MyCode® results reported

1046 patient-participants have received results*
from the Genomic Screening and Counseling Program

Geisinger mycode | 200,000+ PARTICIPANTS

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

December 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)	286	*BRCA1*	100
		BRCA2	186
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

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Risk Condition	Patients per risk condition	Gene	Patients per gene
Cardiovascular risk *(continued from front)*			
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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Risk Condition	Patients per risk condition	Gene	Patients per gene
Cancer risk *(continued from previous page)*			
Retinoblastoma (early eye cancer)	1	*RB1*	1
Other			
Malignant hyperthermia (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)	23	*RYR1*	23
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 telangiecstasia (intestinal polyps, cancer of the intestine, including colon/abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)	1	*SMAD4*	1
Totals	1050		1050

**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.*

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