Comprehensive Inherited

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 Comprehensive Inherited 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 Comprehensive Inherited Caner Precision Panel is indicated as a screening and diagnostic test in those cases where there are:

- 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

Clinical Utility

The clinical utility of this panel is:

- Early and accurate genetic diagnosis allowing the most appropriate clinical management of a patient with personal or family history suggestive of a hereditary cancer syndrome.
- Intensified surveillance and participation on early detection programmes for cancer prevention.
- Early initiation of treatment or surgical intervention.
- Risk assessment of asymptomatic family members according to the mode of inheritance and genetic counselling of relatives.

Genes & Diseases

List of genes included in the Comprehensive Inherited Cancer Precision Panel.
Most relevant genes have been classified according to:

High Risk	Well studied Greater than 4-fold risk of developing one or more cancers Can cause a moderate risk for other cancers Guidelines or expert opinion for cancer screening and prevention
Moderate Risk	Well-studied 2- to 4-fold risk of developing one or more cancers May increase risk for other cancers Limited guidelines for screening and prevention
Research	Not as well-studied Precise lifetime risks and tumor spectrum not yet determined Guidelines for screening and prevention are limited or not available

GENE	RISK	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AIP		Acromegaly, Pituitary Adenoma, ACTH-Secreting, Growth Hormone-Secreting, Pituitary Gigantism, Prolactinoma	AD,AR	100%	103 of 106
AKT1	Moderate	Breast Cancer, Colorectal Cancer, Cowden Syndrome, Meningioma, Proteus Syndrome, Suppressor Of Tumorigenicity	AD	100%	6 of 6
ALK		Neuroblastoma		99.84%	16 of 16
ANKRD26		Thrombocytopenia	AD	98.76%	3 of 23
APC	High	APC-Related Attenuated Familial Adenomatous Polyposis, Cenani-Lenz Syndrome, Colorectal Cancer, Desmoid Disease, Familial Adenomatous Polyposis, Gardner Syndrome, Gastric Cancer, Hepatocellular Carcinoma, Turcot Syndrome With Polyposis	AD	98.92%	1846 of 1882
AR		Androgen Insensitivity Syndrome, Complete Androgen Insensitivity Syndrome, X-linked Hypospadias, Kennedy Disease, Partial Androgen Insensitivity Syndrome, Prostate Cancer, Reifenstein Syndrome, X-linked Spinal And Bulbar Muscular Atrophy	AD,X,XR,G	97.96%	NA of NA
ATM	Moderate	Ataxia-Telangiectasia, Breast Cancer, Mantle Cell Lymphoma	AD,AR	99.93%	1608 of 1632
AXIN2		AXIN2-Related Attenuated Familial Adenomatous Polyposis, Colorectal Cancer, Oligodontia	AD	99.86%	32 of 33
BAP1		Familial Melanoma, Meningioma, Tumor Predisposition Syndrome, Uveal Melanoma	AD	100%	194 of 195
BARD1	Moderate	Breast Cancer, Hereditary Breast And Ovarian Cancer Syndrome	AD	99.86%	195 of 195
BLM		Bloom Syndrome	AR	97.19%	133 of 141
BMPR1A	High	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
BRAF		Cardiofaciocutaneous Syndrome, Craniopharyngioma, Leopard Syndrome, Lung Cancer, Noonan Syndrome With Multiple Lentigines	AD	100%	80 of 80
