MYCODE®

Results reported

6,360 patient-participants have received results* from the Genomic Screening and Counseling Program

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

Geisinger

November 1, 2025

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

Risk Condition	Patients per condition	Gene	Patients per gene
Cardiovascular risk			
Familial hypercholesterolemia§ (early heart attacks and strokes)	812	*APOB*	284
		LDLR	528
Hereditary transthyretin amyloidosis (buildup of amyloid in the body, can lead to heart and nervous system disease)	261	*TTR*	261
Heritable thoracic aortic disease (genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)	59	*ACTA2*	59
Inherited arrhythmias (irregular heartbeat with risk for cardiac arrest)	454	*KCNH2*	47
		KCNQ1	255
		SCN5A	152
Inherited cardiomyopathies (diseases of the heart muscle with dangerous complications)	1,289	*BAG3*	2
		DSC2	46
		DSG2	79
		DSP	100
		FLNC	60
		LMNA	26
		MYBPC3	250
		MYH7	100
		MYL2	10
		MYL3	8
		PKP2	91
		PRKAG2	3
		RBM20	1
		TNNI3	35
		TNNT2	10
		TPM1	6
		TTN	462

(continued on next page)

November 1, 2025

MyCode® results reported *(continued)*

6,360 patient-participants have received results*
from the Genomic Screening and Counseling Program

Risk Condition	Patients per condition	Gene	Patients per gene
Cancer risk			
Familial adenomatous polyposis (intestinal polyps and early colon cancer)	66	*APC*	66
Hereditary breast and ovarian cancer§ (early breast, ovarian, prostate, pancreatic and other cancers)	1,143	*BRCA1*	416
		BRCA2	727
Hereditary pheochromocytomas and paragangliomas (tumors that can release extra hormones and, rarely, become cancer)	130	*SDHAF2*	13
		SDHB	55
		SDHC	22
		SDHD	11
		TMEM127	29
Li-Fraumeni syndrome (early breast, soft tissue, brain, adrenal and other cancers)	30	*TP53*	30
Lynch syndrome§ (early colon, uterine and other cancers)	656	*MLH1*	56
		MSH2	38
		MSH6	283
		PMS2	279
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)	144	*RET*	144
***MUTYH*-associated polyposis** (intestinal polyps and early colon cancer)	10	*MUTYH*	10
Neurofibromatosis, type 2 (noncancerous tumors in nervous system)	1	*NF2*	1
***PALB2*-related cancer risk** (early onset breast, pancreatic, and ovarian cancers)	174	*PALB2*	174
Peutz-Jeghers syndrome (early breast, colon, pancreatic and other cancers)	2	*STK11*	2
Retinoblastoma (early eye cancer)	7	*RB1*	7

(continued on next page)

November 1, 2025

MyCode® results reported *(continued)*

6,360 patient-participants have received results*
from the Genomic Screening and Counseling Program

Risk Condition	Patients per condition	Gene	Patients per gene
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)	3	*WT1*	3
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)	11	*GLA*	11
Hereditary hemochromatosis (too much iron in blood, can lead to liver and heart problems)	648	*HFE*	648
Hereditary hemorrhagic telangiectasia (abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)	59	*ACVRL1* *ENG*	20 39
Juvenile polyposis (intestinal polyps, cancer of the intestine, including colon)	5	*BMPR1A*	5
Juvenile polyposis / hereditary hemorrhagic telangiecstasia (intestinal polyps, cancer of the intestine, including colon/ abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)	5	*SMAD4*	5
Loeys-Dietz syndrome (weakening of the wall of the aorta, leading to swelling and sometimes rupture)	13	*SMAD3* *TGFBR1* *TGFBR2*	7 2 4
Malignant hyperthermia (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)	287	*RYR1*	287
Marfan syndrome (connective tissue disease that can cause heart, eye, and skeletal problems)	32	*FBN1*	32

(continued on next page)

November 1, 2025

MyCode® results reported *(continued)*

6,360 patient-participants have received results*
from the Genomic Screening and Counseling Program

Risk Condition	Patients per condition	Gene	Patients per gene
Miscellaneous phenotypes *(continued)*			
Maturity-onset diabetes of the young (MODY) (Diabetes in the teens or early adulthood)	15	*HNF1A*	15
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)	14	*GAA*	14
***PTEN* hamartoma tumor syndrome** (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)	24	*PTEN*	24
Retinopathy (gradual vision loss, can lead to blindness)	1	*RPE65*	1
Tuberous sclerosis (multiple types of benign tumors)	30	*TSC1* *TSC2*	8 22
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)	17	*COL3A1*	17
Wilson disease (too much copper in the body, can cause liver disease and nervous system issues)	13	*ATP7B*	13
Totals†,‡	6,453		6,453

§*CDC Tier 1 Condition*

**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.*

Geisinger