HEREDITARY CANCER GENE PANEL DG 2.12 (186 genes)

Gene	*Median*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACD	135.2	100	98	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
AIP	154.5	99	99	Pituitary adenoma, ACTH-secreting, 219090 Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634
ALK	129	99	98	{Neuroblastoma, susceptibility to, 3}, 613014
ANKRD26	79.7	89	77	Thrombocytopenia 2, 188000
APC	159	99	98	Adenoma, periampullary, somatic 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	136.7	99	97	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ATM	109.8	99	94	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
ATR	138.2	99	96	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	117.5	100	99	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BAP1	135.5	99	98	Tumor predisposition syndrome, 614327
BARD1	149.7	100	99	{Breast cancer, susceptibility to}, 114480
BLM	116.3	99	96	Bloom syndrome, 210900

BMPR1A	98	99	94	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BRCA1	179.4	98	96	{Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	102.6	99	97	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	117.8	99	97	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BUB1	136.4	99	97	Colorectal cancer with chromosomal instability, somatic
BUB1B	136.4	98	97	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BUB3	146.3	98	97	No OMIM phenotype Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)
CDC73	102.7	99	97	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	125.3	99	99	Endometrial carcinoma, somatic, 608089 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 {Prostate cancer, susceptibility to}, 176807
CDK4	128	100	99	{Melanoma, cutaneous malignant, 3}, 609048

CDKN1A	137.3	100	100	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) ?Parathyroid adenoma (Costa-Guda (2013) Horm Cancer 4, 301) ?Systemic lupus erythematosus, increased risk, association with (Kim (2009) Genes Immun 10, 482)
CDKN1B	93.2	100	99	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	21.3	68	51	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	87.5	93	92	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDKN2B	86.3	100	99	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 Cancer 4, 301)
CDKN2C	153.4	100	100	No OMIM phenotype
CEBPA	46.8	75	65	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CHEK2	102.4	83	80	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast and colorectal cancer, susceptibility to} {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807
CREBBP	123.5	99	96	Rubinstein-Taybi syndrome, 180849
CTC1	119	100	99	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTNNA1	125.7	99	99	Macular dystrophy,patterned,608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CTR9	154.3	100	99	No OMIM phenotype Wilms tumor (Hanks (2014) Nat Commun 5, 4398)
CYLD	119.8	98	93	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606

DDB2	162.8	100	99	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	107.4	80	76	Warsaw breakage syndrome, 613398
DICER1	145.4	99	98	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DKC1	112.6	99	98	Dyskeratosis congenita, X-linked, 305000
DNAJC21	123.4	99	97	Bone marrow failure syndrome 3, 617052
EGFR	161.7	100	99	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 ?Inflammatory skin and bowel disease, neonatal, 2, 616069 {Nonsmall cell lung cancer, susceptibility to}, 211980
ELANE	80.9	99	95	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ERCC1	76.6	100	97	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	123.8	100	99	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	113.3	99	98	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	139.2	100	99	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 ?XFE progeroid syndrome, 610965
ERCC5	139.8	100	99	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	170.4	100	99	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	134	100	99	No OMIM phenotype Medullary thyroid carcinoma (Smith (2016) Hum Mol Genet 25,1836) ?Primary amenorrhea (Asadi (2013) Clin Genet 83,497) ?Breast cancer, increased risk (Pylkas (2012) PLoS Genet 8,e1002734 {Ovulatory defects, association with} (Sundarrajan (2001) J Clin Endocrinol Metab 86,135) {Hyposadias,association with} (Beleza-Meireles (2006) J Endocrinol Invest 29,5)
ETV6	140.3	100	99	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EXT1	105.5	99	98	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	163.6	99	99	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682
EZH2	139.1	99	97	Weaver syndrome, 277590
FAN1	144.1	100	99	Interstitial nephritis, karyomegalic, 614817
FANCA	123.5	99	98	Fanconi anemia, complementation group A, 227650
FANCB	68.8	96	88	Fanconi anemia, complementation group B, 300514
FANCC	116.5	99	96	Fanconi anemia, complementation group C, 227645
FANCD2	128.1	98	95	Fanconi anemia, complementation group D2, 227646
FANCE	108.1	86	84	Fanconi anemia, complementation group E, 600901
FANCF	166.9	100	100	Fanconi anemia, complementation group F, 603467
FANCG	147.9	100	100	Fanconi anemia, complementation group G, 614082
FANCI	152.2	99	97	Fanconi anemia, complementation group I, 609053
FANCL	87.9	99	94	Fanconi anemia, complementation group L, 614083
FANCM	96.7	99	94	No OMIM phenotype Fanconi anemia, complementation group M, 614087
FAS	271.9	100	99	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic {Autoimmune lymphoproliferative syndrome}, 601859
FH	146.1	91	87	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLCN	160.8	100	99	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700

G6PC3	124	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA2	119.6	99	98	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GDNF	186.1	99	98	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GFI1	83.1	99	92	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GPC3	85.8	98	92	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GREM1	95.3	100	100	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	134.6	100	100	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
HABP2	136.3	100	99	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HAX1	136.6	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HNF1A	148.4	100	99	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	127.9	99	97	No OMIM phenotype {Prostate cancer, increased risk} (Lin (2013) Prostate 73, 169)
IPMK	95.5	96	83	No OMIM phenotype Small intestinal carcinoid (Sei (2015) Gastroenterology 149,67)
KIF1B	154.8	100	99	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700

KIT	152.9	100	99	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800
KLLN	117.5	100	100	Cowden syndrome 4, 615107
LIG4	165.3	100	99	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LZTR1	134.3	100	99	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MAX	86.8	99	96	{Pheochromocytoma, susceptibility to}, 171300
MDH2	123.5	98	97	Epileptic encephalopathy, early infantile, 51, 617339
MEN1	123.4	99	96	Adrenal adenoma, somatic Angiofibroma, somatic Carcinoid tumor of lung Lipoma, somatic Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic
MET	184.5	100	99	Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 ?Deafness, autosomal recessive 97, 616705 {Osteofibrous dysplasia,susceptibility to},607278
MITF	155.2	100	99	Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MLH1	162.3	100	99	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MPL	135.2	99	97	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MRE11A	51.2	95	82	Ataxia-telangiectasia-like disorder, 604391

MSH2	113.4	98	93	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	113.4	98	95	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4,617100
MSH6	171.5	100	99	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MUC5B	98.7	87	82	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
MUTYH	165.2	100	99	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
NBN	80.5	99	94	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NF1	129.8	93	90	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	100.3	100	99	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, 162091
NHP2	111.1	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	160.6	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	70.1	88	77	Leukemia, acute myeloid, somatic, 601626
NSD1	155.4	100	100	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NTHL1	98.6	99	95	Familial adenomatous polyposis 3, 616415
OGG1	128.7	100	99	Renal cell carcinoma, clear cell, somatic, 144700
PALB2	152.6	100	99	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348

PARK2	123.7	99	98	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PARN	128.3	99	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	118.9	98	95	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PHOX2B	93	93	87	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PIK3CA	120.5	99	99	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PMS2	95.2	83	80	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMS2CL	NC	NC	NC	No OMIM phenotype
POLD1	101.3	93	90	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	144.3	100	99	FILS syndrome, 615139 {Colorectal cancer, susceptibility to, 12}, 615083
POLH	140.7	100	99	Xeroderma pigmentosum, variant type, 278750
POT1	90.6	99	95	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
POU6F2	141.1	98	98	{Wilms tumor susceptibility-5}, 601583
PPM1D	168.4	100	99	Breast cancer, 114480

PRF1	134.2	100	99	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKAR1A	90.6	99	93	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRSS1	191.1	100	99	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044
PTCH1	114.8	98	95	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCH2	120.4	99	97	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, 155255
PTEN	142.9	99	95	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 PTEN hamartoma tumor syndrome Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
RAD50	99	92	86	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	143.5	100	98	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD51D	154.1	100	99	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291

RB1	88	90	76	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RECQL	127.3	99	96	No OMIM phenotype Breast cancer (Cybulski (2015) Nat Genet 47,643)
RECQL4	149.8	99	96	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REST	130.7	100	100	{Wilms tumor 6, susceptibility to}, 616806
RET	141.3	99	97	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, susceptibility to, 1}, 142623
RHBDF2	98	99	97	Tylosis with esophageal cancer, 148500
RINT1	171.1	99	97	No OMIM phenotype ?Breast cancer (Park (2014) Cancer Discov 4, 804)
RNF43	126.6	100	99	Sessile serrated polyposis cancer syndrome, 617108
RPL11	100	100	99	Diamond-Blackfan anemia 7, 612562
RPL35A	84	99	91	Diamond-Blackfan anemia 5, 612528
RPL5	43.7	81	69	Diamond-Blackfan anemia 6, 612561
RPS10	140.1	99	97	Diamond-Blackfan anemia 9, 613308
RPS17	52.5	87	74	Diamond-Blackfan anemia 4, 612527
RPS19	82.6	99	95	Diamond-Blackfan anemia 1, 105650
RPS20	74.6	99	94	No OMIM phenotype
RPS24	111.3	92	87	Diamond-blackfan anemia 3, 610629
RPS26	107	94	82	Diamond-Blackfan anemia 10, 613309
RPS7	93.5	76	63	Diamond-Blackfan anemia 8, 612563
RTEL1	115.2	99	95	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373

RUNX1	91.6	94	86	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SBDS	212.4	100	99	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SDHA	122.2	84	80	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF2	144.1	94	94	Paragangliomas 2, 601650
SDHB	120.5	100	99	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHC	100	99	96	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	48.5	62	57	Carcinoid tumors, intestinal, 114900 Cowden syndrome 3, 615106 Merkel cell carcinoma, somatic Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SEMA4A	127.6	99	98	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SFTPA1	175.4	99	98	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
SFTPA2	174.3	100	100	Pulmonary fibrosis, idiopathic, 178500
SH2B3	97.5	90	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SLX4	114.4	100	99	Fanconi anemia, complementation group P, 613951

SMAD4	121.2	99	98	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD9	132.7	100	100	Pulmonary hypertension,primary,615342 Polyposis & gastrointestinal ganglioneuromas (Ngeow (2015) Gastroenterology 149,886)
SMARCA4	144.8	100	99	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCB1	214.6	100	100	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCE1	73.6	96	86	Coffin-Siris syndrome 5,616938 {Meningioma, familial, susceptibility to}, 607174
SPINK1	80.2	100	99	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SQSTM1	110.3	98	95	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
STK11	112.2	99	95	Melanoma, malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
SUFU	122.7	100	99	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERF2IP	116.8	100	97	No OMIM phenotype Melanoma (Aoude (2015) J Natl Cancer Inst 107)

TERT	138.6	95	92	{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	183.9	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMEM127	107.5	98	94	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	147	93	91	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TP53	92.1	99	98	Adrenal cortical carcinoma, 202300 Breast cancer, 114480 Choroid plexus papilloma, 260500 Colorectal cancer, 114500 Hepatocellular carcinoma, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, 607107 Osteosarcoma, 259500 Pancreatic cancer, 260350 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800
TSC1	128.9	99	98	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	131.3	100	99	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
USB1	125	99	98	Poikiloderma with neutropenia,604173
VHL	119.6	92	85	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300

WAS	66.6	88	78	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WRAP53	154.5	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	123.6	98	94	Werner syndrome, 277700
WT1	85.3	93	86	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
XPA	52.8	98	88	Xeroderma pigmentosum, group A, 278700
XPC	140.8	100	99	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 : April 14th, 2017.
This list is accurate for panel version DG 2.12
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors