





# MyCode<sup>®</sup> results reported

1068 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).

June 1, 2019

Risk condition 	Patients per risk condition 	Gene 	Patients per gene 
<b>CDC tier 1 conditions</b> <i>(click link)</i>			
<b>Hereditary breast and ovarian cancer</b> (early breast, ovarian, prostate and other cancers)	<b>290</b>	<b>BRCA1</b>	<b>102</b>
		<b>BRCA2</b>	<b>188</b>
<b>Familial hypercholesterolemia</b> (early heart attacks and strokes)	<b>125</b>	<b>APOB</b>	<b>36</b>
		<b>LDLR</b>	<b>89</b>
<b>Lynch syndrome</b> (early colon, uterine and other cancers)	<b>101</b>	<b>PMS2</b>	<b>31</b>
		<b>MSH6</b>	<b>53</b>
		<b>MSH2</b>	<b>11</b>
		<b>MLH1</b>	<b>6</b>
<b>Cardiovascular risk</b>			
<b>Cardiomyopathy</b> (diseases of the heart muscle with dangerous complications)	<b>74</b>	<b>MYH7</b>	<b>12</b>
		<b>MYBPC3</b>	<b>43</b>
		<b>TPM1</b>	<b>2</b>
		<b>TNNI3</b>	<b>3</b>
		<b>TNNT2</b>	<b>5</b>
		<b>MYL3</b>	<b>5</b>
		<b>LMNA</b>	<b>4</b>
<b>Arrhythmia</b> (irregular heartbeat with risk for cardiac arrest)	<b>65</b>	<b>SCN5A</b>	<b>34</b>
		<b>KCNQ1</b>	<b>23</b>
		<b>KCNE1</b>	<b>3</b>
		<b>KCNH2</b>	<b>5</b>
<b>Arrhythmogenic right ventricular cardiomyopathy</b> (disease of the heart muscle with risk for cardiac arrest)	<b>82</b>	<b>DSP</b>	<b>32</b>
		<b>PKP2</b>	<b>19</b>
		<b>DSG2</b>	<b>23</b>
		<b>DSC2</b>	<b>8</b>
<b>Marfan syndrome</b> (connective tissue disease that can cause heart, eye, and skeletal problems)	<b>9</b>	<b>FBN1</b>	<b>9</b>

*(continued on next page)*






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<b>Cardiovascular risk</b> <small>(continued from front)</small>			
<b>Heritable thoracic aortic disease</b> (genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)	11	<b>ACTA2</b> <b>SMAD3</b>	10 1
<b>Cancer risk</b>			
<b>Hereditary pheochromocytomas and paragangliomas</b> (tumors that can release extra hormones and, rarely, become cancer)	17	<b>SDHB</b> <b>SDHC</b> <b>SDHD</b> <b>SDHAF2</b>	7 4 4 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)	21	<b>RET</b>	21
<b>PTEN hamartoma tumor syndrome</b> (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)	5	<b>PTEN</b>	5
<b>Tuberous sclerosis</b> (multiple types of benign [non-cancer] tumors)	4	<b>TSC1</b> <b>TSC2</b>	2 2
<b>Li-Fraumeni syndrome</b> (early breast, soft tissue, brain, adrenal and other cancers)	9	<b>TP53</b>	9
<b>Familial adenomatous polyposis</b> (early colon cancer)	14	<b>APC</b>	14
<b>Von Hippel-Lindau syndrome</b> (early kidney cancer and benign tumors of brain, eye, pancreas and adrenal gland)	1	<b>VHL</b>	1

# MyCode® results reported (continued)

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<b>Cancer risk</b> <small>(continued from previous page)</small>			
<b>Retinoblastoma</b> <small>(early eye cancer)</small>	1	<b>RB1</b>	1
<b>Other</b>			
<b>Malignant hyperthermia</b> <small>(life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)</small>	23	<b>RYR1</b>	23
<b>Fabry disease</b> <small>(enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)</small>	4	<b>GLA</b>	4
<b>Vascular Ehlers-Danlos syndrome</b> <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	<b>COL3A1</b>	6
<b>Hereditary hemochromatosis</b> <small>(too much iron in blood, can lead to liver and heart problems)</small>	203	<b>HFE</b>	203
<b>Hereditary hemorrhagic telangiectasia</b> <small>(abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)</small>	1	<b>ENG</b>	1
<b>Juvenile polyposis syndrome/ hereditary hemorrhagic telangiectasia</b> <small>(intestinal polyps, cancer of the intestine, including colon/abnormal blood vessel formation in skin, mucous membranes, lungs, liver &amp; brain)</small>	1	<b>SMAD4</b>	1
<b>Totals</b>	<b>1073</b>		<b>1073</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.