### Results reported

3712 patient-participants have received results from the Genomic Screening and Counseling Program. For the latest results, see geisinger.org/MyCode-results.

October 1, 2022

300,000+ participants have made the success of MyCode possible

<table>
<thead>
<tr>
<th>Risk Condition [click link]</th>
<th>Patients per risk condition</th>
<th>Gene</th>
<th>Patients per gene</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Hereditary breast and ovarian cancer</strong> (early breast, ovarian, prostate, pancreatic and other cancers)</td>
<td>810</td>
<td><strong>BRCA1</strong></td>
<td>258</td>
</tr>
<tr>
<td></td>
<td></td>
<td><strong>BRCA2</strong></td>
<td>552</td>
</tr>
<tr>
<td><strong>Familial hypercholesterolemia</strong> (early heart attacks and strokes)</td>
<td>531</td>
<td><strong>APOB</strong></td>
<td>189</td>
</tr>
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<td></td>
<td></td>
<td><strong>LDLR</strong></td>
<td>342</td>
</tr>
<tr>
<td><strong>Lynch syndrome</strong> (early colon, uterine and other cancers)</td>
<td>442</td>
<td><strong>PMS2</strong></td>
<td>191</td>
</tr>
<tr>
<td></td>
<td></td>
<td><strong>MSH6</strong></td>
<td>198</td>
</tr>
<tr>
<td></td>
<td></td>
<td><strong>MSH2</strong></td>
<td>22</td>
</tr>
<tr>
<td></td>
<td></td>
<td><strong>MLH1</strong></td>
<td>31</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Cardiovascular risk</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Arrhythmia</strong> (irregular heartbeat with risk for cardiac arrest)</td>
</tr>
<tr>
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<td></td>
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<tr>
<td></td>
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<tr>
<td></td>
</tr>
<tr>
<td><strong>Cardiomyopathy</strong> (diseases of the heart muscle with dangerous complications)</td>
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<tr>
<td></td>
</tr>
</tbody>
</table>

| Arrhythmogenic right ventricular cardiomyopathy (disease of the heart muscle with risk for cardiac arrest) | 267 |
| | **DSP** | 72 |
| | **PKP2** | 79 |
| | **DSG2** | 75 |
| | **DSC2** | 41 |

(continued on next page)
## MyCode® results reported (continued)

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### Cardiovascular risk (continued)

<table>
<thead>
<tr>
<th>Risk Condition</th>
<th>Patients per risk condition</th>
<th>Gene</th>
<th>Patients per gene</th>
</tr>
</thead>
<tbody>
<tr>
<td>Heritable thoracic aortic disease (genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)</td>
<td>39</td>
<td>ACTA2</td>
<td>39</td>
</tr>
<tr>
<td>Hereditary Transthyretin Amyloidosis (buildup of amyloid in the body, can lead to heart and nervous system disease)</td>
<td>7</td>
<td>TTR</td>
<td>7</td>
</tr>
</tbody>
</table>

### Cancer risk

<table>
<thead>
<tr>
<th>Risk Condition</th>
<th>Patients per risk condition</th>
<th>Gene</th>
<th>Patients per gene</th>
</tr>
</thead>
<tbody>
<tr>
<td>Multiple endocrine neoplasia type 2 (early thyroid cancer)</td>
<td>97</td>
<td>RET</td>
<td>97</td>
</tr>
<tr>
<td>Hereditary pheochromocytomas and paragangliomas (tumors that can release extra hormones and, rarely, become cancer)</td>
<td>71</td>
<td>SDHB</td>
<td>38</td>
</tr>
<tr>
<td></td>
<td></td>
<td>SDHC</td>
<td>17</td>
</tr>
<tr>
<td></td>
<td></td>
<td>SDHD</td>
<td>9</td>
</tr>
<tr>
<td></td>
<td></td>
<td>SDHAF2</td>
<td>7</td>
</tr>
<tr>
<td>Familial adenomatous polyposis (early colon cancer)</td>
<td>47</td>
<td>APC</td>
<td>47</td>
</tr>
<tr>
<td>Li-Fraumeni syndrome (early breast, soft tissue, brain, adrenal and other cancers)</td>
<td>24</td>
<td>TP53</td>
<td>24</td>
</tr>
<tr>
<td>Multiple endocrine neoplasia type 1 (tumors that can release extra hormones and, rarely, become cancer)</td>
<td>14</td>
<td>MEN1</td>
<td>14</td>
</tr>
<tr>
<td>Retinoblastoma (early eye cancer)</td>
<td>7</td>
<td>RB1</td>
<td>7</td>
</tr>
<tr>
<td>MUTYH associated polyposis (early colon cancer)</td>
<td>5</td>
<td>MUTYH</td>
<td>5</td>
</tr>
<tr>
<td>Von Hippel-Lindau syndrome (early kidney cancer and benign tumors of brain, eye, pancreas and adrenal gland)</td>
<td>3</td>
<td>VHL</td>
<td>3</td>
</tr>
</tbody>
</table>

