



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 ANKRD26		Neuroblastoma Thrombocytopenia	AD	99.84% 98.76%	16 of 16 3 of 23
АРС	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
ΑΤΜ	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 CDKN1C		Multiple Endocrine Neoplasia Type 1, Multiple Endocrine Neoplasia Type 4 Beckwith-Wiedemann Syndrome, Image Syndrome, Intrauterine Growth Restriction-Short Stature-Early Adult-Onset Diabetes Syndrome, Intrauterine Growth Retardation,	AD AD	99.99% 73.58%	19 of 20 55 of 76
CDKN2A	High	Metaphyseal Dysplasia, Adrenal Hypoplasia Congenita And Genital Anomalies Familial Melanoma, Familial Pancreatic Carcinoma, Li-Fraumeni Syndrome, Melanoma- Astrocytoma Syndrome, Melanoma-Pancreatic Cancer Syndrome, Cutaneous Malignant	40	04.00%	257 of
CDKN2A	High	Melanoma	AD	94.99%	262
CEBPA CEP57		Acute Myeloid Leukemia Mosaic Variegated Aneuploidy Syndrome	AD AR	67.47% 99.64%	14 of 17 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 Familial Multinodular Goiter, Global Developmental Delay, Lung Cysts, Overgrowth, And	AD	99.99%	56 of 56
DICER1		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 Neonatal Inflammatory Skin And Bowel Disease, Lung Cancer, Neonatal Inflammatory Skin	AR	99.94%	NA of NA
EGFR		And Bowel Disease	AD,AR	100%	27 of 27
ELAC2		Combined Oxidative Phosphorylation Deficiency Autosomal Dominant Severe Congenital Neutropenia, Cyclic Hematopoiesis, Cyclic	AR	100%	32 of 32 227 of
ELANE		Neutropenia, Autosomal Dominant Severe Congenital Neutropenia, Cyclic Hernatopolesis, Cyclic Neutropenia, Autosomal Dominant Severe Congenital Neutropenia	AD	100%	227 01





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FANCMInstruct Spermatogenic FailureOligozoospermia, Premature Ovarian Failure, Spermatogenic FailureSpe of 61HighHighEnarrase Deficiency, Hereditary Leiomyomatosis And Renal Cell Cancer, Hereditary Phocothrucytome-Paraganglioma, LeiomyomaDD, AR100% 2322229 of 						
FAILURFailure, Spermatogenic FailureFAILURSpermatogenic FailureSpermatogenic FailureSpermatogen	FANCL			AK	100%	25 OF 26
Image 	FANCM			AR	99.73%	59 of 61
FLCNHighPhecknomocytoma-Paraganginoma, LeomyonaAD100%200GALMT12Coloretal Cancer, Familia Spontaneous Pneumothorax, Potock-Lupski Syndrome, Nonpapillary Renal Cell CarcinomaAD100%200GALMT12Coloretal Cancer, Familia Coloretal Cancer Type 1088.97%14 of 15GATA2Deafness-Lymphedema-Leukemia Syndrome, Dendritic Cell, Monocyte, B Lymphocyte, And Natural Killer Lymphocyte Deficiency, Acute Myeloid Leukemia, Primary Lymphedema, Syndrome Type 1, Wilms TumorAD100%142GEN1Xeroderma Pigmentosum Complementation Group G99.71%6 of 66GR23Nephoblastoma, Simpson-Golabi-Behmel Syndrome Type 1, Wilms TumorAD,X,XR,G99.84%NA of NAGREM1Hereditary Mixed Polyposis Syndrome100%5 of 515HNF1AHereditary Mixed Polyposis Syndrome100%5 of 5HNS13Hereditary Prostal Cancer, Costello Syndrome, Epidermal Nevus, Linear Nevus Sebaceus Syndrome, Sonfamelpennie, Feuerstein-Mins Syndrome, Nonmedullary Thyroid CancerAD100%34 of 34IKZE1Common Variable Immunodeficiency, Stevens-Johnson SyndromeAD99.89%10 of 10KITLGCommon Variable Immunodeficiency, Scutaneous Mastocytosis, Syndrome, Peudoramona, Mixopapillary Ependymona, AD99.89%10 of 10Bullous Diffuse Cutaneous Mastocytosis, Systemic Mastocytosis Syndrome, Syndrome Type 2, Hereditary Prostacese Syndrome, Syndrome Type 4, Hereditary Prostacese Syndrome, Syndrome Type 4, Schamelpenning, Feuerstein-Mins Syndrome, Reset. Peakadian, Reset. Spinalia Prostacese Syndrome, Nonpapillary Renat Cell Carcinom	FH	High	Fumarase Deficiency, Hereditary Leiomyomatosis And Renal Cell Cancer, Hereditary		100%	
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GALNT12Colorectal Cancer, Familial Colorectal Cancer, Type 1088.97%14 of 15GATA2Deafness-Lymphedema-Leukemia Syndrome, Dendritic Cell, Monocyte, B Lymphedema, Primary, With Myelodysplasti, SyndromeAD100%137 of 142GEN1Veroderma Pigmentssun Complementation Group G99.71%6 of 6GPC3Nephroblastoma, Simpson-Golabi-Behmel Syndrome99.81%NA of NAGREM1Hereditary Mixed Polyposis Syndrome99.81%NA of NAGREM1Hereditary Mixed Polyposis Syndrome99.81%S of 5HNF1AHereditary Prostate Cancer, Spinal Cord Ependymoma, Myxopapillary Ependymoma100%5 of 5HAK5Schirmelpenning-Feuerstein-Mins Syndrome, Nonmedullary Thyroid CancerAD100%34 of 34KI71Common Variable Immundeficiency, Stevens-Johnson SyndromeAD99.89%13 of 11KI71Common Variable Immundeficiency, Stevens-Johnson Syndrome, AD99.89%13 of 11KI716Autosomal Dominant Deafness, Familial Progressive Hyperpigmentation, WaardenburgAD99.93%10 of 10Syndrome Type 2Autosomal Dominant Deafness, Familial Progressive Hyperpigmentation, WaardenburgAD99.93%38 of 38KI716Noonan Syndrome, SchwannomatosisAD99.93%10 of 1038 of 38KI716Noonan Syndrome, SchwannomatosisAD99.93%10 of 1038 of 38KI716Noonan Syndrome, SchwannomatosisAD99.99%38 of 38KI716Noonan Syndrome, SchwannomatosisAD99.99%38 of 38<	FLCN	High		AD	100%	
GATA2Deafness-lymphedema-leukemia Syndrome, Dendritic Cell, Monocyte, B Lymphocyte, And Natural Killer Lymphocyte Deficiency, Acute Myeloid Leukemia, Primary Lymphedema, Primary, With Myelodysplasia, Myelodysplasic Syndrome Syndrome Type 1, Wilms TumorAD100%137 of 142GEN1Keroderma Pigmentosum Complementation Group G99.71%6 of 6 of 6 of 6GPC3Hereditary Mixed Polyposis Syndrome Type 1, Wilms TumorADX,XR,G99.84%NA of NA 99.89%5 of 5HMF1AHereditary Mixed Polyposis Syndrome Insulin-Dependent Diabetes Mellitus, Familial Peptic Adenomas, Hyperinsulinism, Maturity-Onset Diabetes Of The Young Type 3, Nonpapillary Renal Cell CarcinomaAD100%\$29 of \$38HOXB13Hereditary Prostate Cancer, Spinal Cord Ependymoma, Myxopapillary EpendymomaAD100%\$4 of 34KRA5Bladder Cancer, Costello Syndrome, Epidermal Nevus, Linear Nevus Sebaceus Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Nonmedullary Thyroid CancerAD99.99%3 of 41KKF18Charcot-Marie-Tooth Disease Type 2A1, Hereditary Pheochromocytoma-Paraganglioma, Neuroblastoma, PheochromocytomaAD99.99%10 of 112KKT7Autosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 112KKT8Aplasia Cuttareous Mastocytosis, Systemic Mastocytosis With Associated Hematologic Neoplasm, Testicular TumorAD99.93%10 of 112KKT6Autosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%3 of 31KKT6Noona	GALNT12				88.97%	
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GENIExerciderma Pigmentosum Complementation Group G99.71%6 of 6GF/3Nephroblastoma, Simpson-Golabi-Behmel Syndrome Type 1, Wilms TumorAD,X,XR,G99.84%NA of NAGREM1Hereditary Mixed Polypois Syndrome99.89%S of 5HNF1AHisulin-Dependent Diabetes Mellitus, Noninsulin-Dependent Diabetes Mellitus, Familial99.89%S of 5HNF1AHepatic Adenomas, Hyperinsulinism, Maturity-Onset Diabetes Of The Young Type 3, Nonpapillary Renal Cell Carcinoma100%S of 5HOXB13Hereditary Postate Cancer, Spinal Cord Ependymoma, Myxopapillary Ependymoma100%S of 5HRA5Bladder Cancer, Costello Syndrome, Epidermal Nevus, Linear Nevus Sebaceus Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Nonmedullary Thyroid CancerAD100%34 of 34 <i>IKZF1</i> Common Variable Immunodeficiency, Stevens-Johnson SyndromeAD99.89%17 of 17Neuroblastoma, PheochromocytomaBullous Diffuse Cutaneous Mastocytosis, Cutaneous Mastocytosis, Gastrointestinal Stromal Tumor, Acute Myeloid Leukemia, Nast Cell Disease, Piebaldism, Seudoxantomatous Diffuse Cutaneous Mastocytosis, Systemic Mastocytosis With Associated HematologicAD99.93%10 of 10 <i>KITLG</i> Aplasia Cutis Congenita With Epibulbar Dermoids, Arteriovenous Malformation Of The Brain, Bladder Cancer, Parea Cancer, Carcindaciocutaneous Syndrome, Noonan Syndrome, Parea Syndrome, Toriello-Lacassie-Droste Syndrome Type 2AD99.99%136 of <i>KITLG</i> Noonan Syndrome, None Autounne Lymphoproliferative Syndrome Type 4, Schimmelpenning-Feuerstein-Mims Syndrome, Toriello-Lacassie-Droste SyndromeAD <td< td=""><td>GATA2</td><td></td><td></td><td>AD</td><td>100%</td><td></td></td<>	GATA2			AD	100%	
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GREM1Hereditary Mixed Polyposis Syndrome99.89%5 of 5HN/LAHereditary Mixed Polyposis Syndrome100%529 of 1538HN/EIAHepatic Adenomas, Hyperinsulinism, Maturity-Onset Diabetes Of The Young Type 3, Nonpapillary Renal Cell CarcinomaAD100%529 of 538HOXB13Hereditary Prostate Cancer, Spinal Cord Ependymoma, Myxopapillary Ependymoma100%5 of 5HRASBladder Cancer, Costell O Syndrome, Epidermal Nevus, Linear Nevus Sebaceus Syndrome, Schimmelpenning-Feuerstein-Mins Syndrome, Nonmedullary Thyroid CancerAD99.98%43 of 34 <i>IKZF1</i> Common Variable Immunodeficiency, Stevens-Johnson SyndromeAD99.89%17 of 17 <i>KIF1B</i> Charcot-Marie-Tooth Disease Type 2A1, Hereditary Pheochromocytoma-Paraganglioma, Neuroblastoma, PheochromocytomaAD99.89%112 of 112 <i>KIT1G</i> Syndrome Type 2 Acute Myeloid Leukemia, Mast Cell Disease, Piebaldism, Pseudoxanthomatous Diffuse Cutaneous Mastocytosis, Systemic Mastocytosis With Associated Hematologic Neoplasm, Testicular Tumor Acute Myeloid Leukemia, Adst Cell Disease, Piebaldism, Pseudoxanthomatous Neoplasm, Testicular Tumor Acute Myeloid Leukemia, Adst Cell Disease, Piebaldism, Pseudoxanthomatous Neoplasm, Testicular Tumor Acute Myeloid Leukemia, Adst Cell Disease, Piebaldism, Pseudoxanthomatous Neoplasm, Testicular Tumor Acute Myeloid Leukemia, Adst Cell Disease, Piebaldism, Pseudoxanthomatous Neoplasm, Testicular Tumor Acute Myeloid Leukemia, Kats Cancer, RAS-Associated Autoimmune Lymphoproliferative Syndrome Type 2AD99.93%10 of 10 <i>KITLG</i> Aplasia Cutis Congenita With Epibulibar Dermoids, Arteriovenous Malformation Of Th			•	AD,X,XR,G		
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HOXB13Hereditary Prostate Cancer, Spinal Cord Ependymoma, Myxopapillary Ependymoma100%5 of 5HRASBladder Cancer, Costello Syndrome, Epidermal Nevus, Linear Nevus Sebaceus Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Nonmedullary Thyroid CancerAD100%34 of 34 <i>IKZF1</i> Common Variable Immunodeficiency, Stevens-Johnson SyndromeAD99.98%43 of 43 <i>KIF1B</i> Charcot-Marie-Tooth Disease Type 2A1, Hereditary Pheochromocytoma-Paraganglioma, Neuroblastoma, PheochromocytomaAD99.89%17 of 17 <i>KIT</i> Bullous Diffuse Cutaneous Mastocytosis, Cutaneous Mastocytosis, Cutaneous Mastocytosis, Cutaneous Mastocytosis With Associated Hematologic Neoplasm, Testicular TumorAD99.93%10 of 10 <i>KITLG</i> Autosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 10 <i>KRAS</i> Aplasia Cutis Congenita With Epibulbar Dermoids, Arteriovenous Malformation Of The Brain, Bladder Cancer, Reast Cancer, Cardiofaciocutaneous Syndrome, Encephalocranicocutaneous Lipomatosis, Familial Progressive Hyperpigmentation, Sastric Cancer, Acute Myeloid Leukemia, Linear Nevus Sebaceus Syndrome, Lung Cancer, Lynch Syndrome, Noonan Syndrome, Pancreatic Cancer, RAS-Associated Autoimmune Lymphoproliferative SyndromeAD100%38 of 38 <i>MAP2KI</i> Cardiofaciocutaneous Syndrome, Neurofibromatosis, Noonan Syndrome, AD99.99%136 of 136 <i>MAP2KI</i> Cardiofaciocutaneous Syndrome, Neurofibromatosis, Noonan Syndrome, AD100%31 of 31 <i>MAP2KI</i> Cardiofaciocutaneous Syndrome, Neurofibromatosis, Noonan Syndrome AD100%31 of 31	HNF1A			AD	100%	
