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Hereditary Cancer Panel

Pathogenic variants in the genes analyzed by this panel cause variable phenotypes and cancer risks, and have been implicated in hereditary cancer. Hereditary predisposition is often characterized by early age of cancer onset (typically before age 50), and the development of two or more cancers, multifocal cancers, or similar cancers in an individual or in a closely related family member(s). See Genes Tested table below for more details regarding the genes and syndromes included on the Hereditary Cancer Panel.

DISEASE OVERVIEW

Etiology

Approximately 5-10% of cancer is associated with a hereditary cause.

Inheritance

- All genes tested on this panel are autosomal dominant with the exception of:
  - *SDHD* gene – autosomal dominant with paternal parent-of-origin effect
  - *MAX* and *SDHAF2* genes – autosomal dominant with possible paternal parent-of-origin effect
  - *MUTYH* gene – autosomal recessive but may also have autosomal dominant risks that are not well-defined
  - *MSH3* and *NTHL1* – autosomal recessive
- Some genes are associated with autosomal recessive childhood cancer predisposition or other syndromes.
- See table below for additional details.

TEST DESCRIPTION

See Genes Tested table for genes included in the panel.

Clinical Sensitivity

Variable, dependent on phenotype/condition

Testing Strategy

- Indications for ordering
  - To diagnose a hereditary cancer syndrome in an individual with a personal and/or family history consistent with more than one cancer syndrome
- Contraindications for ordering
  - Should not be ordered to detect somatic variants associated with malignancy because sensitivity for mosaic variants is low with methodology used for germline assays
  - Individuals with hematological malignancy and/or a previous allogenic bone marrow transplant should not undergo molecular genetic testing on peripheral blood specimen.
    - Testing of cultured fibroblasts is required for accurate interpretation of test results.
  - When a relative has a previously identified pathogenic variant, see Familial Mutation, Targeted Sequencing (2001961).

Limitations

- A negative result does not exclude a heritable form of cancer.
- Diagnostic errors can occur due to rare sequence variations.
- Interpretation of this test result may be impacted if this individual has had an allogeneic stem cell transplantation.
- The following will not be evaluated:
  - Variants outside the coding regions and intron-exon boundaries of the targeted genes
  - Regulatory region variants and deep intronic variants
  - Breakpoints of large deletions/duplications
  - Deletions/duplications in *AXIN2, MSH3, NF1, RECQL, SMARCA4, WT1*

TESTS TO CONSIDER

Hereditary Cancer Panel, Sequencing and Deletion/Duplication 2012032
Method: Massively Parallel Sequencing/Exonic Oligonucleotide-based CGH Microarray

Indication for testing:

- Recommended test to confirm a diagnosis of a hereditary cancer syndrome in individuals with personal or family history consistent with features of more than one cancer syndrome.
- When a relative has a previously identified pathogenic sequence variant, see Familial Mutation, Targeted Sequencing (2001961).

Familial Mutation, Targeted Sequencing 2001961
Method: Polymerase Chain Reaction/Sequencing

Indication for testing:

- Recommended test if there is a known familial sequence variant previously identified in a family member.
- A copy of the family member's test result documenting the familial variant is required.

See Related Tests

- Sequence variants in *EPCAM*
- Noncoding transcripts
- The following exons are not sequenced due to technical limitations of the assay:
  - *CHEK2* (NM_001349956) 4; (NM_001005735) 3; (NM_007194) 10, 12, 13, 14, 15
  - *RECQL* (NM_002907) 14, 15
  - *SDHC* (NM_001035511) 5
  - *SDHD* (NM_001276506) 4

- The following may not be detected:
  - Deletions/duplications/insertions of any size by massively parallel sequencing
  - Deletions/duplications less than 1kb in the targeted genes
  - Some variants due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions
  - Low-level somatic variants
  - Single exon deletions/duplications in the following exons:

Gene	Exon(s)
APC	(NM_001127511) 1
BAP1	(NM_004656) 1
BARD1	(NM_000465) 1
BMPR1A	(NM_004329) 9
BRCA1	(NM_007300) 13
CDH1	(NM_004360) 1
CDKN2A	(NM_000077) 2
CDKN2A	(NM_058195) 2
CHEK2	(NM_001005735) 3
CHEK2	(NM_007194) 11, 12, 14, 15
FH	(NM_000143) 1
FLCN	(NM_144997) 8
MAX	(NM_001320415) 5
MAX	(NM_145113) 5
MRE11	(NM_005591) 2
MSH2	(NM_000251) 1
MSH2	(NM_001258281) 2
MSH6	(NM_000179) 10
MUTYH	(NM_001128425) 1
NF2	(NM_000268) 7, 13, 16
NTHL1	(NM_002528) 3, 4, 5, 6
PALB2	(NM_024675) 1
POLD1	(NM_002691) 6, 18, 25
PTEN	(NM_000314) 8, 9
PTEN	(NM_001304717) 1
RAD51D	(NM_002878) 1
RB1	(NM_000321) 1
RET	(NM_020975) 1
SDHD	(NM_001276506) 4
SMARCB1	(NM_003073) 5
SUFU	(NM_001178133) 11
SUFU	(NM_016169) 1

Gene	Exon(s)
TP53	(NM_001126113) 10
TP53	(NM_001126114) 10
TSC2	(NM_000548) 17, 29, 41
VHL	(NM_000551) 1

Analytical Sensitivity

For massively parallel sequencing:

Variant Class	Analytical Sensitivity (PPA) Estimate[a] (%)	Analytical Sensitivity (PPA) 95% Credibility Region[a] (%)
SNVs	99.2	96.9-99.4
Deletions 1-10 bp	93.8	84.3-98.2
Deletions 11-44 bp	100	87.8-100
Insertions 1-10 bp	94.8	86.8-98.5
Insertions 11-23 bp	100	62.1-100

[a]Genes included on this test are a subset of a larger methods-based validation from which the PPA values are derived.

bp, base pairs; PPA, positive percent agreement; SNVs, single nucleotide variants

Genes Tested

Gene	MIM Number	Disorder/Associated Cancer(s)/Tumor(s)	Inheritance
ALK	105590	Associated cancer(s)/tumor(s): neuroblastoma, ganglioneuroblastoma, ganglioneuroma	AD
APC	611731	Familial adenomatous polyposis (FAP) Attenuated FAP (AFAP) Associated cancer(s)/tumor(s): colon, duodenal, thyroid, pancreas, stomach, medulloblastoma, hepatoblastoma	AD
ATM	607585	Associated cancer(s)/tumor(s): breast, ovarian, [a] colorectal [a]	AD
		Ataxia-telangiectasia (AT)	AR
ATR	601215	Familial cutaneous telangiectasia and cancer syndrome (FCTCS) Associated cancer(s)/tumor(s): oropharyngeal	AD
		Seckel syndrome 1	AR
AXIN2	604025	Oligodontia-colorectal cancer syndrome (OSCRCS) Associated cancer(s): colon [a]	AD
BAP1	603089	*BAP1* tumor predisposition syndrome (*BAP1*-TPDS) Associated cancer(s)/tumor(s): uveal melanoma, malignant mesothelioma, cutaneous melanoma, renal cell carcinoma, basal cell carcinoma	AD
BARD1	601593	Associated cancer(s)/tumor(s): breast [a]	AD
BMPR1A	601299	Juvenile polyposis syndrome (JPS) Associated cancer(s)/tumor(s): colon, stomach, small intestine, pancreas	AD
BRCA1	113705	Hereditary breast and ovarian cancer (HBOC) syndrome Associated cancer(s)/tumor(s): breast, ovarian, prostate, pancreas, melanoma	AD
		Fanconi anemia, complementation group S	AR
BRCA2	600185	Hereditary breast and ovarian cancer (HBOC) syndrome Associated cancer(s)/tumor(s): breast, ovarian, prostate, pancreas, melanoma	AD
		Fanconi anemia, complementation group D1	AR
BRIP1	605882	Associated cancer(s)/tumor(s): ovarian, breast [a]	AD
		Fanconi anemia, complementation group J	AR

Gene	MIM Number	Disorder/Associated Cancer(s)/Tumor(s)	Inheritance
CDH1	192090	Hereditary diffuse gastric cancer (HDGC) Associated cancer(s)/tumor(s): diffuse gastric, lobular breast	AD
CDK4	123829	Associated cancer(s)/tumor(s): cutaneous melanoma	AD
CDKN1B	600778	Multiple endocrine neoplasia (MEN) Type 4 Associated cancer(s)/tumor(s): parathyroid, pituitary, gastrinoma, insulinoma, gastro-entero-pancreatic (GEP), carcinoid, adrenocortical, nonendocrine	AD
CDKN2A	600160	Familial atypical multiple mole melanoma-pancreatic carcinoma (FAMMM-PC) syndrome (also known as melanoma-pancreatic cancer syndrome) Associated cancer(s)/tumor(s): cutaneous melaoma, pancreas	AD
CHEK2	604373	Associated cancer(s)/tumor(s): breast, colorectal, [a] prostate, [a] thyroid [a]	AD
DICER1	606241	*DICER1*-related disorders Associated cancer(s)/tumor(s): pleuropulmonary blastoma, ovarian sex cord-stromal tumors, cystic nephroma, thyroid	AD
EPCAM	185535	Lynch syndrome/hereditary nonpolyposis colorectal cancer (HNPCC) Associated cancer(s)/tumor(s): colorectal, endometrial, stomach, ovarian, and others	AD
FH	136850	Hereditary leiomyomatosis and renal cell cancer (HLRCC) Associated cancer(s)/tumor(s): papillary type 2 renal cancer, cutaneous and uterine leiomyomata	AD
		Fumarase deficiency	AR
FLCN	607273	Birth-Hogg-Dube syndrome (BHDS) Associated cancer(s)/tumor(s): renal	AD
MAX	154950	Associated cancer(s)/tumor(s): pheochromocytoma, paraganglioma	AD[b]
MEN1	613733	Multiple endocrine neoplasia (MEN) type 1 Associated cancer(s)/tumor(s): parathyroid, pituitary, gastrinoma, insulinoma, carcinoid, adrenocorticol	AD
MET	164860	Hereditary papillary renal cell carcinoma (HPRCC) Associated cancer(s)/tumor(s): papillary type 1 renal cancer	AD
MLH1	120436	Lynch syndrome/hereditary nonpolyposis colorectal cancer (HNPCC) Associated cancer(s)/tumor(s): colorectal, endometrial, stomach, ovarian, and others	AD
		Constitutional mismatch repair deficiency (CMMRD)	AR
MRE11/MRE11A	600814	Associated cancer(s)/tumor(s): breast [a]	AD
		Ataxia-telangiectasia-like disorder	AR
MSH2	609309	Lynch syndrome/hereditary nonpolyposis colorectal cancer (HNPCC) Associated cancer(s)/tumor(s): colorectal, endometrial, stomach, ovarian, and others	AD
		Constitutional mismatch repair deficiency (CMMRD)	AR
MSH3	600887	Associated cancer(s)/tumor(s): polyposis, colorectal [a]	AR
MSH6	600678	Lynch syndrome/hereditary nonpolyposis colorectal cancer (HNPCC) Associated cancer(s)/tumor(s): colorectal, endometrial, stomach, ovarian, and others	AD
		Constitutional mismatch repair deficiency (CMMRD)	AR
MUTYH	604933	Associated cancer(s)/tumor(s): breast [a]	AD
		MUTYH-Associated Polyposis (MAP) Associated cancer(s)/tumor(s): colon, duodenal	AR
NBN	602667	Associated cancer(s)/tumor(s): breast	AD
		Nijmegan Breakage syndrome (NBS)	AR

Gene	MIM Number	Disorder/Associated Cancer(s)/Tumor(s)	Inheritance
NF1	613113	Neurofibromatosis type 1 (NF1) Associated cancer(s)/tumor(s): breast, neurofibromas, gliomas, malignant peripheral nerve sheath tumors, gastrointestinal stromal tumor (GIST), leukemia	AD
NF2	607379	Neurofibromatosis type 2 (NF2) Associated cancer(s)/tumor(s): schwannoma, meningioma	AD
NTHL1	602656	Associated cancer(s)/tumor(s): polyposis, colorectal [a]	AR
PALB2	610355	Associated cancer(s)/tumor(s): breast, pancreatic [a]	AD
		Fanconi Anemia, complementation group N	AR
PHOX2B	603851	Congenital central hypoventilation syndrome (CCHS) Associated cancer(s)/tumor(s): neuroblastoma, ganglioneuroblastoma, ganglioneuroma	AD
PMS2	600259	Lynch syndrome/hereditary nonpolyposis colorectal cancer (HNPCC) Associated cancer(s)/tumor(s): colorectal, endometrial, stomach, ovarian, and others	AD
		Constitutional mismatch repair deficiency (CMMRD)	AR
POLD1	174761	Associated cancer(s)/tumor(s): polyposis, colorectal [a]	AD
POLE	174762	Associated cancer(s)/tumor(s): polyposis, colorectal [a]	AD
PTEN	601728	Cowden syndrome/*PTEN* hamartoma tumor syndrome Associated cancer(s)/tumor(s): breast, endometrial, thyroid, colon, renal cell carcinoma	AD
RAD51C	602774	Associated cancer(s)/tumor(s): ovarian	AD
		Fanconi anemia, complementation group O	AR
RAD51D	602954	Associated cancer(s)/tumor(s): ovarian	AD
RB1	614041	Retinoblastoma Associated cancer(s)/tumor(s): retinoblastoma, retinoma, pinealoblastoma, osteosarcoma, soft tissue sarcoma, melanoma	AD
RECQL	600537	Associated cancer(s)/tumor(s): breast [a]	AD
RET	164761	Multiple endocrine neoplasia type 2 (MEN2) Associated cancer(s)/tumor(s): medullary thyroid carcinoma, pheochromocytoma, parathyroid adenoma	AD
SDHAF2	613019	Associated cancer(s)/tumor(s): paraganglioma	AD [b]
SDHB	185470	Associated cancer(s)/tumor(s): paraganglioma, pheochromocytoma, GIST, pulmonary chondroma, renal clear cell carcinoma	AD
SDHC	602413	Associated cancer(s)/tumor(s): paraganglioma, pheochromocytoma, GIST, pulmonary chondroma, renal clear cell carcinoma	AD
SDHD	602690	Associated cancer(s)/tumor(s): paraganglioma, pheochromocytoma, GIST, pulmonary chondroma, renal clear cell carcinoma	AD [c]
SMAD4	600993	Juvenile polyposis syndrome (JPS); hereditary hemorrhagic telangiectasia (HHT) syndrome Associated cancer(s)/tumor(s): colon, stomach, small intestine, pancreas	AD
SMARCA4	603254	Rhabdoid tumor predisposition syndrome Associated cancer(s)/tumor(s): rhabdoid tumor	AD
SMARCB1	601607	Rhabdoid tumor predisposition syndrome Associated cancer(s)/tumor(s): rhabdoid tumor	AD
STK11	602216	Peutz-Jeghers syndrome (PJS) Associated cancer(s)/tumor(s): breast, colon, stomach, small intestine, pancreas, ovary, testes, lung	AD
SUFU	607035	Nevoid basal cell carcinoma syndrome (NBCCS) Associated cancer(s)/tumor(s): basal cell carcinoma, medulloblastoma, fibroma, rhabdomyoma	AD

Gene	MIM Number	Disorder/Associated Cancer(s)/Tumor(s)	Inheritance
TMEM127	613403	Associated cancer(s)/tumor(s): pheochromocytoma, paraganglioma, GIST, pulmonary chondroma, renal clear cell carcinoma	AD
TP53	191170	Li-Fraumeni syndrome (LFS) Associated cancer(s)/tumor(s): soft tissue sarcoma, osteosarcoma, central nervous system (CNS) tumor, breast, adrenocortical carcinoma, choroid plexus carcinoma, rhabdomyosarcoma	AD
TSC1	605284	Tuberous sclerosis complex (TSC) Associated cancer(s)/tumor(s): cardiac rhabdomyoma, retinal and other hamartomas, renal angiomyolipoma, subependymal giant cell astrocytoma (SEGA), fibromas	AD
TSC2	191092	Tuberous sclerosis complex (TSC) Associated cancer(s)/tumor(s): cardiac rhabdomyoma, retinal and other hamartomas, renal angiomyolipoma, subependymal giant cell astrocytoma (SEGA), fibromas	AD
VHL	608537	Von Hippel-Lindau (VHL) syndrome Associated cancer(s)/tumor(s): hemangioblasoma, retinal angioma, renal cell carcinoma, pheochromocytoma, neuroendocrine tumors, endolymphatic sac tumors, epididymal and broad ligament cystadenomas	AD
WT1	607102	*WT1*-related Wilms tumor WAGR syndrome Denys-Drash syndrome (DDS) Frasier syndrome Associated cancer(s)/tumor(s): Wilms tumor	AD

[a]Association is suggested but not well-established at this time

[b]Possible paternal parent-of-origin effect

[c]Paternal parent-of-origin effect

AD, autosomal dominant; AR, autosomal recessive

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RELATED TESTS

Breast and Ovarian Hereditary Cancer Syndrome (BRCA1 and BRCA2) Sequencing and Deletion/Duplication 2011949
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Familial Adenomatous Polyposis Panel: (APC) Sequencing and Deletion/Duplication, (MUTYH) 2 Mutations 2004915
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Hereditary Breast and Ovarian Cancer Panel, Sequencing and Deletion/Duplication 2012026
Method: Massively Parallel Sequencing/Exonic Oligonucleotide-based CGH Microarray

Hereditary Cancer Panel, Sequencing and Deletion/Duplication 2012032
Method: Massively Parallel Sequencing/Exonic Oligonucleotide-based CGH Microarray

Hereditary Gastrointestinal Cancer Panel, Sequencing and Deletion/Duplication 2013449
Method: Massively Parallel Sequencing/Exonic Oligonucleotide-based CGH Microarray/Sequencing/Multiplex Ligation-dependent Probe Amplification

Hereditary Paraganglioma-Pheochromocytoma (SDHB, SDHC, and SDHD) Sequencing and Deletion/Duplication Panel 2007167
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Hereditary Renal Cancer Panel, Sequencing and Deletion/Duplication 2010214
Method: Massively Parallel Sequencing/Exonic Oligonucleotide-based CGH Microarray

HNPCC/Lynch Syndrome (MLH1) Sequencing and Deletion/Duplication 0051650
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

HNPCC/Lynch Syndrome (MSH2) Sequencing and Deletion/Duplication 0051654
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

HNPCC/Lynch Syndrome (MSH6) Sequencing and Deletion/Duplication 0051656
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

HNPCC/Lynch Syndrome (PMS2) Sequencing and Deletion/Duplication 0051737
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Juvenile Polyposis (SMAD4) Sequencing and Deletion/Duplication 2001971
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Juvenile Polyposis Syndrome (BMPR1A) Sequencing and Deletion/Duplication 2004992
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Li-Fraumeni (TP53) Sequencing and Deletion/Duplication 2009313
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Multiple Endocrine Neoplasia Type 1 (MEN1) Sequencing and Deletion/Duplication 2005360
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Multiple Endocrine Neoplasia Type 2 (MEN2), RET Gene Mutations by Sequencing 0051390
Method: Polymerase Chain Reaction/Sequencing

Neurofibromatosis Type 1 (NF1) Sequencing and Deletion/Duplication 2007154
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

Peutz-Jeghers Syndrome (STK11) Sequencing and Deletion/Duplication 2008398
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

PTEN-Related Disorders (PTEN) Sequencing and Deletion/Duplication 2002470
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

von Hippel-Lindau (VHL) Sequencing and Deletion/Duplication 2002965
Method: Polymerase Chain Reaction/Sequencing/Multiplex Ligation-dependent Probe Amplification

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