (click link)			
Familial hypercholesterolemia (early heart attacks and strokes)	601	*APOB*	196
		LDLR	405
Hereditary breast and ovarian cancer (early breast, ovarian, prostate, pancreatic and other cancers)	983	*BRCA1*	335
		BRCA2	648
Lynch syndrome (early colon, uterine and other cancers)	520	*MLH1*	52
		MSH2	30
		MSH6	228
		PMS2	210
Cardiovascular risk			
Hereditary transthyretin amyloidosis (buildup of amyloid in the body, can lead to heart and nervous system disease)	168	*TTR*	168
Heritable thoracic aortic disease (genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)	46	*ACTA2*	46
Inherited arrhythmias (irregular heartbeat with risk for cardiac arrest)	376	*KCNH2*	44
		KCNQ1	201
		SCN5A	131
Inherited cardiomyopathies (diseases of the heart muscle with dangerous complications)	1033	*BAG3*	2
		DSC2	46
		DSG2	79
		DSP	84
		FLNC	42
		LMNA	25
		MYBPC3	197
		MYH7	79
		MYL2	8
		MYL3	8
		PKP2	81

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Cardiovascular risk *(continued)*			
(continued from page 1) **Inherited cardiomyopathies** (diseases of the heart muscle with dangerous complications)		*PRKAG2*	3
		RBM20	1
		TNNI3	24
		TNNT2	10
		TPM1	5
		TTN	339
Cancer risk			
Familial adenomatous polyposis (intestinal polyps and early colon cancer)	63	*APC*	63
Hereditary pheochromocytomas and paragangliomas (tumors that can release extra hormones and, rarely, become cancer)	114	*SDHAF2*	8
		SDHB	48
		SDHC	22
		SDHD	11
		TMEM127	25
Li-Fraumeni syndrome (early breast, soft tissue, brain, adrenal and other cancers)	27	*TP53*	27
Multiple endocrine neoplasia type 1 (tumors that can release extra hormones and, rarely, become cancer)	19	*MEN1*	19
Multiple endocrine neoplasia type 2 (early thyroid cancer)	112	*RET*	112
***MUTYH*-associated polyposis** (intestinal polyps and early colon cancer)	4	*MUTYH*	4
Neurofibromatosis, type 2 (noncancerous tumors in nervous system)	1	*NF2*	1
***PALB2*-related cancer risk** (early onset breast, pancreatic, and ovarian cancers)	134	*PALB2*	134
Peutz-Jeghers syndrome (early breast, colon, pancreatic and other cancers)	2	*STK11*	2
Retinoblastoma (early eye cancer)	7	*RB1*	7

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Cancer risk *(continued)*			
Von Hippel-Lindau syndrome (early kidney cancer and benign tumors of the brain, eye, pancreas and adrenal gland)	4	*VHL*	4
Wilms tumor (malignant kidney tumor)	2	*WT1*	2
Miscellaneous phenotypes			
Biotinidase deficiency (buildup of a B vitamin in the body, can cause issues with the nervous system)	3	*BTD*	3
Fabry disease (enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)	10	*GLA*	10
Hereditary hemochromatosis (too much iron in blood, can lead to liver and heart problems)	515	*HFE*	515
Hereditary hemorrhagic telangiectasia (abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)	46	*ACVRL1* *ENG*	13 33
Juvenile polyposis (intestinal polyps, cancer of the intestine, including colon)	3	*BMPR1A*	3
Juvenile polyposis / hereditary hemorrhagic telangiecstasia (intestinal polyps, cancer of the intestine, including colon/ abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)	4	*SMAD4*	4
Loeys-Dietz syndrome (weakening of the wall of the aorta, leading to swelling and sometimes rupture)	10	*SMAD3* *TGFBR1* *TGFBR2*	5 2 3
Malignant hyperthermia (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)	226	*RYR1*	226
Marfan syndrome (connective tissue disease that can cause heart, eye, and skeletal problems)	27	*FBN1*	27

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Miscellaneous phenotypes *(continued)*			
Maturity-onset diabetes of the young (MODY) (Diabetes in the teens or early adulthood)	13	*HNF1A*	13
Ornithine transcarbamylase deficiency (buildup of ammonia in the blood, can cause altered mental status and seizures)	4	*OTC*	4
Pompe disease (buildup of glycogen which could cause muscle probelms throughout the body)	13	*GAA*	13
***PTEN* hamartoma tumor syndrome** (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)	21	*PTEN*	21
Retinopathy (gradual vision loss, can lead to blindness)	1	*RPE65*	1
Tuberous sclerosis (multiple types of benign tumors)	25	*TSC1* *TSC2*	7 18
Vascular Ehlers-Danlos syndrome (disease of the connective tissues, including arteries and muscles, that can increase the risk for health complications, such as rupture of arteries)	14	*COL3A1*	14
Wilson disease (too much copper in the body, can cause liver disease and nervous system issues)	9	*ATP7B*	9
Totals†,‡	5,168		5,168

**Number of patient-participants with reported results and the number per gene variant/condition may not be equal due to the possibility of a participant having more than one result.*

†Includes some patients (~12%) already aware of their genomic result from clinical genetic testing. The process of clinical confirmation and disclosure may be modified for these patients

‡The gene list designated for return has shifted over time (PMID: 33576083). Totals include fewer than 10 results in genes no longer on the return list.

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