* (continued on next page)
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**Cancer risk (continued)**

<table>
<thead>
<tr>
<th>Risk Condition</th>
<th>Patients per risk condition</th>
<th>Gene</th>
<th>Patients per gene</th>
</tr>
</thead>
<tbody>
<tr>
<td>Wilms tumor and related syndromes (malignant kidney tumor)</td>
<td>2</td>
<td>WT1</td>
<td>2</td>
</tr>
<tr>
<td>Peutz-Jeghers syndrome (early breast, colon, pancreatic and other cancers)</td>
<td>2</td>
<td>STK11</td>
<td>2</td>
</tr>
<tr>
<td>Neurofibromatosis, type 2 (noncancerous tumors in nervous system)</td>
<td>1</td>
<td>NF2</td>
<td>1</td>
</tr>
<tr>
<td>PALB2 -Related Cancer Syndrome (early onset breast, pancreatic, and ovarian cancers)</td>
<td>9</td>
<td>PALB2</td>
<td>9</td>
</tr>
</tbody>
</table>

**Miscellaneous phenotypes**

<table>
<thead>
<tr>
<th>Condition</th>
<th>Patients per risk condition</th>
<th>Gene</th>
<th>Patients per gene</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hereditary hemochromatosis (too much iron in blood, can lead to liver and heart problems)</td>
<td>443</td>
<td>HFE</td>
<td>443</td>
</tr>
<tr>
<td>Malignant hyperthermia (life-threatening condition usually triggered by exposure to certain drugs used for general anesthesia)</td>
<td>192</td>
<td>RYR1</td>
<td>192</td>
</tr>
<tr>
<td>Marfan syndrome (connective tissue disease that can cause heart, eye, and skeletal problems)</td>
<td>23</td>
<td>FBN1</td>
<td>23</td>
</tr>
<tr>
<td>Tuberous sclerosis (multiple types of benign [non-cancer] tumors)</td>
<td>18</td>
<td>TSC1</td>
<td>7</td>
</tr>
<tr>
<td></td>
<td></td>
<td>TSC2</td>
<td>11</td>
</tr>
<tr>
<td>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)</td>
<td>13</td>
<td>COL3A1</td>
<td>13</td>
</tr>
<tr>
<td>PTEN hamartoma tumor syndrome (early breast, thyroid, uterine and other cancers, with intellectual disability in some cases)</td>
<td>11</td>
<td>PTEN</td>
<td>11</td>
</tr>
</tbody>
</table>

*continued on next page*
### MyCode® results reported (continued)

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<tr>
<th>Risk Condition</th>
<th>Patients per risk condition</th>
<th>Gene</th>
<th>Patients per gene</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Miscellaneous phenotypes (continued)</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Fabry disease</strong></td>
<td>8</td>
<td>GLA</td>
<td>8</td>
</tr>
<tr>
<td>(enzyme defect leading to damage of blood vessels in the skin and cells in the kidneys, heart, and nervous system)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Wilson disease</strong></td>
<td>5</td>
<td>ATP7B</td>
<td>5</td>
</tr>
<tr>
<td>(too much copper in the body, can cause liver disease and nervous system issues)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Loeys-Dietz syndrome</strong></td>
<td>6</td>
<td>SMAD3</td>
<td>3</td>
</tr>
<tr>
<td>(genetic predisposition to weakening of the wall of the aorta, leading to swelling and sometimes rupture)</td>
<td></td>
<td>TGFBR1</td>
<td>2</td>
</tr>
<tr>
<td></td>
<td></td>
<td>TGFBR2</td>
<td>1</td>
</tr>
<tr>
<td><strong>Juvenile polyposis</strong></td>
<td>2</td>
<td>BMPR1A</td>
<td>2</td>
</tr>
<tr>
<td>(intestinal polyps, cancer of the intestine, including colon)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Juvenile polyposis / hereditary hemorrhagic telangiectasia</strong></td>
<td>2</td>
<td>SMAD4</td>
<td>2</td>
</tr>
<tr>
<td>(intestinal polyps, cancer of the intestine, including colon/abnormal blood vessel formation in skin, mucous membranes, lungs, liver &amp; brain)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Hereditary hemorrhagic telangiectasia</strong></td>
<td>1</td>
<td>ENG</td>
<td>1</td>
</tr>
<tr>
<td>(abnormal blood vessel formation in skin, mucous membranes, lungs, liver and brain)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Ornithine Transcarbamylase Deficiency</strong></td>
<td>1</td>
<td>OTC</td>
<td>1</td>
</tr>
<tr>
<td>(build up of ammonia in the blood, can cause altered mental status and seizures)</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Totals**

3746

*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.*