Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HEREDITARY CANCER PANEL DG-5.1.0 (251 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
ACD	100%	100%	100%	99.9%	99.3%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
AIP	100%	99.5%	100%	100%	99.8%	Pituitary adenoma 1, multiple types, 102200;Pituitary adenoma predisposition, 102200
AKT1	100%	100%	100%	100%	99.1%	Proteus syndrome, somatic mosaic, 176920;Breast cancer, somatic, 114480;Cowden syndrome 6, 615109;Colorectal cancer, somatic, 114500;Ovarian cancer, somatic, 167000
ALK	100%	100%	100%	100%	99.4%	{Neuroblastoma, susceptibility to, 3}, 613014
AMH	100%	100%	100%	99.9%	99%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100%	100%	100%	100%	99.4%	Persistent Mullerian duct syndrome, type II, 261550
ANKRD26	100%	100%	100%	100%	99.7%	Thrombocytopenia 2, 188000

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

APC	100%	100%	100%	100%	99.8%	Colorectal cancer, somatic, 114500;Brain tumor-polyposis syndrome 2, 175100;Desmoid disease, hereditary, 135290;Adenoma, periampullary, somatic, 175100;Hepatoblastoma, somatic, 114550;Gastric cancer, somatic, 613659;Gastric adenocarcinoma and proximal polyposis of the stomach, 619182;Gardner syndrome, 175100;Adenomatous polyposis coli, 175100
ARMC5	100%	100%	100%	100%	99.4%	{ACTH-independent macronodular adrenal hyperplasia 2}, 615954
ASXL1	100%	100%	100%	100%	99.8%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ATM	100%	100%	100%	100%	99.7%	Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic
ATR	100%	100%	99.9%	99.7%	99.2%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	100%	100%	100%	100%	99.2%	Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BAP1	100%	100%	100%	100%	99.5%	Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661
BARD1	100%	100%	100%	100%	99.7%	{Breast cancer, susceptibility to}, 114480
BLM	94.7%	94.6%	100%	100%	99.9%	Bloom syndrome, 210900
BMPR1A	96.5%	96.5%	100%	100%	99.8%	Polyposis syndrome, hereditary mixed, 2, 610069;Polyposis, juvenile intestinal, 174900
BRAF	100%	100%	100%	99.8%	98.2%	Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980
BRCA1	100%	100%	100%	100%	99.8%	Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BRCA2	100%	100%	100%	100%	99.8%	Fanconi anemia, complementation group D1, 605724;{Glioblastoma 3}, 613029;{Medulloblastoma}, 155255;{Prostate cancer}, 176807;{Breast-ovarian cancer, familial, 2}, 612555;{Breast cancer, male, susceptibility to}, 114480;{Pancreatic cancer 2}, 613347;Wilms tumor, 194070
BRIP1	96%	96%	100%	100%	99.8%	Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480
BUB1	97.3%	97.3%	100%	100%	99.7%	Colorectal cancer with chromosomal instability, somatic, 114500;Microcephaly 30, primary, autosomal recessive, 620183
BUB1B	96.3%	96.3%	100%	100%	99.8%	Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	100%	100%	100%	100%	99.6%	
CARD11	100%	100%	100%	100%	99.5%	B-cell expansion with NFKB and T-cell anergy, 616452;Immunodeficiency 11B with atopic dermatitis, 617638;Immunodeficiency 11A, 615206

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CBL	100%	100%	100%	100%	99.7%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CD27	100%	100%	100%	100%	99.7%	Lymphoproliferative syndrome 2, 615122
CD70	100%	100%	100%	100%	99.1%	Lymphoproliferative syndrome 3, 618261
CDC73	100%	100%	100%	99.9%	99.4%	Hyperparathyroidism, familial primary, 145000;Parathyroid adenoma with cystic changes, 145001;Parathyroid carcinoma, 608266;Hyperparathyroidism-jaw tumor syndrome, 145001
CDH1	100%	100%	100%	100%	99.6%	Ovarian cancer, somatic, 167000;Blepharocheilodontic syndrome 1, 119580;Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215;Endometrial carcinoma, somatic, 608089;Breast cancer, lobular, somatic, 114480
CDH23	100%	100%	100%	100%	99.4%	Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CDK4	100%	100%	100%	100%	99.6%	{Melanoma, cutaneous malignant, 3}, 609048;Microcephaly 31, primary, autosomal recessive, 621507
CDKN1A	100%	100%	100%	100%	99.8%	
CDKN1B	100%	100%	100%	100%	99.6%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	100%	100%	100%	100%	99%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CDKN2A	100%	100%	100%	99.8%	98.5%	{Melanoma and neural system tumor syndrome}, 155755;{Melanoma, cutaneous malignant, 2}, 155601;{Melanoma-pancreatic cancer syndrome}, 606719
CDKN2B	100%	100%	100%	100%	99.5%	
CDKN2C	100%	100%	100%	100%	98.7%	
CEBPA	100%	100%	99.9%	98.8%	92.6%	Leukemia, acute myeloid, somatic, 601626;?Leukemia, acute myeloid, 601626
CHEK2	100%	100%	100%	100%	99.6%	Prostate cancer, somatic, 176807;Osteosarcoma, somatic, 259500;Tumor predisposition syndrome 4, breast/prostate/colorectal, 609265
CREBBP	100%	100%	100%	99.9%	99.5%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CTC1	100%	100%	100%	100%	99.4%	Cerebroretinal microangiopathy with calcifications and cysts, 612199

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CTLA4	93.2%	93.2%	100%	100%	100%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CTNNA1	94.8%	94.8%	100%	100%	99.8%	Macular dystrophy, patterned, 2, 608970
CTR9	100%	100%	100%	100%	99.6%	
CYLD	100%	100%	100%	100%	99.9%	Brooke-Spiegler syndrome, 605041;Cylindromatosis, familial, 132700;Trichoepithelioma, multiple familial, 1, 601606;?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132
DCLRE1B	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal recessive 8, 620133
DDB2	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	100%	100%	100%	100%	99.5%	Warsaw breakage syndrome, 613398
DDX41	100%	100%	100%	100%	99.2%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DGCR8	95.6%	95.6%	100%	100%	99.3%	
DICER1	100%	100%	100%	100%	99.8%	Pleuropulmonary blastoma, 601200;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800;GLOW syndrome, somatic mosaic, 618272;Rhabdomyosarcoma, embryonal, 2, 180295

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DIS3L2	100%	100%	100%	99.9%	99.5%	Perlman syndrome, 267000
DKC1	100%	100%	99%	89.5%	70.6%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLST	100%	100%	100%	99.9%	99.5%	Pheochromocytoma/paraganglioma syndrome 7, 618475
DNAJC21	94.7%	94.7%	100%	100%	99.3%	Bone marrow failure syndrome 3, 617052
DUT	100%	100%	100%	99.8%	99%	Bone marrow failure and diabetes mellitus syndrome, 620044
EGFR	100%	100%	100%	100%	99.7%	Neonatal nephrocutaneous inflammatory syndrome, 616069;Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980;Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980;{Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	88%	88%	100%	99.9%	98.5%	Erythrocytosis, familial, 3, 609820;[Hemoglobin, high altitude adaptation], 609070
EGLN2	100%	100%	100%	100%	99.5%	
ELANE	100%	100%	100%	99.9%	99%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700
ELP1	100%	100%	100%	100%	99.8%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EPAS1	100%	100%	100%	100%	99.6%	Erythrocytosis, familial, 4, 611783
EPCAM	100%	100%	100%	100%	99.3%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
ERCC1	100%	100%	100%	99.9%	99.2%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	96.3%	94.6%	100%	100%	99.3%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100%	100%	100%	100%	99.7%	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651
ERCC4	100%	100%	100%	100%	99.6%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERCC5	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ERCC6	100%	100%	100%	100%	99.6%	UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980
ESR2	100%	100%	100%	100%	99.8%	?Ovarian dysgenesis 8, 618187
ETV6	100%	100%	100%	100%	99.3%	Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626
EXT1	100%	100%	100%	100%	99.5%	Exostoses, multiple, type 1, 133700;Chondrosarcoma, 215300
EXT2	100%	100%	100%	100%	99.6%	Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701
EZH2	100%	100%	100%	100%	99.7%	Weaver syndrome, 277590
FAAP100	100%	100%	100%	100%	99.6%	Fanconi anemia, complementation group X, 621258
FANCA	100%	100%	100%	100%	99.5%	Fanconi anemia, complementation group A, 227650
FANCB	96.2%	96.2%	99.4%	92.2%	74.7%	Fanconi anemia, complementation group B, 300514

