OncoGeneDx: Comprehensive Common Cancer Panel

Panel Gene List: APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM*, FANCC, FANCM, FH, FLCN, HOXB13, MET, MITF, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, POT1, PTEN, RAD51C, RAD51D, RECQL, SCG5/GREM1*, SDHB, SDHC, SDHD, SMAD4, STK11, TP53, TSC1, TSC2, VHL

*Testing includes sequencing and deletion/duplication analysis for all genes except EPCAM and SCG5/GREM1 (del/dup only).

Clinical Features:
Cancer is a common disease affecting approximately 1 in 3 individuals in the U.S. While the majority of cancers are sporadic in nature, some families have hereditary forms of cancer that are associated with increased cancer risks compared with the general population. Approximately 5-10% of cancer cases are thought to be due to a hereditary predisposition. The features of a personal and/or family history of cancer that are suggestive of a hereditary cancer predisposition include: young ages at diagnosis, multiple primary cancers in a single individual, and several relatives affected with the same type of cancer or related cancers spanning multiple generations.

For some of the well-described hereditary conditions discussed below, clinical diagnostic criteria based on personal medical history and family history are available to help identify patients most likely to have a hereditary cancer syndrome. In many cases, however, patients do not meet the clinical diagnostic criteria or the criteria may overlap for multiple conditions, making it difficult to decide which genes should be tested and in what order. The OncoGeneDx Comprehensive Common Cancer Panel offered at GeneDx includes analysis of 47 genes associated with hereditary predisposition to various cancers including the most common hereditary cancer syndromes such as Hereditary Breast and Ovarian Cancer syndrome (BRCA1, BRCA2) and Lynch syndrome (MLH1, MSH2, MSH6, PMS2, EPCAM), as well as newly described genes.

Many of the genes on this panel are involved in the mismatch repair pathway, the Fanconi anemia pathway and/or play a role in DNA damage repair. Newer genes that have been identified in families with cancer have been included in the panel to make it as comprehensive as possible. These genes include, but are not limited to, AXIN2, NTHL1, POT1, and RECQL. The evidence available to date may be derived from a small number of patients with wide confidence intervals or is based upon an ethnic cohort with one specific variant. Accurate risk assessment may be complicated by the low penetrance of pathogenic variants in these genes and/or ascertainment bias.
Inheritance Pattern:
Most genes on this panel are associated with an autosomal dominant cancer risk with the exception of MUTYH and NTHL1, which are associated with an autosomal recessive cancer risk. Some of the genes on this panel are also associated with extremely rare conditions when inherited in an autosomal recessive fashion. The specifics of this inheritance are outlined in the table below.

Test Methods:
Genomic DNA is extracted from the submitted specimen. For skin punch biopsies, fibroblasts are cultured and used for DNA extraction. The DNA is enriched for the complete coding regions and splice junctions of the genes on this panel using a proprietary targeted capture system developed by GeneDx for next-generation sequencing with CNV calling (NGS-CNV). For PTEN nucleotides c.-700 through c.-1300 in the promoter region, and for APC, promoters 1A and 1B are also captured. The enriched targets are simultaneously sequenced with paired-end reads on an Illumina platform. Bi-directional sequence reads are assembled and aligned to reference sequences based on NCBI RefSeq transcripts and human genome build GRCh37/UCSC hg19. After gene specific filtering, data are analyzed to identify sequence variants and most deletions and duplications involving coding exons; however, technical limitations and inherent sequence properties effectively reduce this resolution for some genes. Concurrent MSH2 Exons 1-7 Inversion analysis from NGS data is also performed. For EPCAM and SCG5, deletion/duplication analysis, but not sequencing, is performed. Alternative sequencing or copy number detection methods are used to analyze regions with inadequate sequence or copy number data by NGS. Reported clinically significant variants are confirmed by an appropriate method. Sequence variants are reported according to the Human Genome Variation Society (HGVS) guidelines. Copy number variants are reported based on the probe coordinates, the coordinates of the exons involved, or precise breakpoints when known. Reportable variants include pathogenic variants, likely pathogenic variants and variants of uncertain significance. Likely benign and benign variants, if present, are not routinely reported but are available upon request.

Test Sensitivity:
The clinical sensitivity of sequencing and deletion/duplication analysis of the 47 genes included in the OncoGeneDx Comprehensive Common Cancer Panel depends in part on the patient’s clinical phenotype and family history. In general, the sensitivity is highest for individuals with features suggestive of a hereditary predisposition to cancer as outlined above. DNA sequencing will detect nucleotide substitutions and small insertions and deletions, while NGS-CNV analysis, array CGH, or MLPA will detect exon-level deletions and duplications. These methods are expected to be greater than 99% sensitive in detecting pathogenic variants identifiable by sequencing or CNV technology.
Genetic testing using the methods applied at GeneDx is expected to be highly accurate. Normal findings do not rule out the diagnosis of a genetic disorder since some genetic abnormalities may be undetectable by this test. The methods used cannot reliably detect deletions of 20bp to 250bp in size, or insertions of 10bp to 250 bp in size. Sequencing cannot detect low-level mosaicism. The copy number assessment methods used with this test cannot reliably detect mosaicism and cannot identify balanced chromosome aberrations. Rarely, incidental findings of large chromosomal rearrangements outside the gene of interest may be identified. Regions of certain genes have inherent sequence properties (for example: repeat, homology, or pseudogene regions, high GC content, rare polymorphisms) that yield suboptimal data, potentially impairing accuracy of the results. False negatives may also occur in the setting of bone marrow transplantation, recent blood transfusion, or suboptimal DNA quality. In individuals with active or chronic hematologic neoplasms or conditions, there is a possibility that testing may detect an acquired somatic variant, resulting in a false positive result. As the ability to detect genetic variants and naming conventions can differ among laboratories, rare false negative results may occur when no positive control is provided for testing of a specific variant identified at another laboratory. The chance of a false positive or false negative result due to laboratory errors incurred during any phase of testing cannot be completely excluded. Interpretations are made with the assumption that any clinical information provided, including family relationships, are accurate. Consultation with a genetics professional is recommended for interpretation of results.

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<th>Gene</th>
<th>Protein</th>
<th>Inheritance</th>
<th>Disease Associations</th>
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<tr>
<td>APC&lt;sup&gt;2−5&lt;/sup&gt;</td>
<td>ADENOMATOUS POLYPOSIS COLI PROTEIN</td>
<td>AD</td>
<td>Familial adenomatous polyposis (FAP)-associated condition: colorectal, duodenal or periampullary, gastric, thyroid, pancreatic, brain (medulloblastoma) &amp; liver (hepatoblastoma) cancers, desmoid tumors, gastrointestinal polyps</td>
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<tr>
<td>ATM&lt;sup&gt;6−11&lt;/sup&gt;</td>
<td>SERINE-PROTEIN KINASE ATM</td>
<td>AD</td>
<td>Breast, colon &amp; pancreatic cancers</td>
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<td></td>
<td></td>
<td>AR</td>
<td>Ataxia telangiectasia</td>
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<td>AXIN2&lt;sup&gt;12,13&lt;/sup&gt;</td>
<td>AXIN-2</td>
<td>AD</td>
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<td>BAP1&lt;sup&gt;14,15&lt;/sup&gt;</td>
<td>UBIQUITIN CARBOXYL-TERMINAL HYDROLASE BAP1</td>
<td>AD</td>
<td>Uveal/cutaneous melanoma, mesothelioma, renal cancer</td>
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<tr>
<td>Gene</td>
<td>Distribution</td>
<td>Function</td>
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<tr>
<td>BARD1</td>
<td>16–19</td>
<td>BRCA1-ASSOCIATED RING DOMAIN PROTEIN 1</td>
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<tr>
<td>BMPR1A</td>
<td>4,20–22</td>
<td>BONE MORPHOGENETIC PROTEIN RECEPTOR TYPE-1A</td>
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<tr>
<td>BRCA1</td>
<td>23–33</td>
<td>BREAST CANCER TYPE 1 SUSCEPTIBILITY PROTEIN</td>
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<tr>
<td>BRCA2</td>
<td>23–31,33</td>
<td>BREAST CANCER TYPE 2 SUSCEPTIBILITY PROTEIN</td>
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<tr>
<td>BRIP1</td>
<td>7,34–36</td>
<td>FANCONI ANEMIA GROUP J PROTEIN</td>
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<tr>
<td>CDH1</td>
<td>37–43</td>
<td>CADHERIN 1</td>
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<tr>
<td>CDK4</td>
<td>44–46</td>
<td>CYCLIN-DEPENDENT KINASE 4</td>
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<tr>
<td>CDKN2A</td>
<td>46–51</td>
<td>CYCLIN-DEPENDENT KINASE INHIBITOR 2A, TUMOR SUPPRESSOR ARF</td>
<td></td>
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<tr>
<td>CHEK2</td>
<td>7,8,32,52–58</td>
<td>SERINE/THREONINE-PROTEIN KINASE CHK2</td>
<td></td>
</tr>
<tr>
<td>EPCAM</td>
<td>69–64</td>
<td>EPITHELIAL CELL ADHESION MOLECULE</td>
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</tr>
</tbody>
</table>

