Susceptibility

Cancer Precision Panel

Overview

Hereditary cancer syndromes are encountered in all medical specialties. Although they account for about 5% of all malignancies, it is of special importance to identify these patients because, unlike patients with sporadic cancers, they require special, long-term care as their predisposition can cause them to develop certain tumors at a relatively early age. These cancers can arise in the lungs, kidneys, liver, pancreas, skin, eyes, heart. Most hereditary cancers are associated with a "germline mutation" that will be present in every cell of the human body. Identification of patients at risk of inherited cancer susceptibility is dependent upon the ability to characterize genes and alterations associated with increased cancer risk as well as gathering a detailed personal and family history aiding in the identification of the mode of inheritance as well as other family members at risk of suffering from this susceptibility. Most hereditary cancer syndromes follow an autosomal dominant inheritance, and the penetrance is high.

The Igenomix Susceptibility Cancer Precision Panel provides a comprehensive analysis of the most common hereditary cancer syndromes using next-generation sequencing (NGS) to fully understand the spectrum of relevant cancer predisposition genes.

Indications

The Igenomix Susceptibility Cancer Precision Panel is indicated in those cases where there is:

- Multiple relatives on the same side of the family with the same or related forms of cancer
- Cancer at an early age.
- Early presentation of an aggressive cancer type.
- Multiple primary cancers in an individual
- Asymptomatic patient with no personal or family history who wishes to know genetic susceptibility to cancer.

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis of a patient with personal or family history suggestive of a hereditary cancer syndrome.
- Early initiation of treatment with a multidisciplinary team for appropriate total body screening, early surgical intervention and/or early screening.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

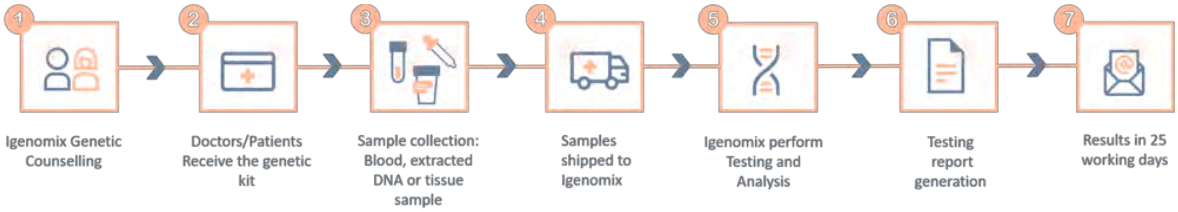
GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
APC	APC-related attenuated Familial Adenomatous Polyposis, Cenani-Lenz Syndrome, Colorectal Cancer, Hereditary Desmoid disease, Desmoid Tumor, Familial Adenomatous Polyposis 1, Familial Adenomatous Polyposis due to 5q22.2 microdeletion, Gardner syndrome, Gastric cancer, Hepatocellular carcinoma, Turcot syndrome with polyposis	AD	98.92	1846 of 1882
ATM	Ataxia-telangiectasia, Breast Cancer, Mantle Cell lymphoma	AD,AR	99.93	1608 of 1632
AXIN2	AXIN2-related attenuated Familial Adenomatous Polyposis, Colorectal Cancer, Oligodontia-Colorectal Cancer Syndrome	AD	99.86	32 of 33
BAP1	Familial Melanoma, Meningioma, Tumor Predisposition Syndrome, Uveal Melanoma	AD	100	194 of 195
BARD1	Breast Cancer, Hereditary Breast and Ovarian Cancer syndrome	AD	99.86	195 of 195
BMPR1A	Familial Colorectal Cancer Type X, Generalized Juvenile Polyposis/Juvenile Polyposis Coli, Hereditary Mixed Polyposis syndrome, Juvenile Polyposis of Infancy, Juvenile Polyposis Syndrome	AD	100	124 of 127
BRCA1	Breast Cancer, Familial Breast-Ovarian Cancer, Familial Pancreatic Carcinoma, Fanconi Anemia Complementation Group S, Hereditary Breast and Ovarian Cancer syndrome, Primary Peritoneal Carcinoma	AD,AR,MU	98.97	2783 of 2894
BRCA2	Breast Cancer, Familial Breast-Ovarian Cancer, Familial Pancreatic Carcinoma, Fanconi Anemia Complementation Group D1, Glioma 3, Hereditary Breast and Ovarian Cancer syndrome, Medulloblastoma, Nephroblastoma, Pancreatic Cancer, Prostate Cancer, Wilms Tumor 1	AD,AR,MU	98.51	3343 of 3451
BRIP1	Breast Cancer, Fanconi Anemia Complementation Group J, Hereditary Breast and Ovarian Cancer syndrome	AD,AR	94.97	235 of 237
CDH1	Blepharo-Cheilo-Odontic syndrome, Breast Cancer, Cleft lip/palate, Endometrial Carcinoma, Gastric Cancer, Prostate Cancer, Suppressor of Tumorigenicity 8	AD	100	361 of 363
CDK4	Familial Melanoma, Cutaneous Malignant Melanoma, Well-differentiated Liposarcoma	AD	100	22 of 22
CDKN2A	Familial Melanoma, Familial Pancreatic Carcinoma, Li-Fraumeni syndrome, Melanoma-Astrocytoma Syndrome, Melanoma-Pancreatic Cancer Syndrome, Cutaneous Malignant Melanoma	AD	94.99	257 of 262
CHEK2	Breast Cancer, Hereditary Breast and Ovarian Cancer Syndrome, Li-Fraumeni Syndrome 2, Osteosarcoma, Prostate Cancer	AD	99.47	307 of 310
EPCAM	Colorectal Cancer, Hereditary Nonpolyposis type 8, Congenital Diarrhea 5 with Tufting Enteropathy, Lynch Syndrome	AR	99.94	52 of 70
FANCC	Fanconi Anemia Complementation Group C	AR	100	75 of 75
FANCM	Fanconi Anemia, Male infertility with azoospermia or oligozoospermia due to single gene mutation, Premature Ovarian Failure 15, Spermatogenic Failure 28	AR	99.73	59 of 61
FH	Fumarase Deficiency, Hereditary Leiomyomatosis and Renal Cell Cancer, Hereditary Pheochromocytoma-Paraganglioma, Multiple Hereditary Leiomyoma of Skin	AD,AR	100	229 of 232
FLCN	Birt-Hogg-Dube Syndrome, Colorectal Cancer, Familial Spontaneous Pneumothorax, Primary Spontaneous Pneumothorax, Potocki-Lupski Syndrome, Nonpapillary Renal Cell Carcinoma	AD	100	200 of 205
HOXB13	Hereditary Prostate Cancer 9		100	5 of 5
MEN1	Familial Isolated Hyperparathyroidism, Insulinoma, Multiple Endocrine Neoplasia Type 1, Pituitary Gigantism, Prolactinoma	AD	99.9	871 of 876
MET	Autosomal Recessive Deafness 97, Hepatocellular Carcinoma, Osteofibrous Dysplasia, Pediatric Hepatocellular Carcinoma, Papillary Renal Cell Carcinoma	AD,AR	99.8	41 of 41

MITF	Coloboma, Osteopetrosis, Microphthalmia, Macrcephaly, Albinism and Deafness, Familial Melanoma, Cutaneous Malignant Melanoma 8, Tietz Syndrome, Waardenburg Syndrome Type 2A, Waardenburg-Shah Syndrome	AD,AR	100	72 of 72
MLH1	Colorectal Cancer, Hereditary Nonpolyposis Type 2, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.94	1079 of 1118
MSH2	Lynch syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.99	1032 of 1057
MSH6	Colorectal Cancer, Hereditary Nonpolyposis Type 5, Endometrial Carcinoma, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.28	613 of 641
MUTYH	Familial Adenomatous Polyposis 2, Gastric Cancer, MUTYH-Related attenuated Familial Adenomatous Polyposis	AR	100	183 of 183
NBN	Aplastic Anemia, Hereditary Breast and Ovarian Cancer Syndrome, Acute Lymphocytic Leukemia, Nijmegen Breakage Syndrome	AR,MU,P	100	200 of 200
NF1	17q11.2 Microduplication Syndrome, Hereditary Pheochromocytoma-Paraganglioma, Juvenile Myelomonocytic Leukemia, Neurofibromatosis Type 1 due to NF1 mutation or intragenic deletion, Neurofibromatosis-Noonan syndrome, Familial Spinal Neurofibromatosis, Watson Syndrome	AD	97.97	3082 of 3166
NTHL1	Familial Adenomatous Polyposis 3, NTHL1-Related attenuated Familial Adenomatous Polyposis	AR	100	13 of 13
PALB2	Breast Cancer, Familial Pancreatic Carcinoma, Fanconi Anemia Complementation Group N, Hereditary Breast and Ovarian Cancer Syndrome	AD,AR	98.78	601 of 617
PC	Pyruvate Carboxylase Deficiency	AR	100	48 of 48
PMS2	Colorectal Cancer, Hereditary Nonpolyposis Type 4, Lynch Syndrome, Mismatch Repair Cancer Syndrome	AD,AR	97.17	264 of 285
POLD1	Colorectal Cancer 10, Mandibular Hypoplasia, Deafness, Progeroid Features and Lipodystrophy Syndrome, Polymerase proofreading-related Adenomatous Polyposis	AD	100	40 of 41
POLE	Colorectal Cancer 12, Facial Dysmorphism, Immunodeficiency, Livedo and Short Stature, IMAGe Syndrome, Intrauterine Growth Retardation, Metaphyseal Dysplasia, Adrenal Hypoplasia Congenita, Genital Anomalies and Immunodeficiency, Polymerase proofreading-related Adenomatous Polyposis	AD,AR	100	100 of 100
POT1	Familial melanoma, Glioma 9, Cutaneous Malignanty Melanoma	AD	99.76	42 of 47
PTEN	Bannayan-Riley-Ruvalcaba syndrome, Cowden Syndrome, Hereditary Breast and Ovarian Cancer Syndrome, Juvenile Polyposis of Infancy, Lhermitte-Duclos Disease, Macrocephaly/Autism Syndrome, Familial Meningioma, Prostate Cancer, Proteus Syndrome, Segmental Outgrowth-Lipomatosis-Arteriovenous Malformation-Epidermal Nevus Syndrome	AD	99.97	609 of 629
RAD51C	Familial Breast-Ovarian Cancer-1, Fanconi Anemia Complementation Group O, Hereditary Breast and Ovarian Cancer Syndrome	AR	100	130 of 130
RAD51D	Hereditary Breast and Ovarian Cancer Syndrome		100	97 of 97
RECQL	Rapadilino Syndrome, Osteoarcoma, Lymphoma		99.71	32 of 34
RET	Congenital Failure of Autonomic Control, Haddad Syndrome, Hereditary Pheochromocytoma-Paraganglioma, Hirschsprung Disease, Multiple Endocrine Neoplasia type 2A and 2B, Pheochromocytoma, Bilateral Renal Agenesis, Sporadic Pheochromocytoma/Secreting Paraganglioma, Familial Medullary Thyroid Carcinoma	AD	100	453 of 454
SCG5	Hereditary Mixed Polyposis syndrome		100	na of na
SDHB	Carney-Stratakis Syndrome, Cowden Syndrome, Gastrointestinal Stromal Tumor, Hereditary Pheochromocytoma-Paraganglioma, Isolated Succinate-CoQ Reductase Deficiency, Paragangliomas 4, Pheochromocytoma, Sporadic Pheochromocytoma/Secreting Paraganglioma	AD	100	261 of 264
SDHC	Carney-Stratakis Syndrome, Cowden Syndrome, Gastrointestinal Stromal Tumor, Hereditary Pheochromocytoma-Paraganglioma, Paragangliomas 3	AD	99.95	62 of 63
SDHD	Carcinoid Syndrome, Carney-Stratakis Syndrome, Cowden Syndrome, Hereditary Pheochromocytoma-Paraganglioma, Isolated Succinate-CoQ Reductase Deficiency, Mitochondrial Complex 2 Deficiency, Paragangliomas 1, Sporadic Pheochromocytoma, Pheochromocytoma/Secreting Paraganglioma	AD,AR	99.98	164 of 166

SMAD4	Familial Pancreatic Carcinoma, Familial Thoracic Aortic Aneurysm and Aortic Dissection, Generalized Juvenile Polyposis/Juvenile Polyposis Coli, Hereditary Hemorrhagic Telangiectasia, Juvenile Polyposis Syndrome, Myhre Syndrome, Pancreatic Cancer	AD	99.56	136 of 136
STK11	Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular Tumor	AD	81.99	456 of 470
TP53	Adrenocortical Carcinoma, Basal Cell Carcinoma, Bone Marrow Failure Syndrome 5, Breast Cancer, Colorectal Cancer, Essential Thrombocythemia, Familial Pancreatic Carcinoma, Glioma 1, Hepatocellular Carcinoma, Hereditary Breast and Ovarian Cancer Syndrome, Li-Fraumeni Syndrome, Nasopharyngeal Carcinoma, Osteosarcoma, Pancreatic Cancer, Papilloma of Chroid Plexus	AD,MU,P	98.92	557 of 563
TSC1	Focal Cortical Dysplasia of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis Complex, Tuberous Sclerosis-1	AD	99.86	390 of 406
TSC2	Focal Cortical Dysplasia of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis Complex, Tuberous Sclerosis-2	AD	100	1157 of 1159
VHL	Familial Erythrocytosis, Hereditary Pheochromocytoma-Paraganglioma, Pheochromocytoma, Nonpapillary Renal Cell Carcinoma, Sporadic Pheochromocytoma/Secreting Paraganglioma,Von Hippel-Lindau Syndrome	AD,AR	100	511 of 544

* Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial
** HGMD: Number of clinically relevant mutations according to HGMD

Methodology



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