





MyCode[®] results reported

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







For the latest results, see [geisinger.org/MyCode-results](https://www.geisinger.org/MyCode-results).

October 1, 2018


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)	284	BRCA1	99
		BRCA2	185
Familial hypercholesterolemia (early heart attacks and strokes)	118	APOB	35
		LDLR	83
Lynch syndrome (early colon, uterine and other cancers)	98	PMS2	30
		MSH6	51
		MSH2	11
		MLH1	6
Cardiovascular risk			
Cardiomyopathy (diseases of the heart muscle with dangerous complications)	72	MYH7	12
		MYBPC3	41
		TPM1	2
		TNNI3	3
		TNNT2	5
		MYL3	5
		LMNA	4
Arrhythmia (irregular heartbeat with risk for cardiac arrest)	59	SCN5A	32
		KCNQ1	20
		KCNE1	3
		KCNH2	4
Arrhythmogenic right ventricular cardiomyopathy (disease of the heart muscle with risk for cardiac arrest)	81	DSP	31
		PKP2	19
		DSG2	23
		DSC2	8
Marfan syndrome (connective tissue disease that can cause heart, eye, and skeletal problems)	9	FBN1	9

(continued on next page)

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Risk Condition 	Patients per risk condition 	Gene 	Patients per gene 
Cardiovascular risk <small>(continued from front)</small>			
Heritable thoracic aortic disease (genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)	11	ACTA2	10
		SMAD3	1
Cancer risk			
Hereditary pheochromocytomas and paragangliomas (tumors that can release extra hormones and, rarely, become cancer)	17	SDHB	7
		SDHC	4
		SDHD	4
		SDHAF2	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)	21	RET	21
PTEN hamartoma tumor syndrome (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)	5	PTEN	5
Tuberous sclerosis (multiple types of benign [non-cancer] tumors)	4	TSC1	2
		TSC2	2
Li-Fraumeni syndrome (early breast, soft tissue, brain, adrenal and other cancers)	9	TP53	9
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)	1	VHL	1

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Cancer risk <small>(continued from previous page)</small>			
Retinoblastoma <small>(early eye cancer)</small>	1	RB1	1
Other			
Malignant hyperthermia <small>(life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)</small>	23	RYR1	23
Fabry disease <small>(enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)</small>	4	GLA	4
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>	6	COL3A1	6
Hereditary hemochromatosis <small>(too much iron in blood, can lead to liver and heart problems)</small>	154	HFE	154
Hereditary hemorrhagic telangiectasia <small>(abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)</small>	1	ENG	1
Juvenile polyposis syndrome/ hereditary hemorrhagic telangiectasia <small>(intestinal polyps, cancer of the intestine, including colon/abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)</small>	1	SMAD4	1
Totals	999		999

*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 condition.