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IKZF1Comme Jean Syndrome, Normed Ulary Inyroid CancerAD99.98%43 of 43KIF1BCommo Variable Immunodeficiency, Stevens-Johnson SyndromeAD99.98%43 of 43KIF1BCharcot-Marie-Tooth Disease Type 2A1, Hereditary Pheochromocytoma-Paraganglioma, Neuroblastoma, PheochromocytomaAD99.89%17 of 17KIT1Buillous 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 TumorAD99.93%100%112 of 112KITLGAutosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 10KRASAplasia 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 SyndromeAD99.99%136 of 136ILZTR1Coman Syndrome, SchwannomatosisNoonan Syndrome, Melorheostosis, Noonan Syndrome ADAD100%31 of 31MAP2K1Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome ADAD100%31 of 31						
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KIF1BNeuroblastoma, PheochromocytomaAD99.89%17 of 17KITBullous Diffuse Cutaneous Mastocytosis, Cutaneous Mastocytoma, Gastrointestinal Stromal Tumor, Acute Myeloid Leukemia, Mast Cell Disease, Piebaldism, Pseudoxanthomatous Diffuse Cutaneous Mastocytosis, Systemic Mastocytosis With Associated HematologicAD100%112 of 112KITLGAutosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 10KITLGAutosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 10KRASAutosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 10KRASAutosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 10KRASAutosomal 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 SyndromeAD99.99%136 of 136MAP2K1Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan Syndrome Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome AD100%31 of 31	IKZF1			AD	99.98%	43 of 43
KITBullous 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 TumorAD100%112 of 112KITLGAutosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2 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 SyndromeAD99.99%136 of 136MAP2K1Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan Syndrome Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome ADAD90.90%31 of 31MAP2K2Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome ADAD100%31 of 31	KIF1B			AD	99.89%	17 of 17
KITDiffuse Cutaneous Mastocytosis, Systemic Mastocytosis With Associated Hematologic Neoplasm, Testicular TumorAD100%112KITLGAutosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 10KITLGAplasia 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 SyndromeAD99.99%136 of 136ILZTR1Noonan Syndrome, SchwannomatosisAD99.99%136 of 136136MAP2K1Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan SyndromeAD100%31 of 31MAP2K2Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan SyndromeAD100%37 of 37			. ,			
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KITLGAutosomal Dominant Deafness, Familial Progressive Hyperpigmentation, Waardenburg Syndrome Type 2AD99.93%10 of 10KRASAplasia 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 SyndromeAD100%38 of 38LZTR1Noonan Syndrome, SchwannomatosisAD99.99%136 of 136MAP2K1Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan SyndromeAD100%31 of 31MAP2K2Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan SyndromeAD100%37 of 37						112
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KRASEncephalocraniocutaneous 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 SyndromeAD100%38 of 38LZTR1Noonan Syndrome, SchwannomatosisAD99.99%136 of 136MAP2K1 MAP2K2Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan SyndromeAD100%31 of 31						
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Syndrome Syndrome 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	1000			, ib	100/0	50 01 50
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MAP2K1Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan SyndromeAD100%31 of 31MAP2K2Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan SyndromeAD100%37 of 37	LZTR1		Noonan Syndrome, Schwannomatosis	AD	99.99%	
