HEREDITARY CANCER GENE PANEL DG 3.00
(242 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
A2ML1	100	99,6	100	100	{Otitis media, susceptibility to}, 166760
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
AIP	100	99	100	100	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
AKT1	100	99,5	100	100	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500
ALK	100	99,4	100	100	{Neuroblastoma, susceptibility to, 3}, 613014
AMH	96,4	83,8	100	99,8	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100	99,5	100	100	Persistent Mullerian duct syndrome, type II, 261550
ANKRD26	95	89,3	97,2	97,2	Thrombocytopenia 2, 188000
APC	100	99,7	100	100	Desmoid disease, hereditary, 135290 Adenomatous polyposis coli, 175100 Gardner syndrome, 175100 Hepatoblastoma, somatic, 114550 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Gastric cancer, somatic, 613659 Adenoma, periampullary, somatic, 0
ARMC5	100	99,4	100	100	ACTH-independent macronodular adrenal hyperplasia 2, 615954

ASXL1	99,8	99,3	99,8	99,8	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ATM	99,8	98,1	100	100	Ataxia-telangiectasia, 208900 {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATR	99,9	99,4	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	100	99,9	100	99,9	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BAP1	84,4	83	100	100	Tumor predisposition syndrome, 614327
BARD1	100	99,8	100	100	{Breast cancer, susceptibility to}, 114480
BLM	99,8	98,3	100	100	Bloom syndrome, 210900
BMPR1A	99,8	96,6	100	100	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900 Juvenile polyposis syndrome, infantile form, 174900
BRAF	91	81,1	100	100	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BRCA1	99,4	98,8	100	100	Fanconi anemia, complementation group S, 617883 {Pancreatic cancer, susceptibility to, 4}, 614320 {Breast-ovarian cancer, familial, 1}, 604370
BRCA2	99,8	98,5	100	100	{Pancreatic cancer 2}, 613347 {Breast cancer, male, susceptibility to}, 114480 {Glioblastoma 3}, 613029 Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724 {Medulloblastoma}, 155255 {Prostate cancer}, 176807 {Breast-ovarian cancer, familial, 2}, 612555
BRIP1	99,9	99	100	100	Fanconi anemia, complementation group J, 609054 {Breast cancer, early-onset, susceptibility to}, 114480

BUB1	99,8	98,8	100	100	Colorectal cancer with chromosomal instability, somatic, 114500
BUB1B	99,6	98,9	100	100	Colorectal cancer, somatic, 114500 [Premature chromatid separation trait], 176430 Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	99,8	99,1	100	100	No OMIM disease ID
CARD11	100	99,9	100	100	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CBL	97,3	97,1	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CD27	99,9	96,9	100	100	Lymphoproliferative syndrome 2, 615122
CD70	99,8	97,7	100	100	Lymphoproliferative syndrome 3, 618261
CDC73	100	99,4	100	100	Parathyroid carcinoma, 608266 Parathyroid adenoma with cystic changes, 145001 Hyperparathyroidism-jaw tumor syndrome, 145001 Hyperparathyroidism, familial primary, 145000
CDH1	99,2	99,1	96,1	96	Endometrial carcinoma, somatic, 608089 {Prostate cancer, susceptibility to}, 176807 Blepharocheilodontic syndrome 1, 119580 {Breast cancer, lobular}, 114480 Ovarian cancer, somatic, 167000 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215
CDH23	100	100	100	100	{Pituitary adenoma 5, multiple types}, 617540 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CDK4	100	99,7	100	100	{Melanoma, cutaneous malignant, 3}, 609048
CDKN1A	100	100	100	100	No OMIM disease ID
CDKN1B	100	99,8	100	100	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	88	77,8	99,3	97,3	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDKN2A	92,3	92,1	100	100	{Melanoma and neural system tumor syndrome}, 155755 {Melanoma, cutaneous malignant, 2}, 155601 {Melanoma-pancreatic cancer syndrome}, 606719