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FANCC	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group C, 227645
FANCD2	100%	100%	100%	100%	99.7%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	100%	100%	99.3%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	100%	99.2%	Fanconi anemia, complementation group F, 603467
FANCG	100%	99.5%	100%	100%	99.3%	Fanconi anemia, complementation group G, 614082
FANCI	100%	99.9%	100%	100%	99.6%	Fanconi anemia, complementation group I, 609053
FANCL	91.5%	89.3%	100%	100%	99.1%	Fanconi anemia, complementation group L, 614083
FANCM	100%	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FAS	100%	100%	100%	100%	99.6%	Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	100%	100%	100%	99.9%	99.6%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980
FBXW7	100%	100%	100%	100%	99.6%	Developmental delay, hypotonia, and impaired language, 620012

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FH	100%	100%	100%	100%	99.8%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FLCN	96.9%	96.9%	100%	100%	99.7%	Birt-Hogg-Dube syndrome, 135150;Colorectal cancer, somatic, 114500;Pneumothorax, primary spontaneous, 173600;Renal carcinoma, chromophobe, somatic, 144700
G6PC3	96.8%	96.8%	100%	100%	99.8%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541
GALNT12	90.6%	90.6%	100%	100%	99.4%	{Colorectal cancer, susceptibility to, 1}, 608812
GATA2	85.7%	85.7%	100%	100%	99.1%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286
GDNF	100%	100%	100%	100%	99.3%	{Hirschsprung disease, susceptibility to, 3}, 613711
GFI1	100%	100%	100%	100%	99.2%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107
GPC3	94%	94%	98.6%	88.7%	70.8%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GPR161	100%	100%	100%	100%	99.5%	{Medulloblastoma predisposition syndrome}, 155255
GREM1	100%	100%	100%	100%	99.7%	
GRHL2	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100%	100%	100%	100%	99.3%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	100%	100%	99.7%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HOXB13	100%	100%	100%	100%	99.4%	{Prostate cancer, hereditary, 9}, 610997
IDH1	100%	100%	100%	99.9%	99.6%	{Glioma, susceptibility to, somatic}, 137800
IDH2	89.7%	89.5%	100%	100%	99.4%	D-2-hydroxyglutaric aciduria 2, 613657
IKZF1	100%	100%	100%	100%	99.5%	Immunodeficiency, common variable, 13, 616873
IPMK	100%	100%	100%	99.9%	99.4%	
ITK	100%	100%	100%	100%	99.6%	Lymphoproliferative syndrome 1, 613011
KIF1B	94.9%	94.9%	100%	100%	99.6%	{Neuroblastoma, susceptibility to, 1}, 256700;Charcot-Marie-Tooth disease, type 2A1, 118210

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KIT	100%	100%	100%	100%	99.8%	Gastrointestinal stromal tumor, familial, 606764;Mastocytosis, cutaneous, 154800;Piebaldism, 172800;Germ cell tumors, somatic, 273300;Mastocytosis, systemic, somatic, 154800;Leukemia, acute myeloid, somatic, 601626
KRAS	100%	100%	100%	100%	100%	Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;Bladder cancer, somatic, 109800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LHCGR	100%	100%	100%	100%	99.7%	Leydig cell adenoma, somatic, with precocious puberty, 176410;Leydig cell hypoplasia with pseudohermaphroditism, 238320;Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320;Luteinizing hormone resistance, female, 238320;Precocious puberty, male, 176410
LIG4	100%	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LZTR1	100%	100%	100%	100%	99.6%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MAD2L2	100%	100%	100%	99.9%	99.1%	?Fanconi anemia, complementation group V, 617243
MAP2K1	95.8%	95.8%	100%	100%	99.5%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	92.4%	92.3%	100%	100%	99.5%	Cardiofaciocutaneous syndrome 4, 615280
MAX	100%	100%	100%	100%	99.7%	Polydactyly-macrocephaly syndrome, 620712;{Pheochromocytoma, susceptibility to}, 171300
MBD4	100%	100%	100%	100%	99.9%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975
MCM8	94.4%	94.4%	100%	100%	99.8%	?Premature ovarian failure 10, 612885

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MCM9	100%	100%	100%	100%	99.8%	Ovarian dysgenesis 4, 616185
MDH2	100%	100%	100%	100%	98.7%	Developmental and epileptic encephalopathy 51, 617339
MEN1	100%	100%	100%	100%	99.1%	Lipoma, somatic;Angiofibroma, somatic;Multiple endocrine neoplasia 1, 131100;Carcinoid tumor of lung;Adrenal adenoma, somatic;Parathyroid adenoma, somatic
MET	100%	100%	100%	100%	99.8%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074;?Arthrogryposis, distal, type 11, 620019;Hepatocellular carcinoma, childhood type, somatic, 114550;{Osteofibrous dysplasia, susceptibility to}, 607278;?Deafness, autosomal recessive 97, 616705
MITF	94.7%	94.7%	100%	100%	99.9%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MLH1	100%	100%	100%	100%	99.7%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MLH3	100%	100%	100%	99.9%	99.2%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MPL	100%	100%	100%	99.9%	99.3%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977
MRE11	97.4%	97.4%	100%	100%	99.9%	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	90.6%	90.6%	100%	100%	99.6%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH3	98%	98%	100%	100%	99.7%	Familial adenomatous polyposis 4, 617100;Endometrial carcinoma, somatic, 608089
MSH6	100%	100%	100%	100%	99.5%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MTAP	91.5%	91.5%	100%	100%	99.4%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MUTYH	100%	100%	100%	99.9%	99.5%	Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NAF1	100%	100%	100%	100%	99.5%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NBN	97.5%	97.5%	100%	100%	99.8%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NF1	99.4%	99.4%	100%	100%	99.8%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NF2	100%	100%	100%	100%	99.5%	Meningioma, NF2-related, somatic, 607174;Schwannomatosis, vestibular, 101000;Schwannomatosis, somatic, 101000
NHP2	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	92.5%	92.5%	100%	100%	99.9%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	87.6%	87.6%	100%	99.9%	99.3%	Leukemia, acute myeloid, somatic, 601626

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NRAS	89.4%	89.4%	100%	100%	99.9%	Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
NTHL1	100%	100%	100%	99.9%	99.1%	Familial adenomatous polyposis 3, 616415
NYNRIN	100%	100%	100%	100%	99.4%	
PALB2	100%	100%	100%	100%	99.7%	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832
PARN	97.2%	96%	100%	100%	99.8%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PAX5	100%	100%	100%	99.9%	99.5%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PDGFB	99.7%	94.7%	100%	100%	99.8%	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483
PDGFRA	100%	100%	100%	100%	99.6%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510;Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PHOX2B	100%	100%	100%	100%	98.6%	{Neuroblastoma, susceptibility to, 2}, 613013;Neuroblastoma with Hirschsprung disease, 613013;Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PIK3CA	100%	100%	100%	100%	99.9%	Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Nevus, epidermal, somatic mosaic, 162900;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108
PMS2	93.4%	93.4%	99.8%	99.6%	98.9%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
PMS2CL						
POLD1	100%	100%	100%	99.9%	99%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

POLE	100%	100%	100%	100%	99.3%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336
POLH	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, variant type, 278750
POT1	100%	100%	100%	99.9%	99.3%	Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367
POU6F2	100%	100%	100%	99.9%	99.2%	{Wilms tumor susceptibility-5}, 601583
PRDM10	100%	100%	100%	100%	99.4%	?Birt-Hogg-Dube syndrome 2, 620459
PRF1	100%	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRKAR1A	100%	100%	100%	100%	99.6%	Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Adrenocortical tumor, somatic;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PRKN	100%	100%	100%	100%	99.8%	Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000
PRSS1	100%	100%	100%	99.6%	96.8%	Pancreatitis, hereditary, 167800
PTCH1	100%	100%	100%	100%	99.2%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828
PTEN	89.6%	89.6%	100%	100%	99.6%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
PTPN11	89.2%	89.2%	100%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
RAD50	100%	100%	100%	100%	99.7%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	87.3%	85.8%	100%	100%	99.2%	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390
RAD51D	100%	100%	100%	100%	99.8%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RAF1	96.1%	93.8%	100%	100%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RB1	96%	96%	100%	100%	99.8%	Small cell cancer of the lung, somatic, 182280;Bladder cancer, somatic, 109800;Retinoblastoma, trilateral, 180200;Osteosarcoma, somatic, 259500;Retinoblastoma, 180200
RECQL4	100%	100%	100%	100%	99.4%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
REST	100%	100%	100%	99.9%	99.5%	Deafness, autosomal dominant 27, 612431;{Wilms tumor 6, susceptibility to}, 616806;Fibromatosis, gingival, 5, 617626
RET	100%	100%	100%	100%	99.4%	{Hirschsprung disease, susceptibility to, 1}, 142623;Multiple endocrine neoplasia IIA, 171400;{Hirschsprung disease, protection against}, 142623;Medullary thyroid carcinoma, 155240;Pheochromocytoma, 171300;Multiple endocrine neoplasia IIB, 162300
RHBDF2	100%	100%	100%	99.9%	98.5%	Tylosis with esophageal cancer, 148500
RIT1	100%	100%	100%	100%	99.8%	Noonan syndrome 8, 615355

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RNASEL	100%	100%	100%	100%	99.6%	Prostate cancer 1, 601518
RNF43	100%	100%	100%	100%	99.5%	Sessile serrated polyposis cancer syndrome, 617108
RPA1	93.6%	93.3%	100%	100%	99.6%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RPL11	96.3%	94.4%	100%	100%	99.9%	Diamond-Blackfan anemia 7, 612562
RPL15	100%	98.5%	100%	100%	100%	Diamond-Blackfan anemia 12, 615550
RPL18	100%	100%	100%	100%	99.6%	?Diamond-Blackfan anemia 18, 618310
RPL27	100%	100%	100%	100%	100%	?Diamond-Blackfan anemia 16, 617408
RPL35A	100%	100%	100%	100%	99.1%	Diamond-Blackfan anemia 5, 612528
RPL5	100%	100%	100%	100%	99.9%	Diamond-Blackfan anemia 6, 612561
RPS10	89.1%	89.1%	100%	100%	99.7%	Diamond-Blackfan anemia 9, 613308
RPS15A	63%	63%	100%	100%	99.6%	?Diamond-Blackfan anemia 20, 618313
RPS17	100%	100%	100%	100%	99.6%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	100%	100%	100%	99.2%	Diamond-Blackfan anemia 1, 105650
RPS20	100%	100%	100%	100%	99.6%	
RPS24	88.4%	86.1%	100%	100%	99.7%	Diamond-blackfan anemia 3, 610629
RPS26	100%	99.9%	100%	100%	99.7%	Diamond-Blackfan anemia 10, 613309
RPS27	100%	100%	100%	100%	99.7%	?Diamond-Blackfan anemia 17, 617409

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RPS28	100%	100%	100%	100%	99.9%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	100%	100%	100%	99.4%	Diamond-Blackfan anemia 13, 615909
RPS7	100%	100%	100%	100%	99.6%	Diamond-Blackfan anemia 8, 612563
RTEL1	100%	100%	100%	99.9%	99.4%	Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373
RUNX1	100%	100%	100%	99.7%	97.9%	Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626
SAMD9	100%	100%	100%	100%	99.8%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMD9L	100%	100%	100%	100%	99.9%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SBDS	100%	100%	100%	100%	99.9%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SDHA	92.6%	89.3%	100%	100%	99.7%	Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma/paraganglioma syndrome 5, 614165
SDHAF2	100%	99.7%	100%	100%	99.6%	Pheochromocytoma/paraganglioma syndrome 2, 601650
SDHB	94%	94%	100%	100%	99.7%	Pheochromocytoma/paraganglioma syndrome 4, 115310;Mitochondrial complex II deficiency, nuclear type 4, 619224;Gastrointestinal stromal tumor, 606764;Paraganglioma and gastric stromal sarcoma, 606864
SDHC	77.5%	77.5%	100%	100%	99.9%	Pheochromocytoma/paraganglioma syndrome 3, 605373;Paraganglioma and gastric stromal sarcoma, 606864;Gastrointestinal stromal tumor, 606764
SDHD	78.9%	78.9%	100%	100%	99.5%	Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167
SEMA4A	100%	100%	100%	100%	98.9%	Retinitis pigmentosa 35, 610282;Cone-rod dystrophy 10, 610283
SFTPA1	96.2%	95.1%	100%	100%	99.8%	Interstitial lung disease 1, 619611