MAP2K2 Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome AD 100% 37 of 37	MAP2K1		Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan Syndrome	AD	100%	
MAX Hereditary Pheochromocytoma-Paraganglioma, Pheochromocytoma AD 99.32% 33 of 33	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
ΜΕΤ		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	AD	99.98%	32 of 32
MRE11		Carcinoma, Lynch Syndrome 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
МЅНЗ		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 Familial Adenomatous Polyposis, Gastric Cancer, MUTYH-Related Attenuated Familial		100%	16 of 16 183 of
MUTYH	High	Adenomatous Polyposis	AR	100%	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 17q11.2 Microduplication Syndrome, Hereditary Pheochromocytoma-Paraganglioma,	AR,MU,P	100%	200 of 200
NF1	High	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 PHB		Gastrointestinal Stromal Tumor, Hypereosinophilic Syndrome Breast Cancer	AD AD	100% 100%	24 of 24
PND		Congenital Failure Of Autonomic Control, Haddad Syndrome, Hirschsprung Disease-	AD	100%	1 of 1
РНОХ2В		Ganglioneuroblastoma Syndrome, Neuroblastoma, Neuroblastoma With Hirschsprung Disease, Ondine Syndrome	AD	90.74%	58 of 71
РІКЗСА		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 PPM1D		Familial Melanoma, Glioma, Cutaneous Malignant Melanoma Breast Cancer, Intellectual Developmental Disorder With Gastrointestinal Difficulties And	AD AD	99.76% 97.82%	42 of 47 76 of 79
PRF1		High Pain Threshold Aplastic Anemia, Familial Hemophagocytic Lymphohistiocytosis, Idiopathic Aplastic Anemia, Non Hodkin Lymphoma	AR	99.99%	196 of
		Non-Hodgkin Lymphoma Acrodysostosis, Acrodysostosis With Multiple Hormone Resistance, Acute Promyelocytic			196
PRKAR1A		Leukemia, Carney Complex Type 1, Familial Atrial Myxoma, Primary Pigmented Nodular Adrenocortical Disease	AD	95.93%	165 of 171
РТСН1		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





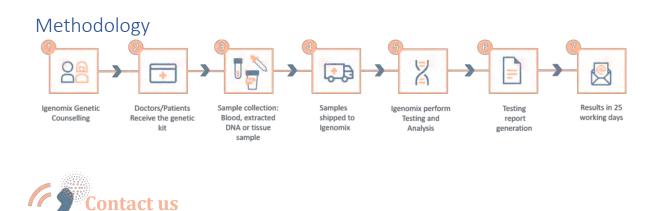
PTEN	High	Bannayan-Riley-Ruvalcaba Syndrome, Cowden Disease, Hereditary Breast And Ovarian Cancer Syndrome, Juvenile Polyposis Of Infancy, Lhermitte-Duclos Disease, Macrocephaly/Autism Syndrome, Familial Meningioma, Prostate Cancer, Proteus Syndrome, Segmental Outgrowth-Lipomatosis-Arteriovenous Malformation-Epidermal	AD	99.97%	609 of 629
		Nevus Syndrome			
PTPN11		Juvenile Myelomonocytic Leukemia, Leopard Syndrome, Metachondromatosis, Noonan	AD	100%	150 of
		Syndrome With Multiple Lentigines	, 10	100/0	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
		Congenital Failure of Autonomic Control, Congenital Failure, Haddad Syndrome, Hereditary			
RET		Pheochromocytoma-Paraganglioma, Hirschsprung Disease, Multiple Endocrine Nerolasia 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
		Dilated Cardiomyopathy, Gastrointestinal Stromal Tumor, Hereditary Pheochromocytoma-			103 of
SDHA		Paraganglioma, Isolated Succinate-CoQ Reductase Deficiency, Leigh Syndrome,	AD,AR,MI	99.98%	103 01
		Mitochondrial Complex II Deficiency, Paragangliomas			105
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,	AD,AR	99.98%	164 of
		Mitochondrial Complex II Deficiency, Paragangliomas, Pheochromocytoma			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	AD	99.56%	136 of 136
SMARCA4		Telangiectasia, Juvenile Polyposis Syndrome, Pancreatic Cancer 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	AD,AR	99.09%	194 of
		Dyskeratosis Congenita, Familial Melanoma, Hoyeraal-Hreidarsson Syndrome, Idiopathic			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
ХРС		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



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