**BARD1**: 16–19
- BRCA1-ASSOCIATED RING DOMAIN PROTEIN 1
- **Function**: Breast & ovarian cancer
- **Related Conditions**: Juvenile Polyposis syndrome (JPS): colorectal, gastric (if gastric polyps), small bowel & pancreatic cancer, gastrointestinal polyps

**BMPR1A**: 4,20–22
- BONE MORPHOGENETIC PROTEIN RECEPTOR TYPE-1A
- **Function**: Juvenile Polyposis syndrome (JPS): colorectal, gastric (if gastric polyps), small bowel & pancreatic cancer, gastrointestinal polyps

**BRCA1**: 23–33
- BREAST CANCER TYPE 1 SUSCEPTIBILITY PROTEIN
- **Function**: Hereditary breast and ovarian cancer (HBOC) syndrome: breast, ovarian, pancreatic, prostate & endometrial serous cancer

**BRCA2**: 23–31,33
- BREAST CANCER TYPE 2 SUSCEPTIBILITY PROTEIN
- **Function**: Hereditary breast and ovarian cancer (HBOC) syndrome: breast, ovarian, pancreatic, prostate, melanoma & endometrial serous cancer

**BRIP1**: 7,34–36
- FANCONI ANEMIA GROUP J PROTEIN
- **Function**: Fanconi anemia

**CDH1**: 37–43
- CADHERIN 1
- **Function**: Hereditary diffuse gastric cancer (HDGC) syndrome: gastric (diffuse), breast & colon (signet ring) cancer

**CDK4**: 44–46
- CYCLIN-DEPENDENT KINASE 4
- **Function**: Melanoma, non-melanoma skin & pancreatic cancer

**CDKN2A**: 46–51
- CYCLIN-DEPENDENT KINASE INHIBITOR 2A, TUMOR SUPPRESSOR ARF
- **Function**: Familiar atypical multiple mole melanoma (FAMMM) syndrome: melanoma, pancreatic cancer & astrocytoma

**CHEK2**: 7,8,32,52–58
- SERINE/THREONINE-PROTEIN KINASE CHK2
- **Function**: Breast, colon, prostate, gastric & thyroid cancer

**EPCAM**: 69–64
- EPITHELIAL CELL ADHESION MOLECULE
- **Function**: Lynch syndrome (LS): colorectal, endometrial, ovarian, gastric, pancreatic, biliary tract, urinary tract, small bowel,
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<th>Gene</th>
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<td>FANCC&lt;sup&gt;65,66&lt;/sup&gt;</td>
<td>FANCONI ANEMIA GROUP C PROTEIN</td>
<td>AD</td>
<td>Breast cancer</td>
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<td></td>
<td></td>
<td>AR</td>
<td>Constitutional mismatch repair deficiency syndrome</td>
</tr>
<tr>
<td>FANCM&lt;sup&gt;67–69&lt;/sup&gt;</td>
<td>FANCONI ANEMIA GROUP M PROTEIN</td>
<td>AD</td>
<td>Breast &amp; ovarian cancer</td>
</tr>
<tr>
<td></td>
<td></td>
<td>AR</td>
<td>Fanconi anemia-like cancer susceptibility</td>
</tr>
<tr>
<td>FH&lt;sup&gt;70–74&lt;/sup&gt;</td>
<td>FUMARATE HYDRATASE, MITOCHONDRIAL</td>
<td>AD</td>
<td>Hereditary leiomyomatosis and renal cell cancer (HLRCC): renal cancer (type II papillary), leiomyomas, pheochromocytoma, paraganglioma</td>
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<tr>
<td></td>
<td></td>
<td>AR</td>
<td>Fumarate hydratase deficiency</td>
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<tr>
<td>FLCN&lt;sup&gt;75–79&lt;/sup&gt;</td>
<td>FOLLICULIN</td>
<td>AD</td>
<td>Birt-Hogg-Dubé syndrome (BHD): renal cancer</td>
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<td>HOXB13&lt;sup&gt;80–82&lt;/sup&gt;</td>
<td>HOMEBOX PROTEIN HOX-B13</td>
<td>AD</td>
<td>Prostate cancer</td>
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<td>MET&lt;sup&gt;83–86&lt;/sup&gt;</td>
<td>HEPATOCYTE GROWTH FACTOR RECEPTOR</td>
<td>AD</td>
<td>Hereditary papillary renal carcinoma (HPRC): renal cancer (type I papillary)</td>
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<td>MITF&lt;sup&gt;87–89&lt;/sup&gt;</td>
<td>MICROPHTHALMIA-ASSOCIATED TRANSCRIPTION FACTOR</td>
<td>AD</td>
<td>Renal cancer, melanoma</td>
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<td>MLH1&lt;sup&gt;59–63,90,91&lt;/sup&gt;</td>
<td>DNA MISMATCH REPAIR PROTEIN MLH1</td>
<td>AD</td>
<td>Lynch syndrome (LS): colorectal, endometrial, ovarian, gastric, pancreatic, biliary tract, urinary tract, small bowel, prostate &amp; brain cancer, sebaceous neoplasms</td>
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<tr>
<td></td>
<td></td>
<td>AR</td>
<td>Constitutional mismatch repair deficiency syndrome</td>
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<tr>
<td>MSH2&lt;sup&gt;59–64,90,91&lt;/sup&gt;</td>
<td>DNA MISMATCH REPAIR PROTEIN MSH2</td>
<td>AD</td>
<td>Lynch syndrome (LS): colorectal, endometrial, ovarian, gastric, pancreatic, biliary tract, urinary tract, small bowel, prostate &amp; brain cancer, sebaceous neoplasms</td>
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<tr>
<td>Gene</td>
<td>Description</td>
<td>Inheritance</td>
<td>Conditions</td>
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<tr>
<td><strong>MSH6</strong></td>
<td>DNA MISMATCH REPAIR PROTEIN MSH6</td>
<td>AR</td>
<td>Constitutional mismatch repair deficiency syndrome</td>
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<tr>
<td><strong>MUTYH</strong></td>
<td>ADENINE DNA GLYCOSYLASE</td>
<td>AR</td>
<td><em>MUTYH</em>-associated polyposis (MAP): colorectal, small bowel &amp; endometrial serous cancer, gastrointestinal polyps</td>
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<td><strong>NBN</strong></td>
<td>NIBRIN</td>
<td>AD</td>
<td>Breast &amp; prostate cancer, non-Hodgkin lymphoma</td>
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<td><strong>NF1</strong></td>
<td>NEUROFIBROMIN</td>
<td>AD</td>
<td>Neurofibromatosis type 1 (NF1) syndrome: breast cancer, GIST, optic nerve gliomas, pheochromocytoma, MPNST, neurofibromas, brain tumors</td>
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<tr>
<td><strong>NTHL</strong></td>
<td>ENDONUCLEASE III-LIKE 1</td>
<td>AR</td>
<td>Colon cancer, colon polyps</td>
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<tr>
<td><strong>PALB2</strong></td>
<td>PARTNER AND LOCALIZER OF BRCA2</td>
<td>AD</td>
<td>Breast, pancreatic &amp; ovarian cancer</td>
</tr>
<tr>
<td><strong>PMS2</strong></td>
<td>MISMATCH REPAIR ENDONUCLEASE PMS2</td>
<td>AD</td>
<td>Lynch syndrome (LS): colorectal, endometrial, ovarian, gastric, pancreatic, biliary tract, urinary tract, small bowel, prostate &amp; brain cancer, sebaceous neoplasms</td>
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<tr>
<td>Gene</td>
<td>Description</td>
<td>Inheritance</td>
<td>Associated Conditions</td>
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<td><strong>POLD1</strong>&lt;sup&gt;126,127&lt;/sup&gt;</td>
<td>DNA POLYMERASE DELTA CATALYTIC SUBUNIT</td>
<td>AR</td>
<td>Constitutional mismatch repair deficiency syndrome</td>
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<td><strong>POLE</strong>&lt;sup&gt;126,128–130&lt;/sup&gt;</td>
<td>DNA POLYMERASE EPSILON CATALYTIC SUBUNIT A</td>
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<td>Colon &amp; endometrial cancer, colon polyps</td>
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<td><strong>POT1</strong>&lt;sup&gt;131–136&lt;/sup&gt;</td>
<td>PROTECTION OF TELOMETERS 1</td>
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<td><strong>PTEN</strong>&lt;sup&gt;4,137–140&lt;/sup&gt;</td>
<td>PHOSPHATIDYLINOSITOL 3,4,5-TRISPHOSPHATE 3-PHOSPHATASE AND DUAL-SPECIFICITY PROTEIN PHOSPHATASE PTEN</td>
<td>AD</td>
<td>PTEN hamartoma tumor syndrome (PHTS): breast, thyroid, endometrial, colon, melanoma, renal cancer, gastrointestinal polyps, Lhermitte-Duclos Disease</td>
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<tr>
<td><strong>RAD51C</strong>&lt;sup&gt;141–144&lt;/sup&gt;</td>
<td>DNA REPAIR PROTEIN RAD51 HOMOLOG 3</td>
<td>AD</td>
<td>Breast &amp; ovarian cancer</td>
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<td><strong>RAD51D</strong>&lt;sup&gt;141,142,145,146&lt;/sup&gt;</td>
<td>DNA REPAIR PROTEIN RAD51 HOMOLOG 4</td>
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<td>Breast &amp; ovarian cancer</td>
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<tr>
<td><strong>RECQL</strong>&lt;sup&gt;147–150&lt;/sup&gt;</td>
<td>RECQ PROTEIN-LIKE</td>
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<td>Breast cancer</td>
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<td><strong>SCG5/GREM1</strong>&lt;sup&gt;151–153&lt;/sup&gt;</td>
<td>NEUROENDOCRINE PROTEIN 7B2/GREMLIN-1</td>
<td>AD</td>
<td>Hereditary mixed polyposis syndrome (HMPS): colon cancer, colon polyps</td>
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<tr>
<td><strong>SDHB</strong>&lt;sup&gt;154–157&lt;/sup&gt;</td>
<td>SUCCINATE DEHYDROGENASE [UBIQUINONE] IRON-SULFUR SUBUNIT, MITOCHONDRIAL</td>
<td>AD</td>
<td>Hereditary paraganglioma/pheochromocytoma (PGL/PCC) syndrome: paraganglioma, pheochromocytoma, renal cancer, GIST</td>
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<tr>
<td><strong>SDHC</strong>&lt;sup&gt;154,155,158–160&lt;/sup&gt;</td>
<td>SUCCINATE DEHYDROGENASE CYTOCHROME B560 SUBUNIT, MITOCHONDRIAL</td>
<td>AD</td>
<td>Hereditary paraganglioma/pheochromocytoma (PGL/PCC) syndrome: paraganglioma,</td>
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<tr>
<td>Genes/Proteins</td>
<td>Description</td>
<td>Mode of Inheritance</td>
<td>Conditions</td>
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<tr>
<td><strong>SDHD</strong>&lt;sup&gt;154–156,161,162&lt;/sup&gt;</td>
<td>Succinate Dehydrogenase [Ubiquinone] Cytochrome B Small Subunit, Mitochondrial</td>
<td>AD</td>
<td>Hereditary paraganglioma/ pheochromocytoma (PGL/PCC) syndrome: paraganglioma, pheochromocytoma, renal cancer, GIST, thyroid cancer</td>
</tr>
<tr>
<td><strong>SMAD4</strong>&lt;sup&gt;4,21,22,163,164&lt;/sup&gt;</td>
<td>Mothers Against Decapentaplegic Homolog 4</td>
<td>AD</td>
<td>Juvenile polyposis syndrome (JPS): colorectal, gastric (if gastric polyps), small bowel &amp; pancreatic cancer, gastrointestinal polyps</td>
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<tr>
<td><strong>STK11</strong>&lt;sup&gt;4,165–167&lt;/sup&gt;</td>
<td>Serine/Threonine-Protein Kinase STK11</td>
<td>AD</td>
<td>Peutz-Jeghers syndrome (PJS): breast, colorectal, pancreatic, gastric, small bowel, ovarian, lung, cervical &amp; endometrial cancer, testicular tumors (LCCSCT), gastrointestinal polyps</td>
</tr>
<tr>
<td><strong>TP53</strong>&lt;sup&gt;32,168–172&lt;/sup&gt;</td>
<td>Cellular Tumor Antigen P53</td>
<td>AD</td>
<td>Li-Fraumeni syndrome (LFS): breast cancer, sarcoma, brain cancer, hematologic malignancies, adrenocortical carcinoma, among others**</td>
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<td><strong>TSC1</strong>&lt;sup&gt;173–175&lt;/sup&gt;</td>
<td>Hamartin</td>
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<td>Tuberous sclerosis complex (TSC): renal cancer/tumors, CNS tumors (subependymal nodules and subependymal giant cell astrocytomas), hamartomatous tumors (cardiac rhabdomyomas)</td>
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<tr>
<td>Gene</td>
<td>Tumor Type</td>
<td>AD</td>
<td>Description</td>
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<tr>
<td>TSC2\textsuperscript{173–175}</td>
<td>Tuberous sclerosis complex (TSC): renal cancer/tumors, CNS tumors (subependymal nodules and subependymal giant cell astrocytomas), hamartomatous tumors (cardiac rhabdomyomas and angiomyolipomas)</td>
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<td>VHL\textsuperscript{176–179}</td>
<td>Tumor Suppressor</td>
<td>AD</td>
<td>von Hippel-Lindau (VHL) disease: renal cancer (clear cell), pancreatic neuroendocrine tumors, hemangioblastoma, pheochromocytoma, endolymphatic sac tumors</td>
</tr>
</tbody>
</table>

Because of evolving and expanding phenotypes, this list of cancer/tumor types is not exhaustive. Gene-specific risk for some of the cancers and other features listed are not well-defined.

** High overall risk of cancer: 75% lifetime risk for males to develop cancer, nearly 100% risk for females.

Abbreviations:
- AD – Autosomal Dominant
- AR – Autosomal Recessive
- CGH – Comparative genomic hybridization
- GIST – Gastrointestinal stromal tumor
- LCCSCT - Large cell-calcifying Sertoli cell tumors
- MLPA – Multiplex ligation-dependent probe amplification
- MPNST - Malignant peripheral nerve sheath tumors

References:


