HEREDITARY CANCER GENE PANEL DG 2.7/DG 2.8

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACD	130.8	99%	97%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ALK	147.9	99%	97%	{Neuroblastoma, susceptibility to, 3}, 613014
ANKRD26	89.4	88%	78%	Thrombocytopenia 2, 188000
APC	175.4	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	130	99%	98%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ATM	124.3	98%	93%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
ATR	160.3	98%	96%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BAP1	154.2	99%	98%	Tumor predisposition syndrome, 614327
BARD1	170	99%	99%	{Breast cancer, susceptibility to}, 114480
BLM	139.6	98%	95%	Bloom syndrome, 210900
BMPR1A	116.8	99%	95%	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BRCA1	210.6	98%	96%	{Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	107.9	98%	96%	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	137.1	99%	96%	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BUB1	155	99%	97%	Colorectal cancer with chromosomal instability, somatic
BUB1B	164.8	98%	97%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BUB3	159.9	98%	94%	No OMIM phenotype Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)
CDC73	111.9	99%	95%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	129.6	99%	98%	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	137	100%	99%	{Melanoma, cutaneous malignant, 3}, 609048
CDKN2A	77.7	92%	90%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CEBPA	53.3	82%	69%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CENPJ	163.1	99%	97%	?Seckel syndrome 4, 613676 Microcephaly 6, primary, autosomal recessive, 608393
CHEK2	114.7	82%	79%	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	147.6	99%	96%	Rubinstein-Taybi syndrome, 180849
CTC1	118.9	99%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTNNA1	148.9	99%	98%	Macular dystrophy,patterned,608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CTR9	167.2	100%	99%	No OMIM phenotype Wilms tumor (Hanks (2014) Nat Commun 5, 4398)
CYLD	135.1	97%	93%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
DDB2	173.8	100%	99%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	99.2	80%	73%	Warsaw breakage syndrome, 613398
DICER1	168.2	99%	98%	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DKC1	138.1	99%	98%	Dyskeratosis congenita, X-linked, 305000
EGFR	172.2	99%	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	103.8	99%	97%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ERCC1	95.6	99%	98%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	143.5	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	117.6	99%	98%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	168.4	99%	98%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 ?XFE progeroid syndrome, 610965

ERCC5	161.6	99%	99%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	192.3	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	152.8	99%	98%	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	145.6	100%	99%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EXT1	106.1	99%	97%	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	197.4	99%	99%	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682
EZH2	153.3	99%	98%	Weaver syndrome, 277590
FAN1	167.4	100%	99%	Interstitial nephritis, karyomegalic, 614817
FANCA	130.4	99%	98%	Fanconi anemia, complementation group A, 227650
FANCB	78.3	95%	86%	Fanconi anemia, complementation group B, 300514
FANCC	123.2	98%	96%	Fanconi anemia, complementation group C, 227645
FANCD2	142.7	98%	95%	Fanconi anemia, complementation group D2, 227646
FANCE	117	88%	85%	Fanconi anemia, complementation group E, 600901
FANCF	158.8	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	149.5	99%	99%	Fanconi anemia, complementation group G, 614082
FANCI	182	99%	97%	Fanconi anemia, complementation group I, 609053
FANCL	86	98%	93%	Fanconi anemia, complementation group L, 614083
FANCM	108.9	97%	92%	No OMIM phenotype

				Fanconi anemia, complementation group M, 614087
FH	175.4	92%	88%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLCN	172.1	99%	98%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
G6PC3	138.4	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GDNF	199.9	99%	97%	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GFI1	99.2	98%	92%	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GPC3	121	98%	94%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GREM1	118	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)
HABP2	158.4	100%	99%	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HAX1	148	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HNF1A	154.1	99%	98%	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	142.9	99%	97%	No OMIM phenotype {Prostate cancer, increased risk} (Lin (2013) Prostate 73, 169)
IPMK	100.7	95%	83%	No OMIM phenotype Small intestinal carcinoid (Sei (2015) Gastroenterology 149,67)
KIF1B	177	99%	99%	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210

				{Neuroblastoma, susceptibility to, 1}, 256700
KIT	175	100%	99%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800
KLLN	121.9	100%	99%	Cowden syndrome 4, 615107
LZTR1	157.8	99%	98%	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MAX	96.4	99%	96%	{Pheochromocytoma, susceptibility to}, 171300
MDH2	127.3	98%	97%	No OMIM phenotype Paraganglioma (Cascon (2015) J Natl Cancer Inst 107,djv053)
MEN1	133.5	98%	95%	Adrenal adenoma, somatic Angiofibroma, somatic Carcinoid tumor of lung Lipoma, somatic Multiple endocrine neoplasia 1, 131100 Parathyroid adenoma, somatic
MET	216.4	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	163	99%	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	188.5	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MPL	156.7	99%	97%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MRE11A	57.6	95%	85%	Ataxia-telangiectasia-like disorder, 604391
MSH2	122	98%	92%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300

				Muir-Torre syndrome, 158320
MSH3	126.5	98%	94%	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4,617100
MSH6	183.6	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MUC5B	107.3	87%	82%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
MUTYH	164.2	99%	99%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
NBN	89.5	98%	94%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NF1	140.5	93%	89%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	118.2	100%	100%	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, 162091
NHP2	87.3	99%	98%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	189.4	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NSD1	172.3	100%	99%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NTHL1	113.6	98%	94%	Familial adenomatous polyposis 3, 616415
OGG1	149.1	99%	99%	Renal cell carcinoma, clear cell, somatic, 144700
PALB2	180.1	100%	99%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PARK2	134.2	99%	98%	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116

				{Leprosy, susceptibility to}, 607572
PARN	140.4	99%	97%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	127.8	98%	96%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PHOX2B	111.3	94%	90%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PMS2	95.9	83%	80%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMS2CL				No OMIM phenotype
POLD1	108.5	94%	91%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	160	99%	99%	FILS syndrome, 615139 {Colorectal cancer, susceptibility to, 12}, 615083
POT1	109	99%	96%	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
POU6F2	131.3	98%	98%	{Wilms tumor susceptibility-5}, 601583
PPM1D	192	99%	98%	Breast cancer, 114480
PRF1	130.4	100%	99%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKAR1A	102.8	97%	91%	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
PTCH1	138.6	98%	96%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTEN	152.4	99%	98%	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
PTPRJ	194.2	97%	96%	Colon cancer, somatic, 114500
RAD50	106.1	92%	85%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	164	99%	99%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD51D	177.3	100%	99%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RB1	90.9	88%	76%	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RECQL	145.1	99%	95%	No OMIM phenotype Breast cancer (Cybulski (2015) Nat Genet 47,643)
RECQL4	150	98%	97%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REST	146.8	100%	100%	{Wilms tumor 6, susceptibility to}, 616806
RET	163.1	99%	98%	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
RINT1	200.2	99%	97%	No OMIM phenotype ?Breast cancer (Park (2014) Cancer Discov 4, 804)
RPL11	96.1	100%	99%	Diamond-Blackfan anemia 7, 612562
RPL35A	75.2	95%	84%	Diamond-Blackfan anemia 5, 612528
RPL5	39.9	80%	61%	Diamond-Blackfan anemia 6, 612561

RPS10	129.6	98%	93%	Diamond-Blackfan anemia 9, 613308
RPS17	51.3	83%	69%	Diamond-Blackfan anemia 4, 612527
RPS19	88.3	99%	96%	Diamond-Blackfan anemia 1, 105650
RPS24	116.4	96%	90%	Diamond-blackfan anemia 3, 610629
RPS26	92.9	92%	82%	Diamond-Blackfan anemia 10, 613309
RPS7	112.4	84%	69%	Diamond-Blackfan anemia 8, 612563
RTEL1	127.4	99%	96%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RUNX1	120.5	96%	92%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SBDS	210.4	99%	99%	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SDHA	117.4	84%	78%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF2	163.2	94%	93%	Paragangliomas 2, 601650
SDHB	144	99%	99%	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHC	104.2	99%	94%	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	59.9	62%	58%	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	139.7	99%	98%	Cone-rod dystrophy 10, 610283

				Retinitis pigmentosa 35, 610282
SFTPA1	162	99%	98%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
SFTPA2	159	100%	100%	Pulmonary fibrosis, idiopathic, 178500
SLX4	127.4	100%	99%	Fanconi anemia, complementation group P, 613951
SMAD4	132.5	99%	98%	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD9	159.8	100%	99%	Pulmonary hypertension,primary,615342 Polyposis & gastrointestinal ganglioneuromas (Ngeow (2015) Gastroenterology 149,886)
SMARCA4	156.7	99%	98%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCB1	248.9	100%	100%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SQSTM1	130.7	98%	94%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
STK11	130.1	99%	94%	Melanoma, malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
SUFU	149.5	99%	97%	Basal cell nevus syndrome, 109400 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	127.3	99%	97%	No OMIM phenotype Melanoma (Aoude (2015) J Natl Cancer Inst 107)
TERT	148.3	96%	91%	{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	196.8	100%	99%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMEM127	112.6	97%	94%	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	144.8	93%	91%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TP53	103.8	99%	94%	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	149.6	99%	98%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	144.5	99%	98%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
USB1	144.5	99%	97%	Poikiloderma with neutropenia,604173
VHL	120.5	95%	88%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
WAS	68.7	87%	78%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WRAP53	164.3	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	143.3	97%	93%	Werner syndrome, 277700

WT1	100	95%	89%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
XPA	58.3	95%	84%	Xeroderma pigmentosum, group A, 278700
XPC	163.9	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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors