GenHet Cancer Predisposition

GENOME EXPLORATIONS
A DIVISION OF COMPASS LABORATORY SERVICES LLC

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Introduction

GenHet Cancer Predisposition Panel is a multiplexed PCR-based assay for targeted enrichment of the coding (exonic) regions of the 143 genes commonly mutated in 88 inherited oncogenic diseases. One of the first genes identified as causing a familial cancer syndrome was BRCA1, which increases predisposition to breast and ovarian cancer. Families who are positive for common heritable oncogenic diseases have the opportunity for enhanced cancer surveillance, and may be able to avoid a poor prognosis if a tumor is identified early. There are many heritable diseases that significantly increase familial cancer incidence. For some of these diseases cancer incidence is the only phenotype, while other diseases have additional phenotypes. This panel includes all genes that are known to cause heritable diseases that result in cancer in at least 50% of affected individuals, as well as other genes commonly mutated in cancer. Many of these genes have key mutations annotated that predispose a person to cancer. However, there are potentially other unknown disease-causing mutations in each identified gene. This panel allows researchers to comprehensively study genes involved in heritable cancer syndromes, and was developed using a variety of resources such as OMIM® (Online Mendelian Inheritance in Man), ClinVar, dbSNP, and the Familial Cancer Database.

GenHet Cancer Predisposition Genes

Cancers:

Bloom Syndrome: BLM
Carney Complex: PRKAR1A
Costello Syndrome: HRAS
Cowden Syndrome: AKT1, PIK3CA, PTEN
Fanconi Anemia: BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, SLX4 (BTBD12)
Hereditary Desmoid Disease: APC
Li-Fraumeni Syndrome: CHEK2 (RAD53), TP53
LIG4 Syndrome: LIG4
Lynch Syndrome: EPCAM, MLH1, MSH2, MSH6, PMS2
Mismatch Repair Cancer Syndrome: MLH1, MSH2, MSH6, PMS2
Mosaic Variegated Aneuploidy: BUB1B
Seckel Syndrome 1: ATR
von Hippel-Lindau Syndrome: VHL

Bone Cancers:

Familial Osteosarcoma: TP53
Hereditary Bone Dysplasia with Malignant Fibrous Histiocytoma: MTAP
Multiple Exostoses: EXT1, EXT2
Rothmund-Thompson Syndrome: RECQL4

Breast/Ovarian Cancers:

Hereditary Breast-Ovarian Cancer: BARD1, BRCA1, BRCA2, CHEK2 (RAD53), RAD51B (RAD51L1), RAD51C, RAD51D (RAD51L3)

Colorectal Cancers:

Hereditary Adenomatous Polyposis: APC, MUTYH
Hereditary Colorectal Cancer: GALNT12, MLH3
Oligodontia-Colorectal Cancer Syndrome: AXIN2
POLD1 & POLE Associated Colorectal Adenomas: POLD1, POLE

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Endocrine Cancers:
Hereditary Adrenal Pheochromocytoma: MAX, TMEM127, VHL
Hereditary Primary Pigmented Nodular Adrenocortical Disease: PDE11A, PRKAR1A
Hereditary Adrenocortical Cancer: TP53
Hereditary Adrenal Hyperplasia: CYP21A2
Hereditary Isolated Pituitary Adenoma: AIP
Hereditary Thyroid Cancer: NDUFA13, NTRK1, RET
Hyperparathyroidism-Jaw Tumor Syndrome: CDC73
Multiple Endocrine Neoplasia: CDKN1B (p27KIP1), MEN1
Pallister-Hall Syndrome: GLI3

GI Tract Cancers:
Bannayan-Riley-Ruvalcaba Syndrome: PTEN
Hereditary Barrett Esophagus/Esophageal Adenocarcinoma: ASCC1, MSR1
Hereditary Gastric Carcinoma: CDH1
Hereditary GIST: KIT (CD117), PDGFRA, SDHC
Howel-Evans Syndrome: RHBDF2
Juvenile Polyposis Syndrome: BMPR1A (ALK3), SMAD4
Peutz-Jeghers Syndrome: STK11 (LKB1)

Head & Neck Cancers:
Dyskeratosis Congenita: DKC1, RTEL1, TERT, TINF2
Hereditary Cylindromatosis: CYLD
Tuberous Sclerosis: TSC1, TSC2

Hematopoietic Cancers:
Ataxia Telangetasia: ATM
Ataxia Telangetasia-like Disorder: MRE11A
Chediak-Higashi Syndrome: LYST
Familial Monocytic Leukemia: GATA2
Hereditary Acute Myeloid Leukemia: CEBPA, RUNX1 (AML1)
Hereditary Hodgkin Lymphoma: KLHDC8B
Nijmegen Breakage Syndrome: NBN (NBS1)
Nijmegen Breakage-like Syndrome: RAD50
Schwachman-Diamond Syndrome: SBDS
TERT Mutation-Associated Haematological Disorders: TERT
Wiskott-Aldrich Syndrome: WAS

Kidney Cancers:
Birt-Hogg-Dube Syndrome: FLCN
Denys-Drash Syndrome: WT1
Hereditary Leiomyomatosis & Renal Cell Cancer: FH
Hereditary Melanoma & Renal Cancer: MET, MITF
Hereditary Wilms' Tumor: POU6F2, WT1
Perlman Syndrome: DIS3L2

Liver Cancers:
Hemochromatosis: HFE
Porphyria Cutanea Tarda: UROD
Tyrosinemia: FAH

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Nervous System Cancers:
Carney-Stratakis Syndrome: PRKAR1A, SDHAF2, SDHB, SDHD
Hereditary Medulloblastoma: SUFU
Hereditary Neuroblastoma: ALK, KIF1B, PHOX2B
Hereditary Paragangliomas: SDHA
Neurofibromatosis: NF1, NF2
Retinoblastoma: RB1
Rhabdoid Predisposition Syndrome: SMARCA4, SMARCB1
Schwannomatosis: SMARCB1
Simpson-Golabi-Behmel Syndrome: GPC3

Skin Cancers:
Basal Cell Nevus Syndrome: PTCH1, PTCH2
Birt-Hogg-Dube Syndrome: FLCN
Familial Multiple Glomus Tumors: GLMN
Familial Multiple Trichoepithelioma: CYLD
Familial Uveal Melanoma: BAP1
Ferguson-Smith Syndrome: TGFBR1 (ALK5)
Hereditary Epidermodysplasia Verruciformis: TMC6, TMC8
Hereditary Leiomyomatosis & Renal Cell Cancer: FH
Hereditary Malignant Melanoma: CDK4, CDKN2A (p16INK4), MC1R, MITF, XRCC3
Muir-Torre Syndrome: MSH2
Palmoplantar Keratoderma & Squamous Cell Carcinoma: RSPO1
Xeroderma Pigmentosum: DDB2, ERCC2 (XPD), ERCC3 (XPB), ERCC4, ERCC5, ERCC6, POLH, XPA, XPC

Soft Tissue Cancers:
Hereditary Infantile Hemangioma: ANTXR1, KDR (VEGFR3)
Juvenile Hyaline Fibromatosis: ANTXR2
Opitz Trigonocephaly Syndrome: CD96
Proteus Syndrome: PTEN
SC Phocomelia Syndrome: ESCO2
Werner Syndrome: WRN

Other Cancers:
Familial Pancreatic Cancer: BRCA2, PALB2, PALLD
Hereditary Pleuropulmonary Blastoma: DICER1
Hereditary Prostate Cancer: BRCA2, EHBP1, EPHB2, MSMB, MSR1, RNASEL

Other Gene:
CHEK1