HEREDITARY CANCER GENE PANEL DG 2.18 (231 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
A2ML1	100%	99,60%	100%	100%	No OMIM disease ID
ACD	100%	99,90%	100%	100%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
AIP	100%	99,00%	100%	100%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
AKT1	100%	99,50%	100%	100%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500
ALK	100%	99,40%	100%	100%	No OMIM disease ID
AMH	96,40%	83,80%	100%	99,80%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100%	99,50%	100%	100%	Persistent Mullerian duct syndrome, type II, 261550
ANKRD26	95,00%	89,30%	97,20%	97,20%	Thrombocytopenia 2, 188000
APC	100%	99,70%	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%	99,40%	100%	100%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ASXL1	100%	99,50%	99,90%	99,90%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ATM	99,80%	98,10%	100%	100%	Ataxia-telangiectasia, 208900 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0

ATR	99,90%	99,40%	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	100%	99,90%	100%	99,90%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BAP1	84,40%	83,00%	100%	100%	Tumor predisposition syndrome, 614327
BARD1	100%	99,80%	100%	100%	No OMIM disease ID
BLM	99,80%	98,30%	100%	100%	Bloom syndrome, 210900
BMPR1A	99,80%	96,60%	100%	100%	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900 Juvenile polyposis syndrome, infantile form, 174900
BRAF	95,60%	85,10%	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	99,40%	98,80%	100%	100%	Fanconi anemia, complementation group S, 617883
BRCA2	99,80%	98,50%	100%	100%	Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724
BRIP1	99,90%	99,00%	100%	100%	Fanconi anemia, complementation group J, 609054
BUB1	99,80%	98,80%	100%	100%	Colorectal cancer with chromosomal instability, somatic, 114500
BUB1B	99,60%	98,90%	100%	100%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	99,80%	99,10%	100%	100%	No OMIM disease ID
CARD11	100%	99,90%	100%	100%	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CBL	97,30%	97,10%	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CDC73	100%	99,40%	100%	100%	Parathyroid adenoma with cystic changes, 145001 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism, familial primary, 145000
CDH1	99,20%	99,10%	96,10%	96,00%	Endometrial carcinoma, somatic, 608089 Blepharocheilodontic syndrome 1, 119580 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215 Ovarian cancer, somatic, 167000

CDH23	100%	100%	100%	100%	Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CDK4	100%	99,70%	100%	100%	No OMIM disease ID
CDKN1A	100%	100%	100%	100%	No OMIM disease ID
CDKN1B	100%	99,80%	100%	100%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	86,30%	74,80%	99,20%	96,90%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDKN2A	92,30%	92,10%	100%	100%	No OMIM disease ID
CDKN2B	100%	99,90%	100%	100%	No OMIM disease ID
CDKN2C	100%	100%	100%	100%	No OMIM disease ID
CEBPA	98,60%	83,90%	99,30%	94,70%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CHEK2	85,00%	81,50%	100%	100%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500
CREBBP	99,70%	98,50%	100%	100%	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CTC1	100%	99,60%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTNNA1	99,30%	98,10%	100%	100%	Macular dystrophy, patterned, 2, 608970
CTR9	100%	99,90%	100%	100%	No OMIM disease ID
CYLD	99,80%	98,00%	100%	100%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
DDB2	99,60%	97,50%	100%	100%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	85,20%	80,70%	100%	100%	Warsaw breakage syndrome, 613398
DDX41	100%	100%	100%	100%	No OMIM disease ID
DICER1	99,80%	99,00%	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%	99,80%	100%	100%	Perlman syndrome, 267000
DKC1	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
DLST	96,70%	90,30%	100%	100%	Paragangliomas 7, 618475
DNAJC21	99,90%	99,00%	100%	100%	Bone marrow failure syndrome 3, 617052
EGFR	100%	100%	100%	99,80%	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980
EGLN1	89,30%	82,20%	100%	100%	Erythrocytosis, familial, 3, 609820

EGLN2	100%	99,80%	100%	100%	No OMIM disease ID
ELANE	99,70%	97,40%	100%	100%	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ELP1	99,80%	99,00%	100%	100%	Dysautonomia, familial, 223900
EPCAM	98,60%	90,30%	99,80%	98,30%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
ERCC1	100%	99,30%	100%	100%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100%	99,70%	100%	100%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	100%	99,40%	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC4	100%	99,90%	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%	99,60%	100%	100%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100%	100%	100%	100%	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%	99,70%	100%	100%	?Ovarian dysgenesis 8, 618187
ETV6	100%	99,90%	100%	100%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EXT1	99,90%	98,40%	100%	100%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100%	99,30%	100%	100%	Exostoses, multiple, type 2, 133701 Seizures, scoliosis, and macrocephaly syndrome, 616682
EZH2	100%	99,50%	100%	100%	Weaver syndrome, 277590
FAN1	100%	99,80%	100%	100%	Interstitial nephritis, karyomegalic, 614817
FANCA	100%	99,40%	100%	100%	Fanconi anemia, complementation group A, 227650
FANCB	98,60%	94,10%	100%	100%	Fanconi anemia, complementation group B, 300514
FANCC	99,90%	99,30%	100%	100%	Fanconi anemia, complementation group C, 227645
FANCD2	99,50%	97,50%	98,80%	98,80%	Fanconi anemia, complementation group D2, 227646
FANCE	89,80%	85,10%	100%	99,90%	Fanconi anemia, complementation group E, 600901

FANCF	100%	100%	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	100%	99,90%	100%	100%	Fanconi anemia, complementation group G, 614082
FANCI	99,90%	99,20%	100%	100%	Fanconi anemia, complementation group I, 609053
FANCL	100%	98,60%	100%	100%	Fanconi anemia, complementation group L, 614083
FANCM	99,60%	97,30%	100%	100%	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
FAS	100%	99,60%	100%	100%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	100%	99,60%	100%	100%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FH	92,10%	88,30%	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%	99,90%	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GALNT12	85,80%	82,70%	97,80%	94,60%	No OMIM disease ID
GATA2	100%	98,30%	100%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GDNF	100%	100%	100%	100%	Central hypoventilation syndrome, 209880
GFI1	100%	99,20%	100%	100%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GPC3	99,10%	94,70%	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%	99,80%	100%	100%	MODY, type III, 600496 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520
HOXB13	100%	99,10%	100%	100%	No OMIM disease ID
IDH1	93,30%	80,10%	100%	100%	No OMIM disease ID

IDH2	99,70%	97,40%	100%	99,80%	D-2-hydroxyglutaric aciduria 2, 613657
MR E11	98,90%	93,30%	100%	100%	Ataxia-telangiectasia-like disorder 1, 604391
IPMK	99,20%	92,00%	100%	100%	No OMIM disease ID
KIF1B	100%	99,60%	100%	100%	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210
KIT	100%	99,60%	100%	100%	Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, somatic, 601626 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KRAS	99,50%	96,90%	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	94,10%	92,30%	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%	99,90%	100%	100%	LIG4 syndrome, 606593
LZTR1	100%	99,90%	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAD2L2	100%	99,90%	100%	100%	?Fanconi anemia, complementation group V, 617243
MAP2K1	99,80%	97,10%	100%	100%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	98,50%	95,10%	100%	100%	Cardiofaciocutaneous syndrome 4, 615280
MAX	100%	98,90%	100%	100%	No OMIM disease ID
MDH2	98,00%	97,90%	100%	100%	Epileptic encephalopathy, early infantile, 51, 617339

MEN1	99,90%	99,10%	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%	99,50%	100%	100%	Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
MITF	100%	99,90%	100%	100%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MLH1	100%	99,90%	100%	100%	Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 2, 609310
MPL	100%	99,50%	100%	100%	Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498 Myelofibrosis with myeloid metaplasia, somatic, 254450
MSH2	99,80%	97,70%	100%	100%	Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 1, 120435
MSH3	100%	99,20%	100%	100%	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089
MSH6	100%	99,80%	100%	100%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MTAP	99,10%	93,50%	100%	100%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MUC5B	82,50%	72,90%	100%	100%	No OMIM disease ID
MUTYH	100%	100%	100%	100%	Gastric cancer, somatic, 613659 Adenomas, multiple colorectal, 608456
NBN	99,90%	98,60%	100%	100%	Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260 Leukemia, acute lymphoblastic, 613065
NF1	92,60%	90,20%	100%	100%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210

NF2	100%	99,90%	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%	99,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	98,20%	85,30%	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%	99,90%	100%	100%	Sotos syndrome 1, 117550
NTHL1	100%	99,80%	100%	100%	Familial adenomatous polyposis 3, 616415
PALB2	100%	100%	100%	100%	Fanconi anemia, complementation group N, 610832
PARN	100%	99,90%	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PAX5	98,70%	96,10%	100%	100%	No OMIM disease ID
PDGFB	100%	99,30%	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%	99,70%	99,50%	97,80%	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880
PIK3CA	100%	99,80%	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	84,30%	82,80%	100%	100%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
PMS2CL	NC	NC	NC	NC	No OMIM disease ID
POLD1	98,50%	95,20%	100%	100%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	100%	99,80%	100%	100%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLH	100%	99,60%	100%	100%	Xeroderma pigmentosum, variant type, 278750
POT1	99,90%	99,00%	100%	100%	No OMIM disease ID
POU6F2	100%	100%	100%	100%	No OMIM disease ID
PPM1D	100%	99,90%	100%	100%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PRF1	91,20%	90,80%	100%	100%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553
PRKAR1A	99,30%	93,50%	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	79,80%	78,80%	89,80%	89,80%	Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000 Adenocarcinoma of lung, somatic, 211980
PRSS1	100%	100%	100%	100%	Pancreatitis, hereditary, 167800
PTCH1	99,20%	97,60%	99,90%	99,80%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	99,90%	99,00%	100%	100%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTEN	99,50%	97,00%	100%	100%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
PTPN11	99,10%	93,70%	100%	100%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250

					Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
RAD50	97,50%	91,60%	100%	100%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	100%	99,80%	100%	100%	Fanconi anemia, complementation group O, 613390
RAD51D	100%	99,90%	100%	100%	No OMIM disease ID
RAF1	100%	100%	100%	100%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RB1	98,50%	93,70%	100%	100%	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800 Retinoblastoma, trilateral, 180200 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200
RECQL4	99,80%	98,10%	100%	99,90%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
REST	98,50%	98,20%	98,60%	98,60%	Fibromatosis, gingival, 5, 617626 ?Deafness, autosomal dominant 27, 612431
RET	99,90%	99,10%	100%	100%	Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Central hypoventilation syndrome, congenital, 209880
RHBDF2	99,90%	98,60%	100%	100%	Tylosis with esophageal cancer, 148500
RIT1	100%	100%	100%	100%	Noonan syndrome 8, 615355
RNASEL	100%	99,80%	100%	100%	Prostate cancer 1, 601518
RNF43	99,90%	99,10%	100%	100%	Sessile serrated polyposis cancer syndrome, 617108
RPL11	100%	100%	100%	100%	Diamond-Blackfan anemia 7, 612562
RPL15	86,80%	78,00%	100%	100%	?Diamond-Blackfan anemia 12, 615550
RPL18	100%	100%	100%	100%	?Diamond-Blackfan anemia 18, 618310
RPL27	73,60%	56,50%	100%	100%	?Diamond-Blackfan anemia 16, 617408
RPL35A	97,10%	88,70%	100%	100%	Diamond-Blackfan anemia 5, 612528
RPL5	86,20%	70,00%	100%	100%	Diamond-Blackfan anemia 6, 612561
RPS10	97,70%	91,70%	100%	100%	Diamond-Blackfan anemia 9, 613308
RPS15A	96,90%	86,70%	80,50%	80,40%	?Diamond-Blackfan anemia 20, 618313
RPS17	84,20%	69,80%	100%	100%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	99,60%	100%	100%	Diamond-Blackfan anemia 1, 105650
RPS20	98,60%	93,60%	100%	100%	No OMIM disease ID

RPS24	98,50%	93,40%	100%	100%	Diamond-blackfan anemia 3, 610629
RPS26	95,70%	84,90%	100%	100%	Diamond-Blackfan anemia 10, 613309
RPS27	89,30%	60,90%	100%	99,80%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	94,80%	100%	100%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	98,20%	100%	100%	Diamond-Blackfan anemia 13, 615909
RPS7	80,00%	68,70%	100%	100%	Diamond-Blackfan anemia 8, 612563
RTEL1	99,50%	96,80%	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	99,30%	94,90%	100%	100%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SAMD9	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	100%	100%	100%	100%	Ataxia-pancytopenia syndrome, 159550
SBDS	100%	100%	100%	100%	Shwachman-Diamond syndrome, 260400
SDHA	85,80%	80,40%	100%	100%	Leigh syndrome, 256000 Paragangliomas 5, 614165 Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial respiratory chain complex II deficiency, 252011
SDHAF2	94,60%	94,20%	98,90%	95,40%	Paragangliomas 2, 601650
SDHB	100%	100%	100%	100%	Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paragangliomas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864
SDHC	100%	99,30%	100%	100%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	54,00%	51,60%	80,10%	80,10%	Paragangliomas 1, with or without deafness, 168000 Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SEMA4A	100%	99,80%	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	99,40%	95,10%	100%	99,90%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100

SHOC2	99,90%	99,40%	100%	100%	Noonan syndrome-like with loose anagen hair, 607721
SLX4	100%	99,80%	100%	100%	Fanconi anemia, complementation group P, 613951
SMAD4	100%	99,90%	100%	100%	Polyposis, juvenile intestinal, 174900 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD9	100%	99,90%	100%	100%	Pulmonary hypertension, primary, 2, 615342
SMARCA4	99,90%	99,00%	100%	100%	Coffin-Siris syndrome 4, 614609
SMARCB1	100%	100%	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCE1	96,10%	88,10%	100%	100%	Coffin-Siris syndrome 5, 616938
SOS1	99,80%	98,40%	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SPINK1	100%	99,30%	100%	100%	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189
SPRED1	100%	98,90%	100%	100%	Legius syndrome, 611431
SQSTM1	98,80%	95,50%	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	100%	99,30%	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 Joubert syndrome 32, 617757
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	99,90%	97,30%	100%	100%	No OMIM disease ID
TERT	96,20%	94,50%	100%	100%	No OMIM disease ID
TG	100%	99,40%	100%	100%	Thyroid dyshormonogenesis 3, 274700
THPO	100%	99,50%	100%	100%	Thrombocythemia 1, 187950
TINF2	100%	100%	100%	100%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TMEM127	99,50%	96,50%	100%	100%	No OMIM disease ID
TNFRSF11A	94,60%	93,30%	99,20%	98,00%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301

TP53	99,90%	97,70%	91,70%	91,70%	Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Bone marrow failure syndrome 5, 618165 Nasopharyngeal carcinoma, somatic, 607107 Hepatocellular carcinoma, somatic, 114550
TRIP13	100%	100%	100%	100%	Mosaic variegated aneuploidy syndrome 3, 617598
TSC1	99,80%	98,70%	100%	100%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100%	99,60%	100%	100%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
USB1	100%	99,40%	100%	100%	Poikiloderma with neutropenia, 604173
VHL	96,30%	91,40%	100%	100%	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
WAS	95,90%	85,30%	100%	99,80%	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	99,90%	98,80%	100%	100%	Werner syndrome, 277700
WT1	99,90%	98,30%	100%	100%	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370
XPA	99,60%	95,60%	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 : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors