HEREDITARY CANCER GENE PANEL DG 3.2.0 (240 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
A2ML1	99,9	99,3	100	100	No OMIM disease ID
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
AIP	100	99,6	100	100	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
AKT1	100	99,9	100	100	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Colorectal cancer, somatic, 114500 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000
ALK	100	99,4	100	100	No OMIM disease ID
AMH	99,4	92,9	100	100	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100	99,6	100	100	Persistent Mullerian duct syndrome, type II, 261550
ANKRD26	94,6	88,5	97,2	97	Thrombocytopenia 2, 188000
APC	99,9	99,7	100	100	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	99,1	100	100	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ASXL1	99,8	98,9	100	100	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
ATM	99,4	97,1	100	100	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic,

					T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,
ATR	99,7	99	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	100	99,8	100	100	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BAP1	83,9	82,4	100	100	Tumor predisposition syndrome, 614327
BARD1	100	99,8	100	100	No OMIM disease ID
BLM	99,3	97,7	100	100	Bloom syndrome, 210900
BMPR1A	99,5	94	100	100	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BRAF	89,4	77,6	100	100	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	99,4	98,4	100	100	Fanconi anemia, complementation group S, 617883
BRCA2	99,1	98,2	100	100	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
BRIP1	99,4	98,5	100	100	Fanconi anemia, complementation group J, 609054
BUB1	99,7	98,4	100	99,9	Colorectal cancer with chromosomal instability, somatic, 114500
BUB1B	99,3	98,3	100	100	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	99,1	97,9	100	100	No OMIM disease ID
CARD11	100	99,9	100	100	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206
CBL	97,3	96,9	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CD27	99,9	98,3	100	100	Lymphoproliferative syndrome 2, 615122
CD70	100	96,8	100	100	Lymphoproliferative syndrome 3, 618261
CDC73	99,8	98,3	100	100	Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001

					Parathyroid carcinoma, 608266 Hyperparathyroidism-jaw tumor syndrome, 145001
CDH1	99,2	98,6	99,2	99,1	Ovarian cancer, somatic, 167000 Blepharocheilodontic syndrome 1, 119580 Endometrial carcinoma, somatic, 608089 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215
CDH23	100	100	100	100	Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 12, 601386
CDK4	99,9	99	100	100	No OMIM disease ID
CDKN1A	100	100	100	100	No OMIM disease ID
CDKN1B	99,9	99,3	100	100	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	89,9	81,6	98,9	95,8	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDKN2A	92,3	92,3	100	100	No OMIM disease ID
CDKN2B	100	99,7	100	100	No OMIM disease ID
CDKN2C	100	100	100	100	No OMIM disease ID
CEBPA	95,9	80,1	99,7	97,3	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CHEK2	84,9	80,7	100	100	Osteosarcoma, somatic, 259500 Li-Fraumeni syndrome 2, 609265
CREBBP	99,6	97,8	100	100	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CTC1	100	99,1	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100	100	100	100	Autoimmune lymphoproliferative syndrome, type V, 616100
CTNNA1	98,8	97,2	100	100	Macular dystrophy, patterned, 2, 608970
CTR9	99,9	99,8	100	100	No OMIM disease ID
CYLD	99,6	98,8	100	100	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606 ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132
DDB2	99,7	97,7	100	100	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	84,9	80	100	100	Warsaw breakage syndrome, 613398
DDX41	100	100	100	100	No OMIM disease ID

DICER1	99,5	98,5	100	100	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295
DIS3L2	100	100	100	100	Perlman syndrome, 267000
DKC1	99,7	97,2	100	99,6	Dyskeratosis congenita, X-linked, 305000
DLST	95,7	87,7	100	100	Paragangliomas 7, 618475
DNAJC21	99,5	97,4	100	100	Bone marrow failure syndrome 3, 617052
EGFR	100	100	100	100	?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
EGLN1	89,1	80,1	100	100	Erythrocytosis, familial, 3, 609820
EGLN2	100	100	100	100	No OMIM disease ID
ELANE	99,9	98,8	100	100	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELP1	99,8	98,9	100	100	Dysautonomia, familial, 223900
EPCAM	97,5	89,6	100	100	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
ERCC1	100	96,4	100	100	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100	99,4	100	100	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	96,8	95,6	100	100	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	100	99,9	100	100	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760 Fanconi anemia, complementation group Q, 615272
ERCC5	99,9	99	100	100	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100	100	100	100	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540

					De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946
ESR2	99,9	98,9	100	100	?Ovarian dysgenesis 8, 618187
ETV6	100	99,3	100	100	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626
EXT1	99,6	97,1	100	100	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	99,9	99	100	100	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EZH2	99,7	98	100	100	Weaver syndrome, 277590
FANCA	99,9	98,7	100	100	Fanconi anemia, complementation group A, 227650
FANCB	98	91,7	100	99,6	Fanconi anemia, complementation group B, 300514
FANCC	96,9	95,7	97,3	97,3	Fanconi anemia, complementation group C, 227645
FANCD2	98,7	95,9	98,8	98,8	Fanconi anemia, complementation group D2, 227646
FANCE	90,7	85,5	100	100	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	99,8	98,6	100	100	Fanconi anemia, complementation group I, 609053
FANCL	99,4	97,6	100	100	Fanconi anemia, complementation group L, 614083
FANCM	98,9	96,3	100	100	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAS	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100	99,1	100	100	Autoimmune lymphoproliferative syndrome, type IB, 601859
FBXW7	99,9	98,2	100	99,9	No OMIM disease ID
FH	93,2	87,2	100	100	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FLCN	100	100	100	100	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541

GALNT12	86,4	82,6	97,9	94,7	No OMIM disease ID
GATA2	99,8	97	100	100	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GDNF	100	100	100	100	No OMIM disease ID
GFI1	100	99,9	100	100	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GPC3	98,8	92,9	100	99,9	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870
GPR161	100	100	100	100	No OMIM disease ID
GREM1	100	100	100	100	No OMIM disease ID
GRHL2	100	99,9	100	100	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100	99,8	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HOXB13	100	99,6	100	100	No OMIM disease ID
IDH1	90,6	75,5	100	100	No OMIM disease ID
IDH2	99,8	97,4	100	100	D-2-hydroxyglutaric aciduria 2, 613657
IKZF1	99,3	99,3	100	100	Immunodeficiency, common variable, 13, 616873
IPMK	98,9	89,9	100	100	No OMIM disease ID
ITK	99,8	98,6	100	100	Lymphoproliferative syndrome 1, 613011
KIF1B	99,9	99,2	100	100	Pheochromocytoma, 171300 Charcot-Marie-Tooth disease, type 2A1, 118210
KIT	100	99,4	100	100	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	99	97,8	100	100	Gastric cancer, somatic, 137215 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
LHCGR	96,6	92,4	100	100	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	99,8	99,3	100	100	LIG4 syndrome, 606593
LZTR1	100	99,9	100	100	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAD2L2	100	99,9	100	100	?Fanconi anemia, complementation group V, 617243
MAP2K1	99,6	96,1	100	100	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	98,5	95,3	100	100	Cardiofaciocutaneous syndrome 4, 615280
MAX	99,8	97,7	100	100	No OMIM disease ID
MCM8	99,9	98,8	94,4	94,3	?Premature ovarian failure 10, 612885
MCM9	99,9	99	100	100	Ovarian dysgenesis 4, 616185
MDH2	98	98	100	100	Developmental and epileptic encephalopathy 51, 617339
MEN1	96,2	94,1	100	100	Multiple endocrine neoplasia 1, 131100 Lipoma, somatic, Angiofibroma, somatic, Carcinoid tumor of lung, Adrenal adenoma, somatic, Parathyroid adenoma, somatic,
MET	100	99,4	100	100	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705
MITF	100	99,9	100	100	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306

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	99,8	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MRE11	98,2	88,6	100	100	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	98,5	94,5	100	100	Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome 2, 619096
MSH3	97,8	97,1	100	100	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089
MSH6	100	99,3	100	100	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome 3, 619097
MTAP	98,3	91,8	100	100	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MUTYH	100	100	100	100	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659
NBN	99,2	97,8	100	99,9	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NF1	91,8	89,3	100	100	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NF2	100	99,6	100	100	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100	99,2	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	95,3	84,9	100	100	Leukemia, acute myeloid, somatic, 601626
NRAS	100	100	100	100	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	99,8	100	100	Sotos syndrome 1, 117550
NTHL1	100	99,9	100	100	Familial adenomatous polyposis 3, 616415
PALB2	100	99,9	100	100	Fanconi anemia, complementation group N, 610832
PARN	81,1	80,4	88,3	87,6	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	98,8	95,4	100	100	No OMIM disease ID
PDGFB	100	100	100	100	Meningioma, SIS-related, 607174 Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907
PDGFRA	100	100	100	100	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PHOX2B	100	100	99,8	98,5	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880
PIK3CA	97,7	97,3	100	100	CLOVE syndrome, somatic, 612918 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 Macrodactyly, somatic, 155500 CLAPO syndrome, somatic, 613089 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108
PMS2	83,9	81,6	100	100	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
PMS2CL	NC	NC	NC	NC	No OMIM disease ID
POLD1	98,4	95,1	100	100	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	100	99,5	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLH	100	99,1	100	100	Xeroderma pigmentosum, variant type, 278750

POT1	99,5	98,5	100	100	No OMIM disease ID
POU6F2	95,2	95,1	100	100	No OMIM disease ID
PPM1D	100	99,9	100	100	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PRF1	91,2	90,1	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRKAR1A	97	89,1	100	100	Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Adrenocortical tumor, somatic,
PRKN	66,9	65,8	75,4	75,3	Adenocarcinoma of lung, somatic, 211980 Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000
PRSS1	100	100	100	100	Pancreatitis, hereditary, 167800
PTCH1	99,3	96,6	100	99,9	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
PTEN	99,5	97,2	100	100	Lhermitte-Duclos syndrome, 158350 Cowden syndrome 1, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309
PTPN11	97,7	87,6	100	100	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAD50	96,6	89,7	100	100	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	99,8	99,4	100	100	Fanconi anemia, complementation group O, 613390
RAD51D	100	99,6	100	100	No OMIM disease ID
RAF1	99,9	99,2	100	100	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RB1	96,3	93,2	100	99,9	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800 Retinoblastoma, trilateral, 180200

					Osteosarcoma, somatic, 259500 Retinoblastoma, 180200
RECQL4	99,9	98,6	100	100	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
REST	98,4	97,4	98,6	98,6	?Deafness, autosomal dominant 27, 612431 Fibromatosis, gingival, 5, 617626
RET	100	98,7	100	100	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIB, 162300
RHBDF2	99,8	98,4	100	100	Tylosis with esophageal cancer, 148500
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEL	100	99,6	100	100	Prostate cancer 1, 601518
RNF43	99,8	98,3	100	100	Sessile serrated polyposis cancer syndrome, 617108
RPL11	99,9	97,9	100	100	Diamond-Blackfan anemia 7, 612562
RPL15	84,9	70,4	100	99,5	?Diamond-Blackfan anemia 12, 615550
RPL18	100	99,9	100	100	?Diamond-Blackfan anemia 18, 618310
RPL27	68	56,6	100	100	?Diamond-Blackfan anemia 16, 617408
RPL35A	94,7	84,9	100	100	Diamond-Blackfan anemia 5, 612528
RPL5	81,9	59,7	100	100	Diamond-Blackfan anemia 6, 612561
RPS10	96,6	87,6	100	100	Diamond-Blackfan anemia 9, 613308
RPS15A	95,3	84,1	80,4	80,4	?Diamond-Blackfan anemia 20, 618313
RPS17	85	67,8	100	100	Diamond-Blackfan anemia 4, 612527
RPS19	100	99,9	100	100	Diamond-Blackfan anemia 1, 105650
RPS20	96,6	87,5	100	100	No OMIM disease ID
RPS24	96,2	90,3	100	100	Diamond-blackfan anemia 3, 610629
RPS26	93,2	81,2	100	100	Diamond-Blackfan anemia 10, 613309
RPS27	95,5	70	100	100	?Diamond-Blackfan anemia 17, 617409

RPS28	99,7	86,3	100	100	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	78	70,5	100	100	Diamond-Blackfan anemia 13, 615909
RPS7	81,7	66,9	100	100	Diamond-Blackfan anemia 8, 612563
RTEL1	99,7	97,2	100	100	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RUNX1	98,6	93	100	100	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SAMD9	99,9	99,8	100	100	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100	99,9	100	100	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SBDS	100	99,9	100	100	Shwachman-Diamond syndrome, 260400
SDHA	84,5	77,9	100	100	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011 Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165
SDHAF2	94,6	93,5	99,4	96,4	Paragangliomas 2, 601650
SDHB	100	100	100	100	Paragangliomas 4, 115310 Mitochondrial complex II deficiency, nuclear type 4, 619224 Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864
SDHC	100	98,9	100	100	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	53,8	49	80,1	80,1	Paragangliomas 1, with or without deafness, 168000 Paraganglioma and gastric stromal sarcoma, 606864 Mitochondrial complex II deficiency, nuclear type 3, 619167 Pheochromocytoma, 171300
SEMA4A	100	99,4	100	100	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283
SFTPA1	100	100	100	100	No OMIM disease ID
SFTPA2	100	100	100	100	Pulmonary fibrosis, idiopathic, 178500

SH2B3	99	94,7	100	100	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	97,8	92,9	100	100	Lymphoproliferative syndrome, X-linked, 1, 308240
SHOC2	99,8	99,6	100	99,9	Noonan syndrome-like with loose anagen hair 1, 607721
SLX4	100	99,9	100	100	Fanconi anemia, complementation group P, 613951
SMAD4	99,9	99,9	100	100	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD9	100	99,3	100	100	Pulmonary hypertension, primary, 2, 615342
SMARCA4	99,9	99,2	100	100	Coffin-Siris syndrome 4, 614609
SMARCB1	100	99,9	100	100	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCE1	93,7	85,9	100	100	Coffin-Siris syndrome 5, 616938
SOS1	99,6	97,9	100	99,9	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SPINK1	99,9	99	100	100	Tropical calcific pancreatitis, 608189 Pancreatitis, hereditary, 167800
SPRED1	99,8	98,2	100	100	Legius syndrome, 611431
SQSTM1	99,8	97,8	100	100	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	92,4	91,9	100	100	Melanoma, malignant, somatic, 155600 Pancreatic cancer, somatic, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
SUCLG2	91,7	79,1	100	100	No OMIM disease ID
SUFU	100	100	100	100	Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 Basal cell nevus syndrome, 109400
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	100	99,9	83,7	83,7	No OMIM disease ID

TERT	97	94,8	100	100	No OMIM disease ID
TG	99,9	98,5	100	100	Thyroid dyshormonogenesis 3, 274700
THPO	81,4	78,7	100	100	Thrombocythemia 1, 187950
TINF2	100	100	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMEM127	99,9	97,7	100	100	No OMIM disease ID
TNFRSF11A	94,9	93,8	99,1	97,7	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TP53	99	95,2	91,7	91,7	Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Nasopharyngeal carcinoma, somatic, 607107 Bone marrow failure syndrome 5, 618165
TRIM28	97,7	96,3	99,8	99,2	No OMIM disease ID
TRIM37	98,3	97,1	98,7	98,6	Mulibrey nanism, 253250
TRIP13	100	99,9	100	100	Oocyte maturation defect 9, 619011 Mosaic variegated aneuploidy syndrome 3, 617598
TSC1	99,5	98,2	100	100	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSC2	100	99,8	100	100	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254
USB1	100	98,8	100	100	Poikiloderma with neutropenia, 604173
VHL	95,5	90,6	100	100	Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic,
WAS	94,1	83,7	100	100	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WRAP53	100	100	100	99,9	Dyskeratosis congenita, autosomal recessive 3, 613988

WRN	99,3	98,2	100	99,9	Werner syndrome, 277700
WT1	97,6	96,1	97,7	97,7	Mesothelioma, somatic, 156240 Meacham syndrome, 608978 Frasier syndrome, 136680 Nephrotic syndrome, type 4, 256370 Denys-Drash syndrome, 194080 Wilms tumor, type 1, 194070
XPA	99,2	97,3	100	100	Xeroderma pigmentosum, group A, 278700
XPC	100	99,9	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.
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-protein coding genes.
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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors