

# Results reported

June 1, 2026

7,167 patient-participants have received results\* from the Genomic Screening and Counseling Program

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

**350,000+ participants have made the success of MyCode possible**

## Summary of results reported by category

	Total	Percentage
<b>Cardiovascular risk</b>	3,275	45%
<b>Cancer risk</b>	2,727	38%
<b>Other**</b>	1,263	17%

## Breakdown of results reported

Risk Condition 	Patients per condition 	Gene 	Patients per gene 
<b>Cardiovascular risk</b>			
<b>Familial hypercholesterolemia<sup>§</sup></b> (early heart attacks and strokes)	897	APOB	285
		LDLR	612
<b>Hereditary transthyretin amyloidosis</b> (buildup of amyloid in the body, can lead to heart and nervous system disease)	267	TTR	267
<b>Heritable thoracic aortic disease</b> (genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)	62	ACTA2	62
<b>Inherited arrhythmias</b> (irregular heartbeat with risk for cardiac arrest)	523	KCNH2	58
		KCNQ1	290
		RYR2	1
		SCN5A	174

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# MyCode® results reported (continued)

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Risk Condition 	Patients per condition 	Gene 	Patients per gene 
<b>Cardiovascular risk</b> <small>(continued)</small>			
<b>Inherited cardiomyopathies</b> (diseases of the heart muscle with dangerous complications)	<b>1,523</b>	BAG3	3
		DSC2	78
		DSG2	92
		DSP	118
		FLNC	80
		LMNA	35
		MYBPC3	277
		MYH7	124
		MYL2	11
		MYL3	8
		PKP2	118
		PRKAG2	3
		RBM20	1
		TNNI3	40
		TNNT2	10
TPM1	6		
TTN	519		
<b>Cancer risk</b>			
<b>Familial adenomatous polyposis</b> (intestinal polyps and early colon cancer)	<b>85</b>	APC	85
<b>Hereditary breast and ovarian cancer<sup>S</sup></b> (early breast, ovarian, prostate, pancreatic and other cancers)	<b>1,307</b>	BRCA1	458
		BRCA2	849
<b>Hereditary pheochromocytomas and paragangliomas</b> (tumors that can release extra hormones and, rarely, become cancer)	<b>167</b>	SDHAF2	15
		SDHB	72
		SDHC	30
		SDHD	14
		TMEM127	36
<b>Li-Fraumeni syndrome</b> (early breast, soft tissue, brain, adrenal and other cancers)	<b>31</b>	TP53	31

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Risk Condition 	Patients per condition 	Gene 	Patients per gene 
<b>Cancer risk</b> <small>(continued)</small>			
<b>Lynch syndrome<sup>§</sup></b> (early colon, uterine and other cancers)	722	MLH1 MSH2 MSH6 PMS2	63 51 314 294
<b>Multiple endocrine neoplasia type 1</b> (tumors that can release extra hormones and, rarely, become cancer)	26	MEN1	26
<b>Multiple endocrine neoplasia type 2</b> (early thyroid cancer)	149	RET	149
<b>MUTYH-associated polyposis</b> (intestinal polyps and early colon cancer)	9	MUTYH	9
<b>Neurofibromatosis, type 2</b> (noncancerous tumors in nervous system)	1	NF2	1
<b>PALB2-related cancer risk</b> (early onset breast, pancreatic, and ovarian cancers)	212	PALB2	212
<b>Peutz-Jeghers syndrome</b> (early breast, colon, pancreatic and other cancers)	2	STK11	2
<b>Retinoblastoma</b> (early eye cancer)	9	RB1	9
<b>Von Hippel-Lindau syndrome</b> (early kidney cancer and benign tumors of the brain, eye, pancreas and adrenal gland)	4	VHL	4
<b>Wilms tumor</b> (malignant kidney tumor)	3	WT1	3
<b>Miscellaneous phenotypes</b>			
<b>Biotinidase deficiency</b> (buildup of a B vitamin in the body, can cause issues with the nervous system)	3	BTD	3
<b>Fabry disease</b> (enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)	14	GLA	14

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Risk Condition 	Patients per condition 	Gene 	Patients per gene 
<b>Miscellaneous phenotypes</b> <i>(continued)</i>			
<b>Hereditary hemochromatosis</b> (too much iron in blood, can lead to liver and heart problems)	648	HFE	648
<b>Hereditary hemorrhagic telangiectasia</b> (abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)	63	ACVRL1 ENG	18 45
<b>Juvenile polyposis</b> (intestinal polyps, cancer of the intestine, including colon)	6	BMPR1A	6
<b>Juvenile polyposis / hereditary hemorrhagic telangiectasia</b> (intestinal polyps, cancer of the intestine, including colon/abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)	6	SMAD4	6
<b>Loeys-Dietz syndrome</b> (weakening of the wall of the aorta, leading to swelling and sometimes rupture)	16	SMAD3 TGFB1 TGFB2	10 2 4
<b>Malignant hyperthermia</b> (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)	321	RYR1 CACNA1S	319 2
<b>Marfan syndrome</b> (connective tissue disease that can cause heart, eye, and skeletal problems)	39	FBN1	39
<b>Maturity-onset diabetes of the young (MODY)</b> (Diabetes in the teens or early adulthood)	18	HNF1A	18
<b>Ornithine transcarbamylase deficiency</b> (buildup of ammonia in the blood, can cause altered mental status and seizures)	4	OTC	4
<b>Pompe disease</b> (buildup of glycogen which could cause muscle problems throughout the body)	20	GAA	20
<b>PTEN hamartoma tumor syndrome</b> (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)	28	PTEN	28

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<b>Miscellaneous phenotypes</b> <i>(continued)</i>			
<b>Retinopathy</b> (gradual vision loss, can lead to blindness)	1	RPE65	1
<b>Tuberous sclerosis</b> (multiple types of benign tumors)	35	TSC1	10
		TSC2	25
<b>Vascular Ehlers-Danlos syndrome</b> (disease of the connective tissues, including arteries and muscles, that can increase the risk for health complications, such as rupture of arteries)	24	COL3A1	24
<b>Wilson disease</b> (too much copper in the body, can cause liver disease and nervous system issues)	12	ATP7B	12

<b>Totals</b> <sup>†‡</sup>	7,265		7,265
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<sup>§</sup>CDC Tier 1 Condition

\*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.

\*\* Other - Includes conditions with metabolic presentation or syndromic cancer or cardiovascular risk

<sup>†</sup>Includes some patients (~12%) already aware of their genomic result from clinical genetic testing. The process of clinical confirmation and disclosure may be modified for these patients

<sup>‡</sup>The gene list designated for return has shifted over time (PMID: 33576083). Totals include fewer than 10 results in genes no longer on the return list.