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SFTPA2	100%	100%	100%	100%	99.5%	Interstitial lung disease 2, 178500
SH2B3	100%	100%	100%	99.9%	99.3%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	99.4%	91.9%	73.1%	Lymphoproliferative syndrome, X-linked, 1, 308240
SHOC2	100%	100%	100%	100%	99.9%	Noonan syndrome-like with loose anagen hair 1, 607721
SLC25A11	100%	100%	100%	100%	99.3%	Pheochromocytoma/paraganglioma syndrome 6, 618464
SLX4	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group P, 613951
SMAD4	95.4%	95.4%	100%	100%	99.8%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD9	100%	100%	100%	100%	99.1%	Pulmonary hypertension, primary, 2, 615342
SMARCA4	100%	100%	100%	99.9%	99.3%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SMARCB1	95.2%	95.2%	100%	100%	99.7%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCE1	100%	100%	100%	100%	99.8%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938
SOS1	98.8%	98.6%	100%	100%	99.6%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SPINK1	100%	100%	100%	100%	100%	Tropical calcific pancreatitis, 608189;Pancreatitis, hereditary, 167800;{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPRED1	97.6%	97.6%	100%	100%	100%	Legius syndrome, 611431
SQSTM1	100%	100%	100%	100%	98.9%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158;Paget disease of bone 3, 167250
STK11	99.9%	98.5%	100%	99.9%	98.8%	Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300
SUCLG2	100%	100%	100%	100%	99.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SUFU	100%	100%	100%	100%	99.6%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
TERC						Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	97%	95.4%	100%	99.9%	99.3%	
TERT	99.6%	97.7%	100%	100%	99.4%	Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626
TG	100%	100%	100%	99.9%	99.2%	{Autoimmune thyroid disease, susceptibility to, 3}, 608175;Thyroid dyshormonogenesis 3, 274700
TGFBR1	100%	100%	100%	100%	99.6%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
THPO	100%	100%	100%	100%	99.3%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TINF2	100%	100%	100%	100%	99.7%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TMEM127	100%	100%	100%	100%	99.5%	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	100%	100%	100%	100%	99.5%	Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810
TP53	95.1%	95.1%	100%	99.9%	99.4%	{Basal cell carcinoma 7}, 614740;{Adrenocortical carcinoma, pediatric}, 202300;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Li-Fraumeni syndrome, 151623;Pancreatic cancer, somatic, 260350;Nasopharyngeal carcinoma, somatic, 607107;{Osteosarcoma}, 259500;{Choroid plexus papilloma}, 260500;{Colorectal cancer}, 114500;{Glioma susceptibility 1}, 137800;Bone marrow failure syndrome 5, 618165
TRIM28	100%	100%	100%	100%	99.4%	Wilms tumor 7, 621332
TRIM37	98.3%	97.6%	100%	100%	99.7%	Mulibrey nanism, 253250
TRIP13	100%	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 9, 619011;Mosaic variegated aneuploidy syndrome 3, 617598

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TSC1	100%	100%	100%	100%	99.6%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSC2	100%	100%	100%	100%	99.3%	Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254
USB1	93.9%	93.2%	100%	100%	99.1%	Poikiloderma with neutropenia, 604173
VHL	87.7%	87.7%	100%	99.9%	98.8%	Hemangioblastoma, cerebellar, somatic;Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300
WAS	99.6%	95.8%	98.5%	87.7%	68.3%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900
WRAP53	100%	99.9%	100%	99.9%	99.2%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	100%	100%	100%	100%	99.7%	Werner syndrome, 277700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

WT1	100%	100%	100%	99.9%	99.3%	Mesothelioma, somatic, 156240;Meacham syndrome, 608978;Frasier syndrome, 136680;Nephrotic syndrome, type 4, 256370;Denys-Drash syndrome, 194080;Wilms tumor, type 1, 194070
XPA	100%	100%	100%	100%	99.9%	Xeroderma pigmentosum, group A, 278700
XPC	100%	100%	100%	100%	99.6%	Xeroderma pigmentosum, group C, 278720
ZCCHC8	100%	100%	100%	100%	99.5%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors