





Results reported

5047 patient-participants have received results* from the Genomic Screening and Counseling Program

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

May 1, 2024

320,000+ participants have made the success of MyCode possible

Risk Condition 	Patients per condition 	Gene 	Patients per gene 
<u>CDC tier 1 conditions (click link)</u>			
Familial hypercholesterolemia <small>(early heart attacks and strokes)</small>	589	APOB LDLR	193 396
Hereditary breast and ovarian cancer <small>(early breast, ovarian, prostate, pancreatic and other cancers)</small>	970	BRCA1 BRCA2	334 636
Lynch syndrome <small>(early colon, uterine and other cancers)</small>	518	MLH1 MSH2 MSH6 PMS2	52 31 226 210
Cardiovascular risk			
Hereditary transthyretin amyloidosis <small>(buildup of amyloid in the body, can lead to heart and nervous system disease)</small>	163	TTR	163
Heritable thoracic aortic disease <small>(genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)</small>	46	ACTA2	46
Inherited arrhythmias <small>(irregular heartbeat with risk for cardiac arrest)</small>	377	KCNE1 KCNH2 KCNQ1 SCN5A	3 44 200 130
Inherited cardiomyopathies <small>(diseases of the heart muscle with dangerous complications)</small>	1030	BAG3 DSC2 DSG2 DSP FLNC LMNA MYBPC3 MYH7 MYL2 MYL3 PKP2	2 46 79 83 42 25 196 79 8 8 81

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MyCode® results reported (continued)

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Risk Condition	Patients per condition	Gene	Patients per gene
Cardiovascular risk <small>(continued)</small>			
<i>(continued from page 1)</i>			
Inherited cardiomyopathies			
<small>(diseases of the heart muscle with dangerous complications)</small>			
		PRKAG2	3
		RBM20	1
		TNNI3	23
		TNNT2	10
		TPM1	5
		TTN	339
Cancer risk			
Familial adenomatous polyposis	63	APC	63
<small>(intestinal polyps and early colon cancer)</small>			
Hereditary pheochromocytomas and paragangliomas	113	SDHAF2	8
<small>(tumors that can release extra hormones and, rarely, become cancer)</small>		SDHB	47
		SDHC	22
		SDHD	11
		TMEM127	25
Li-Fraumeni syndrome	27	TP53	27
<small>(early breast, soft tissue, brain, adrenal and other cancers)</small>			
Multiple endocrine neoplasia type 1	19	MEN1	19
<small>(tumors that can release extra hormones and, rarely, become cancer)</small>			
Multiple endocrine neoplasia type 2	112	RET	112
<small>(early thyroid cancer)</small>			
MUTYH-associated polyposis	4	MUTYH	4
<small>(intestinal polyps and early colon cancer)</small>			
Neurofibromatosis, type 2	1	NF2	1
<small>(noncancerous tumors in nervous system)</small>			
PALB2-related cancer risk	134	PALB2	134
<small>(early onset breast, pancreatic, and ovarian cancers)</small>			
Peutz-Jeghers syndrome	2	STK11	2
<small>(early breast, colon, pancreatic and other cancers)</small>			
Retinoblastoma	7	RB1	7
<small>(early eye cancer)</small>			

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MyCode® results reported (continued)





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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>	502	HFE	502
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>	225	RYR1	225
Marfan syndrome <small>(connective tissue disease that can cause heart, eye, and skeletal problems)</small>	27	FBN1	27

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Risk Condition 	Patients per condition 	Gene 	Patients per gene 
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[†]

5114



5114

*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 already aware of their genomic result from clinical genetic testing. The process of clinical confirmation and disclosure may be modified for these patients