BRCA1	High	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	High	Breast Cancer, Familial Breast-Ovarian Cancer, Familial Pancreatic Carcinoma, Fanconi Anemia Complementation Group D1, Glioma Susceptibility, Hereditary Breast And Ovarian Cancer Syndrome, Medulloblastoma, Nephroblastoma, Pancreatic Cancer, Prostate Cancer, Wilms Tumor	AD,AR,MU	98.51%	3343 of 3451
BRE		Brain Glioma, Synchronous Bilateral Breast Carcinoma		98.20%	NA of NA
BRIP1	Moderate	Breast Cancer, Fanconi Anemia Complementation Group J, Hereditary Breast And Ovarian Cancer Syndrome	AD,AR	94.97%	235 of 237
BUB1B		Colorectal Cancer, Mosaic Variegated Aneuploidy Syndrome	AD,AR	99.84%	30 of 31
CBL		Aggressive Systemic Mastocytosis, Juvenile Myelomonocytic Leukemia, Noonan Syndrome With Or Without Juvenile Myelomonocyticleukemia	AD	100%	46 of 47
CD70		Lymphoproliferative Syndrome	AR	99.89%	4 of 4
CD82		Penile Cancer, Renal Oncocytoma, Chromophobe Renal Cell Carcinoma, Gallbladder Adenocarcinoma		100%	NA of NA
CDC73		Familial Isolated Hyperparathyroidism, Hyperparathyroidism-Jaw Tumor Syndrome, Parathyroid Carcinoma	AD	100%	95 of 95
CDH1	High	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
CDKN1B		Multiple Endocrine Neoplasia Type 1, Multiple Endocrine Neoplasia Type 4	AD	99.99%	19 of 20
CDKN1C		Beckwith-Wiedemann Syndrome, Image Syndrome, Intrauterine Growth Restriction-Short Stature-Early Adult-Onset Diabetes Syndrome, Intrauterine Growth Retardation, Metaphyseal Dysplasia, Adrenal Hypoplasia Congenita And Genital Anomalies	AD	73.58%	55 of 76
CDKN2A	High	Familial Melanoma, Familial Pancreatic Carcinoma, Li-Fraumeni Syndrome, Melanoma-Astrocytoma Syndrome, Melanoma-Pancreatic Cancer Syndrome, Cutaneous Malignant Melanoma	AD	94.99%	257 of 262
CEBPA		Acute Myeloid Leukemia	AD	67.47%	14 of 17
CEP57		Mosaic Variegated Aneuploidy Syndrome	AR	99.64%	6 of 6
CHEK2	Moderate	Breast Cancer, Hereditary Breast And Ovarian Cancer Syndrome, Li-Fraumeni Syndrome, Osteosarcoma, Prostate Cancer	AD	99.47%	307 of 310
CYLD		Brooke-Spiegler Syndrome, Familial Cylindromatosis, Familial Multiple Trichoepithelioma	AD	99%	114 of 116
DDB2		Xeroderma Pigmentosum Complementation Group E	AR	100%	17 of 17
DDX41		Myeloproliferative/Lymphoproliferative Neoplasms	AD	99.99%	56 of 56
DICER1		Familial Multinodular Goiter, Global Developmental Delay, Lung Cysts, Overgrowth, And Wilms Tumor, Multinodular Goiter, Pleuropulmonary Blastoma, Embryonal Rhabdomyosarcoma	AD	99.98%	178 of 180
DIS3L2		Nephroblastoma, Perlman Syndrome	AR	99.99%	12 of 13
DKC1		X-linked Dyskeratosis Congenita, Hoyeraal-Hreidarsson Syndrome	X,XR,G	100%	NA of NA
EFL1		Shwachman-Diamond Syndrome	AR	99.94%	NA of NA
EGFR		Neonatal Inflammatory Skin And Bowel Disease, Lung Cancer, Neonatal Inflammatory Skin And Bowel Disease	AD,AR	100%	27 of 27
ELAC2		Combined Oxidative Phosphorylation Deficiency	AR	100%	32 of 32
ELANE		Autosomal Dominant Severe Congenital Neutropenia, Cyclic Hematopoiesis, Cyclic Neutropenia, Autosomal Dominant Severe Congenital Neutropenia	AD	100%	227 of 227

ENG		Familial Cerebral Saccular Aneurysm, Generalized Juvenile Polyposis/Juvenile Polyposis Coli, Hereditary Hemorrhagic Telangiectasia	AD	100%	467 of 471
EPCAM	High	Hereditary Nonpolyposis Colorectal Cancer Type 8, Congenital Diarrhea With Tufting Enteropathy, Lynch Syndrome	AR	99.94%	52 of 70
ERCC1		Cerebrooculofacioskeletal Syndrome, Cockayne Syndrome Type 2	AR	93.12%	6 of 6
ERCC2		Cerebrooculofacioskeletal Syndrome, Cofs Syndrome, Trichothiodystrophy, Xeroderma Pigmentosum-Cockayne Syndrome Complex, Xeroderma Pigmentosum Complementation Group D	AR	100%	102 of 102
ERCC3		Trichothiodystrophy, Xeroderma Pigmentosum-Cockayne Syndrome Complex, Xeroderma Pigmentosum Complementation Group B	AR	99.98%	24 of 24
ERCC4		Cockayne Syndrome Type 1, Fanconi Anemia Complementation Group Q, Xeroderma Pigmentosum-Cockayne Syndrome Complex, Xeroderma Pigmentosum Complementation Group F, Progeroid Syndrome	AR	99.68%	69 of 72
ERCC5		Cerebrooculofacioskeletal Syndrome, Cofs Syndrome, Xeroderma Pigmentosum-Cockayne Syndrome Complex, Xeroderma Pigmentosum Complementation Group G	AR	99.94%	58 of 58
ETV6		Acute Myeloid Leukemia, Thrombocytopenia	AD	100%	41 of 41
EXO1		Werner Syndrome, Aicardi-Goutieres Syndrome, Lynch Syndrome, Colorectal Cancer		99.86%	17 of 17
EXT1		Chondrosarcoma, Multiple Exostoses Type 1, Multiple Osteochondromas, Trichorhinophalangeal Syndrome Type 2	AD,AR	99.97%	518 of 525
EXT2		Multiple Exostoses Type 2, Multiple Osteochondromas, Potocki-Shaffer Syndrome, Seizures-Scoliosis-Macrocephaly Syndrome	AD,AR	100%	251 of 254
EZH2		Weaver Syndrome	AD	99.82%	40 of 41
FAM111B		Poikiloderma, Hereditary Fibrosing, With Tendon Contractures, Myopathy, and Pulmonary Fibrosis	AD	97.88%	10 of 10
FAM175A		Ovarian Cancer, Breast Cancer, Fanconi Anemia Complementation Group A		94.81%	NA of NA
FANCA		Fanconi Anemia	AR	95.17%	497 of 502
FANCB		Fanconi Anemia Complementation Group B, VACTERL Association With Hydrocephalus	X,XR,G	95.53%	NA of NA
FANCC		Fanconi Anemia Complementation Group C	AR	100%	75 of 75
FANCD2		Fanconi Anemia Complementation Group D2	AR	100%	62 of 63
FANCE		Fanconi Anemia Complementation Group E	AR	97%	17 of 18
FANCF		Fanconi Anemia Complementation Group F	AR	99.31%	17 of 18
FANCG		Fanconi Anemia Complementation Group G		100%	94 of 94
FANCI		Fanconi Anemia Complementation Group I	AR	100%	53 of 54
FANCL		Fanconi Anemia Complementation Group L	AR	100%	25 of 26
FANCM		Fanconi Anemia, Male Infertility With Azoospermia Or Oligozoospermia, Premature Ovarian Failure, Spermatogenic Failure	AR	99.73%	59 of 61
FH	High	Fumarase Deficiency, Hereditary Leiomyomatosis And Renal Cell Cancer, Hereditary Pheochromocytoma-Paraganglioma, Leiomyoma	AD,AR	100%	229 of 232
FLCN	High	Birt-Hogg-Dube Syndrome, Colorectal Cancer, Familial Spontaneous Pneumothorax, Potocki-Lupski Syndrome, Nonpapillary Renal Cell Carcinoma	AD	100%	200 of 205
GALNT12		Colorectal Cancer, Familial Colorectal Cancer Type 10		88.97%	14 of 15
GATA2		Deafness-Lymphedema-Leukemia Syndrome, Dendritic Cell, Monocyte, B Lymphocyte, And Natural Killer Lymphocyte Deficiency, Acute Myeloid Leukemia, Primary Lymphedema, Primary, With Myelodysplasia, Myelodysplastic Syndrome	AD	100%	137 of 142
GEN1		Xeroderma Pigmentosum Complementation Group G		99.71%	6 of 6
GPC3		Nephroblastoma, Simpson-Golabi-Behmel Syndrome Type 1, Wilms Tumor	AD,X,XR,G	99.84%	NA of NA
GREM1		Hereditary Mixed Polyposis Syndrome		99.89%	5 of 5
HNF1A		Insulin-Dependent Diabetes Mellitus, Noninsulin-Dependent Diabetes Mellitus, Familial Hepatic Adenomas, Hyperinsulinism, Maturity-Onset Diabetes Of The Young Type 3, Nonpapillary Renal Cell Carcinoma	AD	100%	529 of 538
HOXB13		Hereditary Prostate Cancer, Spinal Cord Ependymoma, Myxopapillary Ependymoma		100%	5 of 5
HRAS		Bladder Cancer, Costello Syndrome, Epidermal Nevus, Linear Nevus Sebaceus Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Nonmedullary Thyroid Cancer	AD	100%	34 of 34
IKZF1		Common Variable Immunodeficiency, Stevens-Johnson Syndrome	AD	99.98%	43 of 43
KIF1B		Charcot-Marie-Tooth Disease Type 2A1, Hereditary Pheochromocytoma-Paraganglioma, Neuroblastoma, Pheochromocytoma	AD	99.89%	17 of 17
KIT		Bullous Diffuse Cutaneous Mastocytosis, Cutaneous Mastocytoma, Gastrointestinal Stromal Tumor, Acute Myeloid Leukemia, Mast Cell Disease, Piebaldism, Pseudoxanthomatous Diffuse Cutaneous Mastocytosis, Systemic Mastocytosis With Associated Hematologic Neoplasm, Testicular Tumor	AD	100%	112 of 112
KITLG		Autosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2	AD	99.93%	10 of 10
KRAS		Aplasia Cutis Congenita With Epibulbar Dermoids, Arteriovenous Malformation Of The Brain, Bladder Cancer, Breast Cancer, Cardiofaciocutaneous Syndrome, Encephalocraniocutaneous Lipomatosis, Familial Pancreatic Carcinoma, Gastric Cancer, Acute Myeloid Leukemia, Linear Nevus Sebaceus Syndrome, Lung Cancer, Lynch Syndrome, Noonan Syndrome, Pancreatic Cancer, RAS-Associated Autoimmune Lymphoproliferative Syndrome Type 4, Schimmelpenning-Feuerstein-Mims Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100%	38 of 38
LZTR1		Noonan Syndrome, Schwannomatosis	AD	99.99%	136 of 136
MAP2K1		Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan Syndrome	AD	100%	31 of 31
MAP2K2		Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome	AD	100%	37 of 37
MAX		Hereditary Pheochromocytoma-Paraganglioma, Pheochromocytoma	AD	99.32%	33 of 33

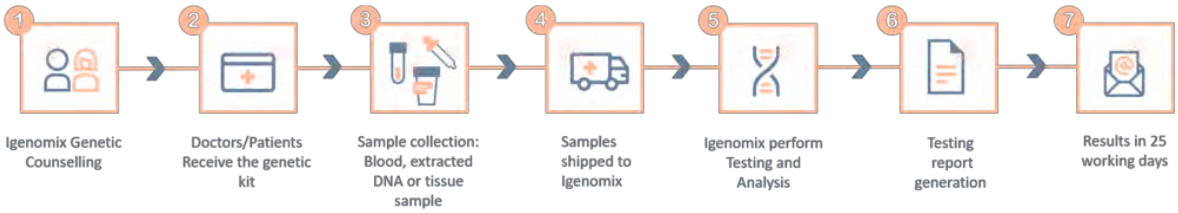
MEN1		Familial Isolated Hyperparathyroidism, Insulinoma, Multiple Endocrine Neoplasia Type 1, Pituitary Gigantism, Prolactinoma	AD	99.90%	871 of 876
MET		Autosomal Recessive Deafness, Hepatocellular Carcinoma, Osteofibrous Dysplasia, Pediatric Hepatocellular Carcinoma, Papillary Renal Cell Carcinoma	AD,AR	99.80%	41 of 41
MITF		Coloboma, Osteopetrosis, Microphthalmia, Macrocephaly, Albinism, And Deafness, Familial Melanoma, Cutaneous Malignanty Melanoma, Tietz Syndrome, Waardenburg Syndrome Type 2, Waardenburg-shah Syndrome	AD,AR	100%	72 of 72
MLH1	High	Hereditary Nonpolyposis Colorectal Cancer Type 2, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.94%	1079 of 1118
MLH3		Colorectal Cancer, Hereditary Nonpolyposis Colorectal Cancer Type 7, Endometrial Carcinoma, Lynch Syndrome	AD	99.98%	32 of 32
MRE11		Ataxia-Telangiectasia-Like Disorder, Hereditary Breast And Ovarian Cancer Syndrome	AR	99.95%	NA of NA
MSH2	High	Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.99%	1032 of 1057
MSH3		Endometrial Carcinoma, Familial Adenomatous Polyposis, MSH3-related Attenuated Familial Adenomatous Polyposis	AD,AR	99.42%	23 of 24
MSH6	High	Hereditary Nonpolyposis Colorectal Cancer Type 5, Endometrial Carcinoma, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.28%	613 of 641
MSR1		Barrett Esophagus		100%	16 of 16
MUTYH	High	Familial Adenomatous Polyposis, Gastric Cancer, MUTYH-Related Attenuated Familial Adenomatous Polyposis	AR	100%	183 of 183
MXI1		Prostate Cancer	AD	94.55%	NA of NA
NBN		Aplastic Anemia, Hereditary Breast And Ovarian Cancer Syndrome, Acute Lymphocytic Leukemia, Nijmegen Breakage Syndrome	AR,MU,P	100%	200 of 200
NF1	High	17q11.2 Microduplication Syndrome, Hereditary Pheochromocytoma-Paraganglioma, Juvenile Myelomonocytic Leukemia, Neurofibromatosis Type 1, Neurofibromatosis-Noonan Syndrome, Watson Syndrome	AD	97.97%	3082 of 3166
NF2		Familial Meningioma, Neurofibromatosis Type 2, Schwannomatosis	AD	100%	359 of 362
NRAS		Colorectal Cancer, Epidermal Nevus, Giant Pigmented Hairy Nevus, Large Congenital Melanocytic Nevus, Linear Nevus Sebaceus Syndrome, Neurocutaneous Melanosis, Noonan Syndrome, RAS-associated Autoimmune Lymphoproliferative Syndrome Type 4, Schimmelpenning-Feuerstein-Mims Syndrome, Nonmedullary Thyroid Cancer	AD	100%	15 of 15
NSD1		5q35 Microduplication Syndrome, Sotos Syndrome, Weaver Syndrome	AD	99.80%	451 of 459
NSUN2		Autosomal Recessive Non-Syndromic Intellectual Disability, Dubowitz Syndrome, Autosomal Recessive Mental Retardation	AR	99.99%	8 of 8
NTHL1		Familial Adenomatous Polyposis 3, NTHL1-Related Attenuated Familial Adenomatous Polyposis	AR	100%	13 of 13
PALB2	High	Breast Cancer, Familial Pancreatic Carcinoma, Fanconi Anemia Complementation Group N, Hereditary Breast And Ovarian Cancer Syndrome	AD,AR	98.78%	601 of 617
PAX5		Acute Lymphoblastic Leukemia, Gray Zone Lymphoma, Lymphoblastic Lymphoma, Mediastinal Gray Zone Lymphoma		100%	8 of 8
PDGFRA		Gastrointestinal Stromal Tumor, Hypereosinophilic Syndrome	AD	100%	24 of 24
PHB		Breast Cancer	AD	100%	1 of 1
PHOX2B		Congenital Failure Of Autonomic Control, Haddad Syndrome, Hirschsprung Disease-Ganglioneuroblastoma Syndrome, Neuroblastoma, Neuroblastoma With Hirschsprung Disease, Ondine Syndrome	AD	90.74%	58 of 71
PIK3CA		Breast Cancer, Capillary Malformation Of The Lower Lip, Lymphatic Malformation Of Face And Neck, Asymmetry Of Face And Limbs, And Partial/Generalized Overgrowth, Colorectal Cancer, Congenital Lipomatous Overgrowth, Vascular Malformations, And Epidermal Nevi, Cowden Syndrome, Epidermal Nevus, Gastric Cancer, Hemihyperplasia-Multiple Lipomatosis Syndrome, Hepatocellular Carcinoma, Seborrheic Keratosis, Lung Cancer, Lynch Syndrome, Suppressor Of Tumorigenicity	AD	99.58%	54 of 58
PMS1		Lynch Syndrome	AD	99.92%	32 of 33
PMS2	High	Hereditary Nonpolyposisi Colorectal Cancer Type 4, Lynch Syndrome, Mismatch Repair Cancer Syndrome	AD,AR	97.17%	264 of 285
POLD1		Colorectal Cancer, Mandibular Hypoplasia, Deafness, Progeroid Features, And Lipodystrophy Syndrome, Polymerase Proofreading-Related Adenomatous Polyposis	AD	100%	40 of 41
POLE		Colorectal Cancer, 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
POLH		Xeroderma Pigmentosum Variant Type	AR	99.49%	73 of 76
POT1		Familial Melanoma, Glioma, Cutaneous Malignant Melanoma	AD	99.76%	42 of 47
PPM1D		Breast Cancer, Intellectual Developmental Disorder With Gastrointestinal Difficulties And High Pain Threshold	AD	97.82%	76 of 79
PRF1		Aplastic Anemia, Familial Hemophagocytic Lymphohistiocytosis, Idiopathic Aplastic Anemia, Non-Hodgkin Lymphoma	AR	99.99%	196 of 196
PRKAR1A		Acrodysostosis, Acrodysostosis With Multiple Hormone Resistance, Acute Promyelocytic Leukemia, Carney Complex Type 1, Familial Atrial Myxoma, Primary Pigmented Nodular Adrenocortical Disease	AD	95.93%	165 of 171
PTCH1		Alobar Holoprosencephaly, Basal Cell Carcinoma, Basal Cell Nevus Syndrome, Gorlin Syndrome, Holoprosencephaly, Monosomy 9q22.3, Semilobar Holoprosencephaly, Septopreoptic Holoprosencephaly	AD	98.89%	498 of 502

PTEN	High	Bannayan-Riley-Ruvalcaba Syndrome, Cowden Disease, 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
PTPN11		Juvenile Myelomonocytic Leukemia, Leopard Syndrome, Metachondromatosis, Noonan Syndrome With Multiple Lentigines	AD	100%	150 of 151
RAD50		Hereditary Breast And Ovarian Cancer Syndrome, Nijmegen Breakage Syndrome-like Disorder	AR	99.94%	117 of 120
RAD51C	Moderate	Familial Breast-Ovarian Cancer, Fanconi Anemia Complementation Group O, Hereditary Breast And Ovarian Cancer Syndrome	AR	100%	130 of 130
RAD51D	Moderate	Hereditary Breast And Ovarian Cancer Syndrome		100%	97 of 97
RAD54L		Breast Cancer, Non-Hodgkin Lymphoma	AD	100%	4 of 4
RAF1		Dilated Cardiomyopathy, Leopard Syndrome, Noonan Syndrome With Multiple Lentigines	AD	100%	64 of 64
RASA2		Noonan Syndrome		99.82%	5 of 5
RB1		Bladder Cancer, Monosomy 13q14, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of The Lung	AD	99.41%	941 of 995
RB1CC1		Breast Cancer	AD	99.30%	1 of 1
RECQL		Inherited Cancer-Predisposing Syndrome, Werner Syndrome, Rothmund Thomson Syndrome Type 2, Bloom Syndrome, Baller-Gerold Syndrome		99.71%	32 of 34
RECQL4		Baller-Gerold Syndrome, Rapadilino Syndrome, Rothmund-Thomson Syndrome Type 2	AR	96.72%	134 of 135
REST		Autosomal Dominant Deafness, Hereditary Gingival Fibromatosis, Nephroblastoma, Wilms Tumor	AD	99.83%	15 of 16
RET		Congenital Failure of Autonomic Control, Congenital Failure, Haddad Syndrome, Hereditary Pheochromocytoma-Paraganglioma, Hirschsprung Disease, Multiple Endocrine Neoplasia Type 2A, 2B, Pheochromocytoma, Renal Agenesis, Bilateral, Sporadic Pheochromocytoma/Secreting Paraganglioma, Familial Medullary Thyroid Carcinoma	AD	100%	453 of 454
RHBDF2		Palmoplantar Keratoderma-Esophageal Carcinoma Syndrome, Tylosis With Esophageal Cancer	AD	99.27%	5 of 5
RINT1		Infantile Liver Failure Syndrome	AR	99.96%	16 of 16
RIT1		Noonan Syndrome	AD	99.85%	27 of 27
RNASEL		Hereditary Prostate Cancer	AD	99.83%	7 of 7
RPS20		Familial Colorectal Cancer Type X		99.97%	1 of 1
RRAS		Noonan Syndrome		95.86%	3 of 3
RUNX1		Aggressive Systemic Mastocytosis, Chronic Myeloid Leukemia, Acute Myeloid Leukemia, Familial Platelet Disorder With Associated Myeloid Malignancy	AD	99.83%	90 of 90
SAMD9		Mirage Syndrome, Familial Normophosphatemic Tumoral Calcinosis	AD,AR	99.72%	45 of 46
SAMD9L		Ataxia-Pancytopenia Syndrome, Ataxia-Pancytopenia Syndrome	AD	99.81%	39 of 39
SBDS		Aplastic Anemia, Shwachman-Diamond Syndrome	AR	100%	77 of 79
SDHA		Dilated Cardiomyopathy, Gastrointestinal Stromal Tumor, Hereditary Pheochromocytoma-Paraganglioma, Isolated Succinate-CoQ Reductase Deficiency, Leigh Syndrome, Mitochondrial Complex II Deficiency, Paragangliomas	AD,AR,MI	99.98%	103 of 103
SDHAF2		Hereditary Pheochromocytoma-Paraganglioma	AD	96.78%	8 of 8
SDHB	High	Carney-Stratakis Syndrome, Cowden Syndrome, Gastrointestinal Stromal Tumor, Hereditary Pheochromocytoma-Paraganglioma, Isolated Succinate-CoQ Reductase Deficiency, Paragangliomas, Pheochromocytoma, Sporadic Pheochromocytoma/Secreting Paraganglioma	AD	100%	261 of 264
SDHC		Carney-Stratakis Syndrome, Cowden Syndrome, Gastrointestinal Stromal Tumor, Hereditary Pheochromocytoma-Paraganglioma, Paragangliomas	AD	99.95%	62 of 63
SDHD		Carcinoid Syndrome, Carney-Stratakis Syndrome, Cowden Syndrome, Hereditary Pheochromocytoma-Paraganglioma, Isolated Succinate-CoQ Reductase Deficiency, Mitochondrial Complex II Deficiency, Paragangliomas, Pheochromocytoma	AD,AR	99.98%	164 of 166
SHOC2		Noonan Syndrome-Like Disorder With Loose Anagen Hair	AD	99.98%	8 of 8
SLC22A18		Breast Cancer, Lung Cancer, Rhabdomyosarcoma	AD,AR	99.98%	1 of 1
SLX4		Fanconi Anemia Complementation Group P	AR	99.92%	76 of 76
SMAD4	High	Familial Pancreatic Carcinoma, Familial Thoracic Aortic Aneurysm And Aortic Dissection, Generalized Juvenile Polyposis/Juvenile Polyposis Coli, Hereditary Hemorrhagic Telangiectasia, Juvenile Polyposis Syndrome, Pancreatic Cancer	AD	99.56%	136 of 136
SMARCA4		Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome	AD	100%	68 of 69
SMARCB1		Atypical Teratoid Rhabdoid Tumor, Coffin-Siris Syndrome, Meningioma, Rhabdoid Tumor Predisposition Syndrome, Shwannomatosis	AD	100%	97 of 99
SOS1		Hereditary Gingival Fibromatosis, Noonan Syndrome	AD	100%	103 of 104
SOS2		Noonan Syndrome	AD	99.48%	6 of 7
SPRED1		Legius Syndrome	AD	100%	84 of 84
SRP72		Bone Marrow Failure Syndrome	AD	99.95%	3 of 3
STK11	High	Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular Tumor	AD	81.99%	456 of 470
SUFU		Acrocallosal Syndrome, Basal Cell Nevus Syndrome, Gorlin Syndrome, Joubert Syndrome, Medulloblastoma, Familial Meningioma, Microform Holoprosencephaly	AD,AR	99.99%	43 of 43
TERC		Autosomal Dominant Dyskeratosis Congenita, Idiopathic Aplastic Anemia, Idiopathic Pulmonary Fibrosis	AD	na	na
TERT		Aplastic Anemia, Autosomal Dominant Dyskeratosis Congenita, Autosomal Recessive Dyskeratosis Congenita, Familial Melanoma, Hoyeraal-Hreidarsson Syndrome, Idiopathic	AD,AR	99.09%	194 of 197

		Aplastic Anemia, Idiopathic Pulmonary Fibrosis, Acute Myeloid Leukemia, Cutaneous Malignant Melanoma, Meningioma, Pulmonary Fibrosis And/Or Bone Marrow Failure			
TGFBR2		Hereditary Nonpolyposis Colorectal Cancer Type 6, Esophageal Cancer, Familial Thoracic Aortic Aneurysm And Aortic Dissection, Loeys-Dietz Syndrome Type 1B, Lynch Syndrome, Squamous Cell Carcinoma Of The Esophagus	AD	99.90%	165 of 166
TINF2		Autosomal Dominant Dyskeratosis Congenita, Hoyeraal-Hreidarsson Syndrome, Revesz Syndrome	AD	99.94%	47 of 47
TMEM127		Hereditary Pheochromocytoma-Paraganglioma, Pheochromocytoma	AD	99.68%	60 of 60
TP53	High	Adrenocortical Carcinoma, Basal Cell Carcinoma, Bone Marrow Failure Syndrome, Breast Cancer, Colorectal Cancer, Essential Thrombocythemia, Familial Pancreatic Carcinoma, Familial Glioma Of Brain, Hepatocellular Carcinoma, Hereditary Breast And Ovarian Cancer Syndrome, Li-Fraumeni Syndrome, Nasopharyngeal Carcinoma, Osteosarcoma, Pancreatic Cancer, Papilloma Of Choroid Plexus	AD,MU,P	98.92%	557 of 563
TSC1	High	Focal Cortical Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis Complex 1	AD	99.86%	390 of 406
TSC2	High	Focal Cortical Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis Complex 2	AD	100%	1157 of 1159
VHL	High	Familial Erythrocytosis, Hereditary Pheochromocytoma-Paraganglioma, Nonpapillary Renal Cell Carcinoma, Von Hippel-Lindau Syndrome	AD,AR	100%	511 of 544
WRN		Werner Syndrome	AR	99.97%	107 of 109
WT1		46,XY Complete Gonadal Dysgenesis, 46,XY Partial Gonadal Dysgenesis, Aniridia, Denys-Drash Syndrome, Desmoplastic Small Round Cell Tumor, Frasier Syndrome, Meacham Syndrome, Mesothelioma, Malignant, Nephroblastoma, Nephrotic Syndrome, WAGR Syndrome	AD	98.92%	178 of 185
XPA		Xeroderma Pigmentosum Complementation Group A	AR	99.91%	49 of 49
XPC		Xeroderma Pigmentosum Complementation Group C	AR	99.83%	86 of 87
XRCC2		Fanconi Anemia, Complementation Group U, Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation	AR	98.39%	28 of 28
XRCC3		Breast Cancer	AD	100%	11 of 11

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

Methodology



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