





MyCode[®] results reported

1489 patient-participants have received results*
from the Genomic Screening and Counseling Program



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

December 1, 2019

Risk Condition 	Patients per risk condition 	Gene 	Patients per gene 
CDC tier 1 conditions (click link)			
Hereditary breast and ovarian cancer (early breast, ovarian, prostate and other cancers)	374	BRCA1 BRCA2	121 253
Familial hypercholesterolemia (early heart attacks and strokes)	157	APOB LDLR	36 121
Lynch syndrome (early colon, uterine and other cancers)	175	PMS2 MSH6 MSH2 MLH1	72 78 14 11
Cardiovascular risk			
Cardiomyopathy (diseases of the heart muscle with dangerous complications)	106	MYH7 MYBPC3 TPM1 TNNI3 TNNT2 MYL2 MYL3 LMNA	25 55 2 4 6 3 5 6
Arrhythmia (irregular heartbeat with risk for cardiac arrest)	139	SCN5A KCNQ1 KCNE1 KCNH2	50 77 3 9
Arrhythmogenic right ventricular cardiomyopathy (disease of the heart muscle with risk for cardiac arrest)	92	DSP PKP2 DSG2 DSC2	35 24 24 9
Marfan syndrome (connective tissue disease that can cause heart, eye, and skeletal problems)	12	FBN1	12

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MyCode[®] results reported *(continued)*


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Risk Condition 	Patients per risk condition 	Gene 	Patients per gene 
Cardiovascular risk <i>(continued from front)</i>			
Heritable thoracic aortic disease (genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)	20	ACTA2 SMAD3 TGFBR1	18 1 1
Cancer risk			
Hereditary pheochromocytomas and paragangliomas (tumors that can release extra hormones and, rarely, become cancer)	32	SDHB SDHC SDHD SDHAF2	16 9 5 2
Multiple endocrine neoplasia type 1 (tumors that can release extra hormones and, rarely, become cancer)	6	MEN1	6
Multiple endocrine neoplasia type 2 (early thyroid cancer)	41	RET	41
PTEN hamartoma tumor syndrome (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)	7	PTEN	7
Tuberous sclerosis (multiple types of benign [non-cancer] tumors)	6	TSC1 TSC2	3 3
Li-Fraumeni syndrome (early breast, soft tissue, brain, adrenal and other cancers)	13	TP53	13
Familial adenomatous polyposis (early colon cancer)	14	APC	14
Von Hippel-Lindau syndrome (early kidney cancer and benign tumors of brain, eye, pancreas and adrenal gland)	2	VHL	2
Retinoblastoma (early eye cancer)	1	RB1	1

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MyCode® results reported (continued)

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Risk Condition	Patients per risk condition	Gene	Patients per gene
Other			
Malignant hyperthermia (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)	85	RYR1	85
Fabry disease (enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)	4	GLA	4
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)	6	COL3A1	6
Hereditary hemochromatosis (too much iron in blood, can lead to liver and heart problems)	203	HFE	203
Hereditary hemorrhagic telangiectasia (abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)	1	ENG	1
Juvenile polyposis syndrome/ hereditary hemorrhagic telangiectasia (intestinal polyps, cancer of the intestine, including colon/abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)	1	SMAD4	1
Totals	1497		1497

*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.