

Results reported

5,365 patient-participants have received results* from the Genomic Screening and Counseling Program

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

April 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)	661	APOB LDLR	229 432
Hereditary breast and ovarian cancer (early breast, ovarian, prostate, pancreatic and other cancers)	1,018	BRCA1 BRCA2	352 666
Lynch syndrome (early colon, uterine and other cancers)	550	MLH1 MSH2 MSH6 PMS2	52 30 237 231
Cardiovascular risk			
Hereditary transthyretin amyloidosis (buildup of amyloid in the body, can lead to heart and nervous system disease)	215	TTR	215
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)	383	KCNH2 KCNQ1 SCN5A	44 205 134
Inherited cardiomyopathies (diseases of the heart muscle with dangerous complications)	1,055	BAG3 DSC2 DSG2 DSP FLNC LMNA MYBPC3 MYH7 MYL2 MYL3 PKP2	2 46 79 84 44 25 207 79 8 8 81

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MyCode® results reported (continued)





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Cardiovascular risk <small>(continued)</small>			
<i>(continued from page 1)</i> Inherited cardiomyopathies <small>(diseases of the heart muscle with dangerous complications)</small>		PRKAG2 RBM20 TNNI3 TNNT2 TPM1 TTN	3 1 26 10 5 347
Cancer risk			
Familial adenomatous polyposis <small>(intestinal polyps and early colon cancer)</small>	63	APC	63
Hereditary pheochromocytomas and paragangliomas <small>(tumors that can release extra hormones and, rarely, become cancer)</small>	114	SDHAF2 SDHB SDHC SDHD TMEM127	8 48 22 11 25
Li-Fraumeni syndrome <small>(early breast, soft tissue, brain, adrenal and other cancers)</small>	27	TP53	27
Multiple endocrine neoplasia type 1 <small>(tumors that can release extra hormones and, rarely, become cancer)</small>	19	MEN1	19
Multiple endocrine neoplasia type 2 <small>(early thyroid cancer)</small>	114	RET	114
MUTYH-associated polyposis <small>(intestinal polyps and early colon cancer)</small>	4	MUTYH	4
Neurofibromatosis, type 2 <small>(noncancerous tumors in nervous system)</small>	1	NF2	1
PALB2-related cancer risk <small>(early onset breast, pancreatic, and ovarian cancers)</small>	143	PALB2	143
Peutz-Jeghers syndrome <small>(early breast, colon, pancreatic and other cancers)</small>	2	STK11	2
Retinoblastoma <small>(early eye cancer)</small>	7	RB1	7

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Risk Condition 	Patients per condition 	Gene 	Patients per gene 
Cancer risk <small>(continued)</small>			
Von Hippel-Lindau syndrome <small>(early kidney cancer and benign tumors of the brain, eye, pancreas and adrenal gland)</small>	4	VHL	4
Wilms tumor <small>(malignant kidney tumor)</small>	2	WT1	2
Miscellaneous phenotypes			
Biotinidase deficiency <small>(buildup of a B vitamin in the body, can cause issues with the nervous system)</small>	3	BTD	3
Fabry disease <small>(enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)</small>	10	GLA	10
Hereditary hemochromatosis <small>(too much iron in blood, can lead to liver and heart problems)</small>	559	HFE	559
Hereditary hemorrhagic telangiectasia <small>(abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)</small>	46	ACVRL1 ENG	13 33
Juvenile polyposis <small>(intestinal polyps, cancer of the intestine, including colon)</small>	3	BMPR1A	3
Juvenile polyposis / hereditary hemorrhagic telangiectasia <small>(intestinal polyps, cancer of the intestine, including colon/ abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)</small>	4	SMAD4	4
Loeys-Dietz syndrome <small>(weakening of the wall of the aorta, leading to swelling and sometimes rupture)</small>	10	SMAD3 TGFB1 TGFB2	5 2 3
Malignant hyperthermia <small>(life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)</small>	233	RYR1	233
Marfan syndrome <small>(connective tissue disease that can cause heart, eye, and skeletal problems)</small>	29	FBN1	29

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

Totals^{†,‡}

5,433



5,433

*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.