MyCode®

Results reported

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

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

Geisinger

April 1, 2024

320,000+ 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)	589	*APOB*	193
		LDLR	396
Hereditary breast and ovarian cancer (early breast, ovarian, prostate, pancreatic and other cancers)	970	*BRCA1*	334
		BRCA2	636
Lynch syndrome (early colon, uterine and other cancers)	518	*MLH1*	52
		MSH2	31
		MSH6	226
		PMS2	210
Cardiovascular risk			
Hereditary transthyretin amyloidosis (buildup of amyloid in the body, can lead to heart and nervous system disease)	163	*TTR*	163
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)	377	*KCNE1*	3
		KCNH2	44
		KCNQ1	200
		SCN5A	130
Inherited cardiomyopathies (diseases of the heart muscle with dangerous complications)	1030	*BAG3*	2
		DSC2	46
		DSG2	79
		DSP	83
		FLNC	42
		LMNA	25
		MYBPC3	196
		MYH7	79
		MYL2	8
		MYL3	8
		PKP2	81

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

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Cancer risk *(continued)*			
Von Hippel-Lindau syndrome (early kidney cancer and benign tumors of the brain, eye, pancreas and adrenal gland)	4	*VHL*	4
Wilms tumor (malignant kidney tumor)	2	*WT1*	2
Miscellaneous phenotypes			
Biotinidase deficiency (buildup of a B vitamin in the body, can cause issues with the nervous system)	3	*BTD*	3
Fabry disease (enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)	10	*GLA*	10
Hereditary hemochromatosis (too much iron in blood, can lead to liver and heart problems)	502	*HFE*	502
Hereditary hemorrhagic telangiectasia (abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)	46	*ACVRL1* *ENG*	13 33
Juvenile polyposis (intestinal polyps, cancer of the intestine, including colon)	3	*BMPR1A*	3
Juvenile polyposis / hereditary hemorrhagic telangiecstasia (intestinal polyps, cancer of the intestine, including colon/ abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)	4	*SMAD4*	4
Loeys-Dietz syndrome (weakening of the wall of the aorta, leading to swelling and sometimes rupture)	10	*SMAD3* *TGFBR1* *TGFBR2*	5 2 3
Malignant hyperthermia (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)	225	*RYR1*	225
Marfan syndrome (connective tissue disease that can cause heart, eye, and skeletal problems)	27	*FBN1*	27

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Miscellaneous phenotypes *(continued)*			
Maturity-onset diabetes of the young (MODY) (Diabetes in the teens or early adulthood)	13	*HNF1A*	13
Ornithine transcarbamylase deficiency (buildup of ammonia in the blood, can cause altered mental status and seizures)	4	*OTC*	4
Pompe disease (buildup of glycogen which could cause muscle probelms throughout the body)	13	*GAA*	13
***PTEN* hamartoma tumor syndrome** (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)	21	*PTEN*	21
Retinopathy (gradual vision loss, can lead to blindness)	1	*RPE65*	1
Tuberous sclerosis (multiple types of benign tumors)	25	*TSC1* *TSC2*	7 18
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)	14	*COL3A1*	14
Wilson disease (too much copper in the body, can cause liver disease and nervous system issues)	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

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