

# Hereditary Cancer Panels

## Lifetime Risks of Cancers and/or Tumors

### High-Risk Genes

Well-studied • Greater than 4-fold risk of developing one or more cancers • Can cause a moderate risk for other cancers • National or expert opinion guidelines for screening and prevention are established

### Moderate-Risk Genes

Well-studied • Approximately 2- to 4-fold risk of developing one or more cancers • May increase risk for other cancers • Limited guidelines for screening and prevention

### Newer Genes

Not as well-studied • Precise lifetime risks and tumor spectrum not yet determined • Guidelines for screening and prevention are limited or not available

For a complete list of available testing options, please visit: [www.genedx.com/oncology](http://www.genedx.com/oncology)

## Tumor Spectrum and Lifetime Risks Associated with Pathogenic Variants\*

	Gene	Lifetime Cancer and/or Tumor Risks*
High-Risk Genes	<i>ALK</i>	Neuroblastic tumors (up to 57%)
	<i>APC</i>	Colorectal (up to 93%), Duodenal or periampullary (4-12%), Gastric, Thyroid (up to 3%), Pancreatic, Brain-medulloblastoma, Liver-hepatoblastoma, Desmoid tumors, Gastrointestinal polyps
	<i>BMPR1A</i>	Colorectal (up to 68%), Gastric (up to 21% if gastric polyps), Small bowel, Pancreatic, Gastrointestinal polyps
	<i>BRCA1</i>	Female breast (55-87%), Ovarian (39-59%), Prostate, Male breast, Pancreatic, Fallopian tube, Primary peritoneal, Endometrial-serous
	<i>BRCA2</i>	Female breast (33-84%), Prostate (up to 34%), Ovarian (11-27%), Pancreatic (up to 7%), Male breast (up to 7%), Melanoma, Fallopian tube, Primary peritoneal, Endometrial-serous
	<i>CDC73</i>	Hyperparathyroidism, Parathyroid cancer and tumors, Jaw tumors-ossifying fibromas, Renal tumors, Uterine tumors
	<i>CDH1</i>	Gastric-diffuse, Female breast-lobular (39-55%), Colorectal
	<i>CDKN2A</i>	Melanoma (28-67%), Pancreatic (17%), Brain-astrocytoma
	<i>EPCAM**</i>	Colorectal (69-75%), Endometrial (12-55%), Ovarian, Gastric, Pancreatic, Biliary tract, Urinary tract-transitional cell, Small bowel, Brain, Sebaceous neoplasms, Prostate
	<i>FH</i>	Renal-type II papillary (10-19%), Paranglioma/Pheochromocytoma, Leiomyomas-cutaneous and uterine
	<i>FLCN</i>	Renal cancer and tumors (6-41%)
	<i>MEN1</i>	Hyperparathyroidism, Parathyroid tumors (95%), Neuroendocrine tumors of the gastro-entero-pancreatic (GEP) tract (up to 80%), Anterior pituitary tumors (20-65%), Carcinoid tumors, Adrenal tumors (pheochromocytomas and adrenocortical tumors), and other tumors
	<i>MLH1</i>	Colorectal (34-46%), Endometrial (18-54%), Ovarian (10-20%), Gastric (6-20%), Urinary tract-transitional cell (1-4%), Pancreatic (1-4%), Biliary tract (2-3%), Small bowel (4-12%), Brain, Sebaceous neoplasms, Prostate
	<i>MSH2</i>	Colorectal (37-48%), Endometrial (21-57%), Ovarian (10-24%), Urinary tract-transitional cell (8-20%), Gastric (<1-9%), Pancreatic (1-4%), Biliary tract, Small bowel (1%), Brain, Sebaceous neoplasms, Prostate
	<i>MSH6**</i>	Colorectal (20-44%), Endometrial (16-71%), Ovarian (1-13%), Gastric, Pancreatic, Biliary tract, Urinary tract-transitional cell, Small bowel, Brain, Sebaceous neoplasms, Prostate
	<i>MUTYH</i>	Colorectal (up to 80%), Duodenal (up to 4%), Gastrointestinal polyps
	<i>NF1</i>	Neurofibromas, Optic nerve gliomas (15%), Pheochromocytomas (1-13%), Malignant peripheral nerve sheath tumors (6-16%), Brain tumors (2-3%), Female breast (up to 26%), Gastrointestinal stromal tumor (GIST)
	<i>NF2</i>	Schwannomas-vestibular (greater than 90%) and other cranial nerves (24-51%), Central nervous system tumors-spinal tumors (60-90%) and meningioma (50-80%)
	<i>PALB2</i>	Female breast (up to 58%), Male breast, Pancreatic, Ovarian, Prostate
	<i>PHOX2B</i>	Neuroblastic tumors (up to 50%) including: neuroblastoma; ganglioneuroblastoma; ganglioneuroma
<i>PMS2**</i>	Colorectal (11-20%), Endometrial (12-26%), Ovarian, Gastric, Pancreatic, Biliary tract, Urinary tract-transitional cell, Small bowel, Brain, Sebaceous neoplasms, Prostate	

	Gene	Lifetime Cancer and/or Tumor Risks*
High-Risk Genes	<i>PRKAR1A</i>	Myxomas-cardiac (20-40%) and cutaneous, Testicular tumors-large-cell calcifying Sertoli cell tumors, Pituitary tumors (10-20%), Thyroid (10%), Schwannomas-psammomatous melanotic (up to 10%), Primary pigmented nodular adrenocortical disease (25-60%)
	<i>PTCH1</i>	Basal cell carcinoma (up to 90%), Brain-medulloblastoma (~2%), Fibromas-cardiac and ovarian, Jaw tumors-odontogenic keratocysts, Meningioma
	<i>PTEN</i>	Female breast (25-85%), Thyroid (3-38%), Endometrial (5-28%), Colorectal, Renal, Melanoma, Gastrointestinal polyps, Lhermitte-Duclos disease
	<i>RB1</i>	Retinoblastoma (greater than 90%), Brain-pineoblastoma (5-10%), Soft tissue sarcoma-leiomyosarcoma and rhabdomyosarcoma, Osteosarcoma, Melanoma, Retinoma, Bladder cancer, Lung cancer
	<i>RET</i>	Thyroid-medullary (greater than 90%), Pheochromocytoma (up to 50%), Hyperparathyroidism (up to 30%)
	<i>SDHB</i>	Paranglioma/Pheochromocytoma, Renal, Gastrointestinal stromal tumor (GIST)
	<i>SDHD</i>	Paranglioma/Pheochromocytoma , Renal, Gastrointestinal stromal tumor (GIST)
	<i>SMAD4</i>	Colorectal (up to 68%), Gastric (up to 21% if gastric polyps), Small bowel, Pancreatic, Gastrointestinal polyps
	<i>STK11</i>	Female breast (up to 54%), Colorectal (39%), Pancreatic (11-36%), Gastric (29%), Ovarian tumors (21%), Lung (7-17%), Small bowel (13%), Cervical (10%), Testicular tumors (9%), Endometrial (9%), Gastrointestinal polyps
	<i>TERC</i>	Acute myeloid leukemia (AML), Myelodysplastic syndrome (MDS), Bone marrow failure, Squamous cell carcinoma-head and neck, Anogenital cancers
	<i>TERT</i>	Acute myeloid leukemia (AML), Myelodysplastic syndrome (MDS), Bone marrow failure, Squamous cell carcinoma-head and neck, Anogenital cancers
	<i>TP53</i>	Female breast (85%), Soft tissue sarcoma, Osteosarcoma, Brain, Hematologic malignancies-Acute leukemias among others, Adrenocortical carcinoma, among others. Overall risk for cancer: up to 95% in females, 88% in males
	<i>TSC1</i>	Renal cancer (5%) and tumors, Benign central nervous system tumors-subependymal nodules and subependymal giant cell astrocytomas, Hamartomatous tumors-cardiac rhabdomyomas and angiomyolipomas
	<i>TSC2</i>	Renal cancer (5%) and tumors, Benign central nervous system tumors-subependymal nodules and subependymal giant cell astrocytomas, Hamartomatous tumors-cardiac rhabdomyomas and angiomyolipomas
	<i>VHL</i>	Renal-clear cell (up to 69%), Hemangioblastomas-retinal and central nervous system (50-80%), Pheochromocytomas (11-19%), Pancreatic neuroendocrine tumors (8-17%), Endolymphatic sac tumors (up to 10%)
	<i>WT1</i>	Wilms tumor (up to 74%)

	Gene	Lifetime Cancer and/or Tumor Risks*
Moderate-Risk Genes	<i>ATM</i>	Female breast (27-33%), Colorectal, Ovarian, Pancreatic, Prostate
	<i>BRIP1</i>	Ovarian, Prostate, Female Breast
	<i>CHEK2</i>	Female breast, Male breast, Colorectal, Gastric, Prostate, Renal, Thyroid
	<i>RAD51C</i>	Ovarian, Female breast, Prostate
	<i>RAD51D</i>	Ovarian, Female breast, Prostate

	Gene	Lifetime Cancer and/or Tumor Risks*
Newer Genes	<i>AIP</i>	Pituitary adenomas (benign pituitary tumors)
	<i>ANKRD26</i>	Thrombocytopenia, Acute myeloid leukemia (AML) among others, Myelodysplastic syndrome (MDS), Chronic myeloid leukemia (CML)
	<i>AXIN2</i>	Colorectal, Colon polyps
	<i>BAP1</i>	Renal, Melanoma-cutaneous and uveal, Mesothelioma, Basal cell carcinoma
	<i>BARD1</i>	Female breast
	<i>CASR</i>	Hyperparathyroidism, Parathyroid tumors
	<i>CDK4</i>	Melanoma
	<i>CDKN1B</i>	Hyperparathyroidism, Pituitary tumors, Gastro-entero-pancreatic neuroendocrine tumors, Parathyroid tumors
	<i>CEBPA</i>	Acute myeloid leukemia (AML)
	<i>DDX41</i>	Acute myeloid leukemia (AML), Myelodysplastic syndrome (MDS)

	Gene	Lifetime Cancer and/or Tumor Risks*
Newer Genes	<i>DICER1</i>	Lung tumors-pleuropulmonary blastoma, Thyroid tumors-multinodular thyroid goiter and cancer, Renal tumors-cystic nephroma, Ovarian tumors-Sertoli-Leydig, Embryonal rhabdomyosarcoma-cervix, Pituitary blastoma, Pineoblastoma
	<i>ETV6</i>	Thrombocytopenia, Acute lymphocytic leukemia (ALL), Acute myeloid leukemia (AML), Myelodysplastic syndrome (MDS)
	<i>FANCC</i>	Female breast
	<i>FANCM</i>	Female breast
	<i>GATA2</i>	Myelodysplastic syndrome (MDS), Acute myeloid leukemia (AML)
	<i>HOXB13</i>	Prostate
	<i>KIT</i>	Gastrointestinal stromal tumors (GIST)
	<i>LZTR1</i>	Schwannomas – spinal, peripheral, cranial nerve and vestibular
	<i>MAX</i>	Paranglioma/Pheochromocytoma
	<i>MET</i>	Renal-type I papillary
	<i>MITF</i>	Renal, Melanoma
	<i>NBN</i>	Female breast, Non-Hodgkin lymphoma, Prostate
	<i>NTHL1</i>	Colorectal, Colon polyps
	<i>PDGFRA</i>	Gastrointestinal stromal tumors (GIST)
	<i>POLD1</i>	Colorectal, Endometrial, Colon polyps
	<i>POLE</i>	Colorectal, Colon polyps
	<i>POT1</i>	Melanoma, Brain-glioma
	<i>RECQL</i>	Female breast
	<i>RUNX1</i>	Myelodysplastic syndrome (MDS), Acute myeloid leukemia (AML) among others, Platelet disorder
	<i>SAMD9</i>	Myelodysplastic syndrome (MDS), Acute myeloid leukemia (AML)
	<i>SAMD9L</i>	Myelodysplastic syndrome (MDS), Acute myeloid leukemia (AML)
	<i>SCG5/ GREM1</i>	Colorectal, Colon polyps
	<i>SDHA</i>	Paranglioma/Pheochromocytoma, Gastrointestinal stromal tumor (GIST)
	<i>SDHAF2</i>	Paranglioma/Pheochromocytoma
	<i>SDHC</i>	Paranglioma/Pheochromocytoma , Renal, Gastrointestinal stromal tumor (GIST)
	<i>SMARCA4</i>	Malignant rhabdoid tumors-atypical teratoid/rhabdoid tumor of the brain and malignant rhabdoid tumors of the kidney, Ovarian-small cell carcinoma of the ovary, hypercalcemic type
	<i>SMARCB1</i>	Malignant rhabdoid tumors-atypical teratoid/rhabdoid tumor of the brain and malignant rhabdoid tumors of the kidney, Schwannomas, Meningiomas
<i>SMARCE1</i>	Meningiomas clear cell type-cranial and spinal	
<i>SRP72</i>	Myelodysplastic syndrome (MDS), Aplastic anemia, Pancytopenia	
<i>SUFU</i>	Brain-medulloblastoma, Basal cell carcinoma, Meningioma	
<i>TMEM127</i>	Paranglioma/Pheochromocytoma	

\* Most commonly associated cancers/tumors listed; lifetime risks provided when available. Risks relate to carriers of a single pathogenic variant with the exception of the *MUTYH* and *NTHL1* genes.

\*\* Tumor spectrum is representative of Lynch syndrome; data are limited with regard to the association of certain cancers with pathogenic variants in *MSH6*, *PMS2* and *EPCAM*.

# Hereditary Cancer Testing Panels

More Comprehensive

More Specific

Panel	Genes	Number of Genes:	TAT:
<b>CUSTOM</b>	<b>OncobedX Custom Panel</b> (B749)	82	3 weeks
<b>MULTIPLE CANCERS</b>	<b>Common Cancer Panel</b> (B275)	47	2 weeks
	<b>Common Cancer Management Panel</b> (B751)	37	2 weeks
<b>BREAST/GYNECOLOGIC CANCER</b>	<b>Breast Cancer Management Panel</b> (J055)	9	2 weeks (8-10 day RUSH)
	<b>Breast/Gyn Cancer Panel</b> (B273)	24	2 weeks
<b>COLORECTAL CANCER</b>	<b>Lynch/ Colorectal High Risk Panel</b> (B522)	7	2 weeks
	<b>Colorectal Cancer Panel</b> (B274)	20	2 weeks
<b>TUMOR SPECIFIC PANELS</b>	<b>Melanoma Panel</b> (B399)	9	2 weeks
	<b>Pancreatic Cancer Panel</b> (B343)	15	2 weeks
	<b>Pediatric Tumor Panel</b> (J318)	27	3 weeks
	<b>PCL/PCC Panel</b> (B395)	12	3 weeks
	<b>Hereditary Prostate Cancer Panel</b> (J665)	16	2 weeks
	<b>Renal Cancer Panel</b> (B394)	18	3 weeks
	<b>Hereditary MDS/Leukemia Panel</b> (T830)	12	3 weeks
	<b>Hyperparathyroidism/Endocrine Tumor Panel</b> (T828)	11	3 weeks
<b>Brain Tumor Panel</b> (T831)	23	3 weeks	

Test Code:

Number of Genes: TAT: