





# MyCode<sup>®</sup> results reported

1612 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).

September 1, 2020

Risk Condition 	Patients per risk condition 	Gene 	Patients per gene 
<b>CDC tier 1 conditions</b> <a href="#">(click link)</a>			
<b>Hereditary breast and ovarian cancer</b> (early breast, ovarian, prostate and other cancers)	<b>413</b>	<b>BRCA1</b> <b>BRCA2</b>	<b>141</b> <b>272</b>
<b>Familial hypercholesterolemia</b> (early heart attacks and strokes)	<b>157</b>	<b>APOB</b> <b>LDLR</b>	<b>36</b> <b>121</b>
<b>Lynch syndrome</b> (early colon, uterine and other cancers)	<b>198</b>	<b>PMS2</b> <b>MSH6</b> <b>MSH2</b> <b>MLH1</b>	<b>84</b> <b>86</b> <b>15</b> <b>13</b>
<b>Cardiovascular risk</b>			
<b>Cardiomyopathy</b> (diseases of the heart muscle with dangerous complications)	<b>126</b>	<b>MYH7</b> <b>MYBPC3</b> <b>TPM1</b> <b>TNNI3</b> <b>TNNT2</b> <b>MYL2</b> <b>MYL3</b> <b>LMNA</b>	<b>30</b> <b>64</b> <b>2</b> <b>6</b> <b>6</b> <b>4</b> <b>6</b> <b>8</b>
<b>Arrhythmia</b> (irregular heartbeat with risk for cardiac arrest)	<b>165</b>	<b>SCN5A</b> <b>KCNQ1</b> <b>KCNE1</b> <b>KCNH2</b>	<b>55</b> <b>93</b> <b>3</b> <b>14</b>
<b>Arrhythmogenic right ventricular cardiomyopathy</b> (disease of the heart muscle with risk for cardiac arrest)	<b>98</b>	<b>DSP</b> <b>PKP2</b> <b>DSG2</b> <b>DSC2</b>	<b>35</b> <b>28</b> <b>25</b> <b>10</b>
<b>Marfan syndrome</b> (connective tissue disease that can cause heart, eye, and skeletal problems)	<b>12</b>	<b>FBN1</b>	<b>12</b>

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<b>Cardiovascular risk</b> <i>(continued from front)</i>			
<b>Heritable thoracic aortic disease</b> (genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)	21	<b>ACTA2</b>	18
		<b>SMAD3</b>	1
		<b>TGFBR1</b>	1
		<b>TGFBR1</b>	1
<b>Cancer risk</b>			
<b>Hereditary pheochromocytomas and paragangliomas</b> (tumors that can release extra hormones and, rarely, become cancer)	35	<b>SDHB</b>	17
		<b>SDHC</b>	9
		<b>SDHD</b>	7
		<b>SDHAF2</b>	2
<b>Multiple endocrine neoplasia type 1</b> (tumors that can release extra hormones and, rarely, become cancer)	6	<b>MEN1</b>	6
<b>Multiple endocrine neoplasia type 2</b> (early thyroid cancer)	42	<b>RET</b>	42
<b>PTEN hamartoma tumor syndrome</b> (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)	7	<b>PTEN</b>	7
<b>Tuberous sclerosis</b> (multiple types of benign [non-cancer] tumors)	6	<b>TSC1</b>	3
		<b>TSC2</b>	3
<b>Li-Fraumeni syndrome</b> (early breast, soft tissue, brain, adrenal and other cancers)	13	<b>TP53</b>	13
<b>Familial adenomatous polyposis</b> (early colon cancer)	15	<b>APC</b>	15
<b>Von Hippel-Lindau syndrome</b> (early kidney cancer and benign tumors of brain, eye, pancreas and adrenal gland)	3	<b>VHL</b>	3
<b>Retinoblastoma</b> (early eye cancer)	1	<b>RB1</b>	1
<b>MUTYH associated polyposis</b> (early colon cancer)	2	<b>MUTYH</b>	2

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<b>Other</b>			
<b>Malignant hyperthermia</b> (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)	<b>86</b>	<b>RYR1</b>	<b>86</b>
<b>Fabry disease</b> (enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)	<b>4</b>	<b>GLA</b>	<b>4</b>
<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)	<b>6</b>	<b>COL3A1</b>	<b>6</b>
<b>Hereditary hemochromatosis</b> (too much iron in blood, can lead to liver and heart problems)	<b>203</b>	<b>HFE</b>	<b>203</b>
<b>Hereditary hemorrhagic telangiectasia</b> (abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)	<b>1</b>	<b>ENG</b>	<b>1</b>
<b>Juvenile polyposis syndrome/ hereditary hemorrhagic telangiectasia</b> (intestinal polyps, cancer of the intestine, including colon/abnormal blood vessel formation in skin, mucous membranes, lungs, liver & brain)	<b>1</b>	<b>SMAD4</b>	<b>1</b>
<b>Totals</b>	<b>1621</b>		<b>1621</b>

\*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.