CDKN2B	100	99,9	100	100	No OMIM disease ID
CDKN2C	100	100	100	100	No OMIM disease ID
CEBPA	98,6	83,9	99,3	94,7	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CHEK2	85	81,5	100	100	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Prostate cancer, familial, susceptibility to}, 176807 {Breast cancer, susceptibility to}, 114480 {Breast and colorectal cancer, susceptibility to}, 0
CREBBP	99,7	98,5	100	100	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CTC1	100	99,6	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Celiac disease, susceptibility to, 3}, 609755 Autoimmune lymphoproliferative syndrome, type V, 616100 {Hashimoto thyroiditis}, 140300
CTNNA1	99,3	98,1	100	100	Macular dystrophy, patterned, 2, 608970
CTR9	100	99,9	100	100	No OMIM disease ID
CYLD	99,8	98	100	100	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
DDB2	99,6	97,5	100	100	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	85,2	80,7	100	100	Warsaw breakage syndrome, 613398
DDX41	100	100	100	100	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DICER1	99,8	99	100	100	GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200
DIS3L2	100	99,8	100	100	Perlman syndrome, 267000
DKC1	99,8	98,7	100	99,7	Dyskeratosis congenita, X-linked, 305000
DLST	96,7	90,3	100	100	Paragangliomas 7, 618475

DNAJC21	99,8	98,7	100	100	Bone marrow failure syndrome 3, 617052
EGFR	100	100	100	99,8	?Inflammatory skin and bowel disease, neonatal, 2, 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	89,3	82,2	100	100	[Hemoglobin, high altitude adaptation], 609070 Erythrocytosis, familial, 3, 609820
EGLN2	100	99,8	100	100	No OMIM disease ID
ELANE	99,7	97,4	100	100	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ELP1	99,8	99	100	100	Dysautonomia, familial, 223900
EPCAM	98,6	90,3	99,8	98,3	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
ERCC1	100	99,3	100	100	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100	99,7	100	100	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	96,9	96,3	100	100	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC4	100	99,9	100	100	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC5	100	99,7	100	100	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100	100	100	100	{Macular degeneration, age-related, susceptibility to, 5}, 613761 {Lung cancer, susceptibility to}, 211980 Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
ESR2	100	99,7	100	100	?Ovarian dysgenesis 8, 618187

ETV6	100	99,9	100	100	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EXT1	99,9	98,4	100	100	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100	99,3	100	100	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EZH2	100	99,5	100	100	Weaver syndrome, 277590
FAN1	100	99,8	100	100	Interstitial nephritis, karyomegalic, 614817
FANCA	100	99,4	100	100	Fanconi anemia, complementation group A, 227650
FANCB	98,6	94,1	100	100	Fanconi anemia, complementation group B, 300514
FANCC	97,2	96,6	97,3	97,3	Fanconi anemia, complementation group C, 227645
FANCD2	99,5	97,5	98,8	98,8	Fanconi anemia, complementation group D2, 227646
FANCE	89,8	85,1	100	99,9	Fanconi anemia, complementation group E, 600901
FANCF	100	100	100	100	Fanconi anemia, complementation group F, 603467
FANCG	100	99,9	100	100	Fanconi anemia, complementation group G, 614082
FANCI	99,9	99,2	100	100	Fanconi anemia, complementation group I, 609053
FANCL	100	98,6	100	100	Fanconi anemia, complementation group L, 614083
FANCM	99,6	97,3	100	100	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
FAS	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IA, 601859 {Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
FBXW7	99,9	99,2	100	100	No OMIM disease ID
FH	92,1	88,3	100	100	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLCN	100	100	100	100	Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500

G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GALNT12	85,8	82,7	97,8	94,6	{Colorectal cancer, susceptibility to, 1}, 608812
GATA2	100	98,3	100	100	Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626
GDNF	100	100	100	100	{Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711 Central hypoventilation syndrome, 209880
GFI1	100	99,2	100	100	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GPC3	99,1	94,7	100	100	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPR161	100	100	100	100	No OMIM disease ID
GREM1	100	100	100	100	No OMIM disease ID
GRHL2	100	100	100	100	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
HAVCR2	100	100	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HNF1A	100	99,8	100	100	{Diabetes mellitus, insulin-dependent}, 222100 MODY, type III, 600496 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520 {Diabetes mellitus, noninsulin-dependent, 2}, 125853
HOXB13	100	99,1	100	100	{Prostate cancer, hereditary, 9}, 610997
IDH1	93,3	80,1	100	100	{Glioma, susceptibility to, somatic}, 137800
IDH2	99,7	97,4	100	99,8	D-2-hydroxyglutaric aciduria 2, 613657
IKZF1	99,3	99,3	100	100	Immunodeficiency, common variable, 13, 616873
IPMK	99,2	92	100	100	No OMIM disease ID

ITK	100	98,9	100	100	Lymphoproliferative syndrome 1, 613011
KIF1B	100	99,6	100	100	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700
KIT	100	99,6	100	100	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, somatic, 601626 Mastocytosis, systemic, somatic, 154800
KRAS	99,5	96,9	100	100	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
LHCGR	94,1	92,3	100	100	Leydig cell adenoma, somatic, with precocious puberty, 176410 Precocious puberty, male, 176410 Luteinizing hormone resistance, female, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320
LIG4	100	99,9	100	100	{Multiple myeloma, resistance to}, 254500 LIG4 syndrome, 606593
LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAD2L2	100	99,9	100	100	?Fanconi anemia, complementation group V, 617243
MAP2K1	99,8	97,1	100	100	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	98,5	95,1	100	100	Cardiofaciocutaneous syndrome 4, 615280

MAX	100	98,9	100	100	{Pheochromocytoma, susceptibility to}, 171300
MCM8	100	99,6	94,4	94,4	?Premature ovarian failure 10, 612885
MCM9	99,9	99,8	100	100	Ovarian dysgenesis 4, 616185
MDH2	98	97,9	100	100	Developmental and epileptic encephalopathy 51, 617339
MEN1	96,9	94,8	100	100	Multiple endocrine neoplasia 1, 131100 Angiofibroma, somatic, 0 Adrenal adenoma, somatic, 0 Parathyroid adenoma, somatic, 0 Lipoma, somatic, 0 Carcinoid tumor of lung, 0
MET	100	99,5	100	100	{Osteofibrous dysplasia, susceptibility to}, 607278 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
MITF	100	99,9	100	100	COMMAD syndrome, 617306 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MLH1	100	99,9	100	100	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MPL	100	99,5	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977
MRE11	98,9	93,3	100	100	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	99	96,9	100	100	Mismatch repair cancer syndrome 2, 619096 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435
MSH3	98	97,3	98	98	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089
MSH6	100	99,8	100	100	{Endometrial cancer, familial}, 608089 Mismatch repair cancer syndrome 3, 619097 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MTAP	99,1	93,5	100	100	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250

MUTYH	100	100	100	100	Gastric cancer, somatic, 613659 Adenomas, multiple colorectal, 608456
NBN	99,9	98,6	100	100	Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135
NF1	92,6	90,2	100	100	Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Watson syndrome, 193520 Neurofibromatosis, type 1, 162200
NF2	100	99,9	100	100	Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091 Neurofibromatosis, type 2, 101000
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100	99,8	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	98,2	85,3	100	100	Leukemia, acute myeloid, somatic, 601626
NRAS	100	100	100	100	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NSD1	100	99,9	100	100	Sotos syndrome 1, 117550
NTHL1	100	99,8	100	100	Familial adenomatous polyposis 3, 616415
PALB2	100	100	100	100	{Pancreatic cancer, susceptibility to, 3}, 613348 Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480
PARN	81,2	81,1	88,1	87,6	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PAX5	98,7	96,1	100	100	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PDGFB	100	99,3	100	100	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174

PDGFRA	100	100	100	100	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PHOX2B	100	99,7	99,5	97,8	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 {Neuroblastoma, susceptibility to, 2}, 613013
PIK3CA	98	97,8	100	100	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Macrodactyly, somatic, 155500 Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
PMS2	84,3	82,8	100	100	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
PMS2CL					No OMIM disease ID
POLD1	98,5	95,2	100	100	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	100	99,8	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336 {Colorectal cancer, susceptibility to, 12}, 615083
POLH	100	99,6	100	100	Xeroderma pigmentosum, variant type, 278750
POT1	99,9	99	100	100	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
POU6F2	95,2	95,2	100	100	{Wilms tumor susceptibility-5}, 601583
PPM1D	100	99,9	100	100	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PRF1	91,2	90,8	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027

PRKAR1A	99,3	93,5	100	100	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0
PRKN	67	66,2	75,3	75,3	Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000 Adenocarcinoma of lung, somatic, 211980
PRSS1	100	100	100	100	Pancreatitis, hereditary, 167800
PTCH1	99,2	97,6	99,9	99,8	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	99,9	99	100	100	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTEN	99,5	97	100	100	Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 {Meningioma}, 607174
PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
RAD50	97,5	91,6	100	100	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	100	99,8	100	100	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 Fanconi anemia, complementation group O, 613390
RAD51D	100	99,9	100	100	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RAF1	100	100	100	100	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RB1	96,8	92,1	100	100	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800 Retinoblastoma, trilateral, 180200 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200

RECQL4	99,8	98,1	100	99,9	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
REST	98,5	98,2	98,6	98,6	{Wilms tumor 6, susceptibility to}, 616806 Fibromatosis, gingival, 5, 617626 ?Deafness, autosomal dominant 27, 612431
RET	99,9	99,1	100	100	Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 {Hirschsprung disease, protection against}, 142623 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 1}, 142623
RHBDF2	99,9	98,6	100	100	Tylosis with esophageal cancer, 148500
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RMRP					Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEL	100	99,8	100	100	Prostate cancer 1, 601518
RNF43	99,9	99,1	100	100	Sessile serrated polyposis cancer syndrome, 617108
RPL11	100	100	100	100	Diamond-Blackfan anemia 7, 612562
RPL15	86,8	78	100	100	?Diamond-Blackfan anemia 12, 615550
RPL18	100	100	100	100	?Diamond-Blackfan anemia 18, 618310
RPL27	73,6	56,5	100	100	?Diamond-Blackfan anemia 16, 617408
RPL35A	97,1	88,7	100	100	Diamond-Blackfan anemia 5, 612528
RPL5	86,2	70	100	100	Diamond-Blackfan anemia 6, 612561
RPS10	98	92,5	100	100	Diamond-Blackfan anemia 9, 613308
RPS15A	96,9	86,7	80,5	80,4	?Diamond-Blackfan anemia 20, 618313
RPS17	84,2	69,8	100	100	Diamond-Blackfan anemia 4, 612527
RPS19	100	99,6	100	100	Diamond-Blackfan anemia 1, 105650

RPS20	98,6	93,6	100	100	No OMIM disease ID
RPS24	98,4	93,1	100	100	Diamond-blackfan anemia 3, 610629
RPS26	95,7	84,9	100	100	Diamond-Blackfan anemia 10, 613309
RPS27	85,9	60,6	100	100	?Diamond-Blackfan anemia 17, 617409
RPS28	100	94,8	100	100	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	82	74,7	100	100	Diamond-Blackfan anemia 13, 615909
RPS7	80	68,7	100	100	Diamond-Blackfan anemia 8, 612563
RTEL1	99,5	96,8	100	100	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RUNX1	99,3	94,9	100	100	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SAMD9	100	99,8	100	100	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041
SAMD9L	100	100	100	100	Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Ataxia-pancytopenia syndrome, 159550
SBDS	100	100	100	100	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400
SDHA	85,8	80,4	100	100	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Paragangliomas 5, 614165 Mitochondrial respiratory chain complex II deficiency, 252011
SDHAF2	94,6	94,2	98,9	95,4	Paragangliomas 2, 601650
SDHB	100	100	100	100	Pheochromocytoma, 171300 Paragangliomas 4, 115310 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864
SDHC	100	99,3	100	100	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	54	51,6	80,1	80,1	Paragangliomas 1, with or without deafness, 168000 Mitochondrial complex II deficiency, 252011

					Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SEMA4A	100	99,8	100	100	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SFTPA1	100	100	100	100	No OMIM disease ID
SFTPA2	100	100	100	100	Pulmonary fibrosis, idiopathic, 178500
SH2B3	99,4	95,1	100	99,9	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	97,2	94	100	100	Lymphoproliferative syndrome, X-linked, 1, 308240
SHOC2	99,9	99,4	100	100	Noonan syndrome-like with loose anagen hair 1, 607721
SLX4	100	99,8	100	100	Fanconi anemia, complementation group P, 613951
SMAD4	100	99,9	100	100	Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350
SMAD9	100	99,9	100	100	Pulmonary hypertension, primary, 2, 615342
SMARCA4	99,9	99	100	100	{Rhabdoid tumor predisposition syndrome 2}, 613325 Coffin-Siris syndrome 4, 614609
SMARCB1	100	100	100	100	Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091 Coffin-Siris syndrome 3, 614608 {Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCE1	95,6	88,8	100	100	{Meningioma, familial, susceptibility to}, 607174 Coffin-Siris syndrome 5, 616938
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SPINK1	100	99,3	100	100	{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189
SPRED1	100	98,9	100	100	Legius syndrome, 611431
SQSTM1	98,8	95,5	100	100	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145

					Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
STK11	92,4	91,7	100	100	Testicular tumor, somatic, 273300 Peutz-Jeghers syndrome, 175200 Pancreatic cancer, somatic, 260350 Melanoma, malignant, somatic, 0
SUFU	100	100	100	100	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174 Joubert syndrome 32, 617757
TERC					{Aplastic anemia}, 614743 Dyskeratosis congenita, autosomal dominant 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERF2IP	99,9	97,8	83,7	83,7	No OMIM disease ID
TERT	96,2	94,5	100	100	{Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 {Leukemia, acute myeloid}, 601626 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989
TG	100	99,4	100	100	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 Thyroid dyshormonogenesis 3, 274700
THPO	81,4	81	100	100	Thrombocythemia 1, 187950
TINF2	100	100	100	100	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TMEM127	99,5	96,5	100	100	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	94,6	93,3	99,2	98	Osteolysis, familial expansile, 174810 {Paget disease of bone 2, early-onset}, 602080 Osteopetrosis, autosomal recessive 7, 612301
TP53	99,9	97,7	91,7	91,7	{Adrenocortical carcinoma, pediatric}, 202300 {Glioma susceptibility 1}, 137800 {Basal cell carcinoma 7}, 614740 Bone marrow failure syndrome 5, 618165 {Colorectal cancer}, 114500 Nasopharyngeal carcinoma, somatic, 607107 Breast cancer, somatic, 114480 {Osteosarcoma}, 259500 {Choroid plexus papilloma}, 260500

					Li-Fraumeni syndrome, 151623 Hepatocellular carcinoma, somatic, 114550 Pancreatic cancer, somatic, 260350
TRIM28	96,8	95,2	99,8	99,3	No OMIM disease ID
TRIM37	98,6	98,1	98,7	98,7	Mulibrey nanism, 253250
TRIP13	100	100	100	100	Mosaic variegated aneuploidy syndrome 3, 617598 Oocyte maturation defect 9, 619011
TSC1	99,8	98,8	100	100	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100	99,6	100	100	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
USB1	100	99,4	100	100	Poikiloderma with neutropenia, 604173
VHL	96,3	91,4	100	100	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
WAS	95,9	85,3	100	99,8	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WRAP53	100	100	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	99,9	98,8	100	100	Werner syndrome, 277700
WT1	97,3	95,4	97,7	97,7	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370
XPA	99,6	95,6	100	100	Xeroderma pigmentosum, group A, 278700
XPC	100	100	100	100	Xeroderma pigmentosum, group C, 278720

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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors