HEREDITARY CANCER GENE PANEL DG 2.16 (210 genes)

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Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
A2ML1	104,6	99.9%	99.5%	{Otitis media, susceptibility to}, 166760
ACD	159,6	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
AIP	137,2	100.0%	99.6%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
ALK	128,1	100.0%	99.4%	{Neuroblastoma, susceptibility to, 3}, 613014
ANKRD26	83,3	95.3%	90.1%	Thrombocytopenia 2, 188000
APC	141,4	99.9%	99.6%	Adenoma, periampullary, somatic, 0 Adenomatous polyposis coli, 175100 Brain tumor-polyposis syndrome 2, 175100 Colorectal cancer, somatic, 114500 Desmoid disease, hereditary, 135290 Gardner syndrome, 175100 Gastric cancer, somatic, 613659 Hepatoblastoma, somatic, 114550
ARMC5	170	100.0%	99.6%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ASXL1	132,4	100.0%	99.5%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ATM	110,9	99.6%	97.2%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0 {Breast cancer, susceptibility to}, 114480
ATR	144,6	99.8%	98.6%	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
AXIN2	124,2	100.0%	99.9%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BAP1	104,8	85.0%	82.9%	Tumor predisposition syndrome, 614327
BARD1	140,6	100.0%	99.9%	{Breast cancer, susceptibility to}, 114480
BLM	111	99.6%	98.0%	Bloom syndrome, 210900

BMPR1A	78,2	99.5%	92.9%	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BRAF	72,5	92.4%	80.2%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
BRCA1	161,4	99.1%	98.1%	Fanconi anemia, complementation group S, 617883 {Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	106,2	99.6%	98.7%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070 {Breast cancer, male, susceptibility to}, 114480 {Breast-ovarian cancer, familial, 2}, 612555 {Glioblastoma 3}, 613029 {Medulloblastoma}, 155255 {Pancreatic cancer 2}, 613347 {Prostate cancer}, 176807
BRIP1	125,8	99.7%	98.8%	Fanconi anemia, complementation group J, 609054 {Breast cancer, early-onset, susceptibility to}, 114480
BUB1	126,2	99.9%	98.6%	Colorectal cancer with chromosomal instability, somatic, 0
BUB1B	122	99.8%	98.7%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BUB3	125,3	99.8%	98.1%	No OMIM phenotype Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)
CBL	126	97.3%	97.0%	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CDC73	113,6	99.9%	98.8%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	104,5	99.2%	99.0%	Blepharocheilodontic syndrome 1, 119580 Endometrial carcinoma, somatic, 608089 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215

				Ovarian cancer, somatic, 167000 {Breast cancer, lobular}, 114480 {Prostate cancer, susceptibility to}, 176807
CDK4	100	100.0%	99.1%	{Melanoma, cutaneous malignant, 3}, 609048
CDKN1A	171,9	100.0%	100.0%	No OMIM phenotype Multiple endocrine neoplasia 1 (Agarwal (2009) J Clin Endocrinol Metab 94, 1826) {Cancer, association with} (Mousses (1995) Hum Mol Genet 4, 1089) {Breast cancer, association with} (Staalesen (2006) Clin Cancer Res 12, 6000)
CDKN1B	151,5	99.9%	99.4%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	100,1	89.8%	81.7%	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	121,7	92.3%	92.3%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, 0 Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDKN2B	121,9	100.0%	100.0%	No OMIM phenotype Renal cell carcinoma (Jafri (2015) Cancer Discov 5, 723) Multiple endocrine neoplasia 1 (Agarwal (2009) J Clin Endocrinol Metab 94, 1826) ?Melanoma (Foley (2015) EBioMedicine 2,74) ?Parathyroid adenoma (Costa-Guda (2013) Horm
CDKN2C	139,7	100.0%	100.0%	No OMIM phenotype
CEBPA	139,8	99.9%	99.1%	?Leukemia, acute myeloid, 601626 Leukemia, acute myeloid, somatic, 601626
CHEK2	88,6	83.8%	80.1%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast and colorectal cancer, susceptibility to}, 0 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807
CREBBP	110,7	99.4%	97.0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CTC1	105,5	100.0%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTNNA1	109,6	99.1%	97.2%	Macular dystrophy, patterned, 2, 608970
CTR9	142,5	100.0%	99.9%	No OMIM phenotype Wilms tumor (Hanks (2014) Nat Commun 5, 4398)
CYLD	109,2	99.7%	97.8%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606

DDB2	147,3	99.8%	98.4%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	100,7	86.7%	81.2%	Warsaw breakage syndrome, 613398
DICER1	137,9	99.8%	98.4%	GLOW syndrome, somatic mosaic, 618272 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DIS3L2	143,3	100.0%	99.8%	Perlman syndrome, 267000
DKC1	91,2	99.8%	97.7%	Dyskeratosis congenita, X-linked, 305000
DNAJC21	128,1	99.9%	99.5%	Bone marrow failure syndrome 3, 617052
EGFR	135,2	100.0%	100.0%	?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 {Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	102	97.6%	85.9%	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070
EGLN2	151,2	100.0%	100.0%	No OMIM phenotype
ELANE	141,5	100.0%	99.3%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
EPCAM	76,5	99.7%	95.7%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
ERCC1	85,7	100.0%	98.1%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	128	100.0%	99.8%	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	92	99.9%	98.4%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	132	100.0%	99.8%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965
ERCC5	126,3	99.9%	99.5%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	158,2	100.0%	99.9%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946

				UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ESR2	111,6	100.0%	99.6%	?Ovarian dysgenesis 8, 618187
ETV6	148,3	100.0%	99.4%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EXT1	88,6	99.6%	98.0%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	118	99.9%	99.1%	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EZH2	130	99.4%	97.6%	Weaver syndrome, 277590
FAN1	132,2	100.0%	99.9%	Interstitial nephritis, karyomegalic, 614817
FANCA	112,4	99.9%	98.9%	Fanconi anemia, complementation group A, 227650
FANCB	76,4	98.6%	93.2%	Fanconi anemia, complementation group B, 300514
FANCC	100,8	99.7%	99.2%	Fanconi anemia, complementation group C, 227645
FANCD2	115,6	99.1%	96.6%	Fanconi anemia, complementation group D2, 227646
FANCE	118,2	96.6%	89.9%	Fanconi anemia, complementation group E, 600901
FANCF	244,4	100.0%	100.0%	Fanconi anemia, complementation group F, 603467
FANCG	140,7	100.0%	99.8%	Fanconi anemia, complementation group G, 614082
FANCI	136,2	99.9%	98.9%	Fanconi anemia, complementation group I, 609053
FANCL	105,8	99.7%	98.0%	Fanconi anemia, complementation group L, 614083
FANCM	100,6	99.3%	97.1%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAS	226	99.9%	99.6%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0 {Autoimmune lymphoproliferative syndrome}, 601859
FH	128	95.0%	88.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLCN	152,3	100.0%	100.0%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
G6PC3	114,6	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA2	115	100.0%	99.0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626

				{Myelodysplastic syndrome, susceptibility to}, 614286
GDNF	183,5	100.0%	100.0%	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GFI1	105,7	100.0%	100.0%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, autosomal dominant, 613107
GPC3	75,7	98.7%	92.7%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GREM1	106,8	100.0%	100.0%	No OMIM phenotype {Colorectal cancer, increased risk, association with}(Peters (2012) Hum Genet 131,217) Oligosyndactyly of the hands, Cenani-Linz-like (Dimitrov (2010) J Med Genet 47,569) Mixed polyposis syndrome (Jaeger (2012) Nat Genet 44,699)
GRHL2	116,8	100.0%	100.0%	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
HABP2	109,3	100.0%	99.4%	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HAX1	137,4	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HNF1A	163,2	100.0%	99.9%	Diabetes mellitus, insulin-dependent, 20, 612520 Hepatic adenoma, somatic, 142330 MODY, type III, 600496 Renal cell carcinoma, 144700 {Diabetes mellitus, insulin-dependent}, 222100 {Diabetes mellitus, noninsulin-dependent, 2}, 125853
HOXB13	186,3	100.0%	100.0%	{Prostate cancer, hereditary, 9}, 610997
IDH1	78	89.4%	77.3%	{Glioma, susceptibility to, somatic}, 137800
IDH2	98,5	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IPMK	88	98.5%	89.6%	No OMIM phenotype Small intestinal carcinoid (Sei (2015) Gastroenterology 149,67)
KIF1B	139,6	100.0%	99.6%	?Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIT	136,2	100.0%	99.6%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, 601626 Mastocytosis, cutaneous, 154800 Mastocytosis, systemic, somatic, 154800

				Piebaldism, 172800
KLLN	152,3	100.0%	100.0%	Cowden syndrome 4, 615107
KRAS	67,2	99.4%	97.3%	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LIG4	173,4	100.0%	99.8%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LZTR1	143,6	100.0%	99.7%	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
MAP2K1	92,3	99.5%	96.3%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	124,2	98.5%	94.1%	Cardiofaciocutaneous syndrome 4, 615280
MAX	80	99.9%	98.0%	{Pheochromocytoma, susceptibility to}, 171300
MDH2	109,4	98.0%	97.9%	Epileptic encephalopathy, early infantile, 51, 617339
MEN1	132	100.0%	99.5%	Adrenal adenoma, somatic, 0 Angiofibroma, somatic, 0 Carcinoid tumor of lung, 0 Lipoma, somatic, 0 Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic, 0
MET	151,3	99.9%	99.3%	?Deafness, autosomal recessive 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 {Osteofibrous dysplasia, susceptibility to}, 607278
MITF	141,1	100.0%	99.8%	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456

MLH1	139,2	99.9%	99.3%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MPL	125,8	100.0%	99.8%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MRE11	49,7	97.3%	86.0%	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	111,7	99.4%	96.4%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	139,6	99.8%	99.2%	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4, 617100
MSH6	165,1	100.0%	100.0%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome, 276300 {Endometrial cancer, familial}, 608089
MUC5B	85,5	82.9%	72.7%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
MUTYH	152	100.0%	100.0%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
NBN	93,8	99.8%	98.4%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NF1	106,2	92.5%	89.4%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	94,2	100.0%	99.6%	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, somatic, 162091
NHP2	121,9	100.0%	99.2%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	120,5	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	65	94.5%	83.5%	Leukemia, acute myeloid, somatic, 601626
NRAS	145,5	100.0%	100.0%	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550

				Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NSD1	147	100.0%	99.8%	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NTHL1	121,6	100.0%	100.0%	Familial adenomatous polyposis 3, 616415
PALB2	143,5	100.0%	99.9%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PARN	127,3	99.9%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	105,6	99.2%	96.0%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PDGFB	115,4	100.0%	100.0%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRA	124,7	100.0%	100.0%	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PHOX2B	145,5	100.0%	100.0%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PIK3CA	127,7	100.0%	99.8%	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PMS2	94,7	83.4%	81.0%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMS2CL				No OMIM phenotype
POLD1	124,5	98.0%	93.9%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381

				{Colorectal cancer, susceptibility to, 10}, 612591
POLE	126,9	99.9%	99.4%	FILS syndrome, 615139 IMAGE-I syndrome, 618336 {Colorectal cancer, susceptibility to, 12}, 615083
POLH	116,2	99.9%	98.6%	Xeroderma pigmentosum, variant type, 278750
POT1	97,7	99.9%	98.5%	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
POU6F2	135,1	100.0%	100.0%	{Wilms tumor susceptibility-5}, 601583
PPM1D	170,2	100.0%	99.6%	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450
PRF1	138,1	91.2%	90.6%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKAR1A	79,4	98.6%	92.6%	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRKN	82,1	79.9%	78.1%	Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 Parkinson disease, juvenile, type 2, 600116
PRSS1	141,4	100.0%	99.9%	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044
PTCH1	110,2	99.9%	98.4%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	120,3	99.9%	98.7%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTEN	129,7	99.6%	97.0%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174
PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250

				Noonan syndrome 1, 163950
RAD50	102	97.5%	91.1%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	140,6	99.9%	99.5%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD51D	140,9	100.0%	99.4%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RAF1	108,3	100.0%	99.9%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RB1	89,8	97.8%	93.1%	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RECQL	144,1	99.8%	98.7%	No OMIM phenotype Breast cancer (Cybulski (2015) Nat Genet 47,643)
RECQL4	159,9	100.0%	99.8%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REST	115,5	98.5%	98.4%	Fibromatosis, gingival, 5, 617626 {Wilms tumor 6, susceptibility to}, 616806
RET	136,8	100.0%	99.2%	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, protection against}, 142623 {Hirschsprung disease, susceptibility to, 1}, 142623
RHBDF2	105,1	99.9%	98.9%	Tylosis with esophageal cancer, 148500
RINT1	158	99.8%	98.2%	No OMIM phenotype ?Breast cancer (Park (2014) Cancer Discov 4, 804)
RIT1	139,2	100.0%	100.0%	Noonan syndrome 8, 615355
RNF43	145,9	100.0%	98.8%	Sessile serrated polyposis cancer syndrome, 617108
RPL11	85,4	100.0%	99.3%	Diamond-Blackfan anemia 7, 612562
RPL35A	75,4	96.4%	84.6%	Diamond-Blackfan anemia 5, 612528
RPL5	34,7	85.0%	67.7%	Diamond-Blackfan anemia 6, 612561
RPS10	91,8	98.8%	91.8%	Diamond-Blackfan anemia 9, 613308
RPS17	38,2	87.0%	68.9%	Diamond-Blackfan anemia 4, 612527

RPS19	76,7	99.9%	96.6%	Diamond-Blackfan anemia 1, 105650
RPS20	58,1	97.9%	88.8%	No OMIM phenotype Colorectal cancer, non-polyposis (Nieminen (2014) Gastroenterology 147,595)
RPS24	84,5	95.2%	89.7%	Diamond-blackfan anemia 3, 610629
RPS26	75,9	89.2%	75.8%	Diamond-Blackfan anemia 10, 613309
RPS7	76,6	84.8%	70.0%	Diamond-Blackfan anemia 8, 612563
RTEL1	131,1	99.7%	97.7%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RUNX1	84,6	99.6%	96.3%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SAMD9	163,9	100.0%	99.9%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	171,8	100.0%	100.0%	Ataxia-pancytopenia syndrome, 159550
SBDS	166,2	100.0%	100.0%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SDHA	88,9	85.1%	77.7%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF2	127,7	95.6%	94.6%	Paragangliomas 2, 601650
SDHB	114,8	100.0%	99.9%	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHC	85,8	99.6%	95.3%	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	43,7	52.7%	50.6%	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SEMA4A	124,3	100.0%	99.3%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SFTPA1	155,5	100.0%	100.0%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
SFTPA2	141,1	100.0%	100.0%	Pulmonary fibrosis, idiopathic, 178500
SH2B3	108,4	99.9%	97.6%	Erythrocytosis, somatic, 133100

				Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SHOC2	139,6	99.9%	99.4%	Noonan-like syndrome with loose anagen hair, 607721
SLX4	124,2	100.0%	99.7%	Fanconi anemia, complementation group P, 613951
SMAD4	108,9	100.0%	99.9%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD9	110	100.0%	100.0%	Pulmonary hypertension, primary, 2, 615342
SMARCA4	150,9	100.0%	99.4%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCB1	179,1	100.0%	99.9%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid tumor predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCE1	66,5	94.4%	84.2%	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174
SOS1	102	99.6%	97.4%	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SPINK1	85	100.0%	99.4%	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPRED1	146,5	99.8%	98.8%	Legius syndrome, 611431
SQSTM1	117,8	99.9%	99.2%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250
STK11	131	100.0%	100.0%	Melanoma, malignant, somatic, 0 Pancreatic cancer, somatic, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
SUFU	132,8	100.0%	99.9%	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
TERC				Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743

				{Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERF2IP	128,4	100.0%	99.6%	No OMIM phenotype Melanoma (Aoude (2015) J Natl Cancer Inst 107) Chronic lymphocytic leukaemia (Speedy (2016) Blood 128,2319)
TERT	144,1	99.7%	97.6%	{Dyskeratosis congenita, autosomal dominant 2}, 613989 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742
TINF2	177,1	100.0%	100.0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMEM127	112	100.0%	99.8%	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	131	96.1%	95.2%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TP53	89,2	99.8%	98.5%	Bone marrow failure syndrome 5, 618165 Breast cancer, somatic, 114480 Hepatocellular carcinoma, somatic, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, somatic, 607107 Pancreatic cancer, somatic, 260350 {Adrenocortical carcinoma, pediatric}, 202300 {Basal cell carcinoma 7}, 614740 {Choroid plexus papilloma}, 260500 {Colorectal cancer}, 114500 {Glioma susceptibility 1}, 137800 {Osteosarcoma}, 259500
TRIP13	127,4	100.0%	99.9%	Mosaic variegated aneuploidy syndrome 3, 617598
TSC1	112,5	99.6%	98.2%	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	140,5	100.0%	99.9%	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
USB1	118,2	99.8%	97.2%	Poikiloderma with neutropenia, 604173
VHL	169,6	100.0%	98.3%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300

				Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
WAS	70,4	94.2%	83.6%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WRAP53	162,8	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	124,8	99.7%	98.8%	Werner syndrome, 277700
WT1	90,1	100.0%	99.3%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
XPA	74,7	99.7%	98.2%	Xeroderma pigmentosum, group A, 278700
XPC	143,5	100.0%	99.8%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors