HEREDITARY CANCER GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACD	*139.5*	*100%*	*99%*	*?Dyskeratosis congenita, autosomal recessive 7, 616553* *?Dyskeratosis congenita, autosomal dominant 6, 616553*
ALK	*143.8*	*99%*	*98%*	*{Neuroblastoma, susceptibility to, 3}, 613014*
ANKRD26	*99.3*	*93%*	*85%*	*Thrombocytopenia 2, 188000*
APC	*186.4*	*100%*	*99%*	*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	*145.7*	*100%*	*99%*	*ACTH-independent macronodular adrenal hyperplasia 2, 615954*
ATM	*132.3*	*99%*	*96%*	*Ataxia-telangiectasia, 208900* *Lymphoma, B-cell non-Hodgkin, somatic* *Lymphoma, mantle cell, somatic* *T-cell prolymphocytic leukemia, somatic* *{Breast cancer, susceptibility to}, 114480*
ATR	*175.1*	*99%*	*98%*	*Seckel syndrome 1, 210600* *?Cutaneous telangiectasia and cancer syndrome, familial, 614564*
BAP1	*153.1*	*99%*	*98%*	*Tumor predisposition syndrome, 614327*
BARD1	*186.8*	*100%*	*99%*	*{Breast cancer, susceptibility to}, 114480*
BLM	*139.5*	*99%*	*97%*	*Bloom syndrome, 210900*
BMPR1A	*115.4*	*99%*	*97%*	*Juvenile polyposis syndrome, infantile form, 174900* *Polyposis syndrome, hereditary mixed, 2, 610069* *Polyposis, juvenile intestinal, 174900*
BRCA1	*223.9*	*99%*	*97%*	*{Breast-ovarian cancer, familial, 1}, 604370* *{Pancreatic cancer, susceptibility to, 4}, 614320*
BRCA2	*118.6*	*99%*	*98%*	*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	*149*	*99%*	*98%*	*Breast cancer, early-onset, 114480* *Fanconi anemia, complementation group J, 609054*
BUB1	*172.6*	*100%*	*99%*	*Colorectal cancer with chromosomal instability, somatic*
BUB1B	*158.2*	*98%*	*98%*	*Colorectal cancer, somatic, 114500* *Mosaic variegated aneuploidy syndrome 1, 257300* *[Premature chromatid separation trait], 176430*
BUB3	*167.4*	*99%*	*97%*	*No OMIM phenotype* *Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)*
CDC73	*122.3*	*99%*	*98%*	*Hyperparathyroidism, familial primary, 145000* *Hyperparathyroidism-jaw tumor syndrome, 145001* *Parathyroid adenoma with cystic changes, 145001* *Parathyroid carcinoma, 608266*
CDH1	*129.3*	*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	*128.7*	*100%*	*100%*	*{Melanoma, cutaneous malignant, 3}, 609048*
CDKN2A	*80*	*93%*	*92%*	*Melanoma and neural system tumor syndrome, 155755* *Orolaryngeal cancer, multiple,* *Pancreatic cancer/melanoma syndrome, 606719* *{Melanoma, cutaneous malignant, 2}, 155601*
CEBPA	*55.7*	*86%*	*71%*	*Leukemia, acute myeloid, somatic, 601626* *?Leukemia, acute myeloid, 601626*
CHEK2	*115*	*84%*	*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	*141.3*	*98%*	*96%*	*Rubinstein-Taybi syndrome, 180849*
CTC1	*122.4*	*99%*	*99%*	*Cerebroretinal microangiopathy with calcifications and cysts, 612199*
CTNNA1	*155.3*	*99%*	*99%*	*Macular dystrophy,patterned,608970* *Gastric cancer, diffuse (Majewski (2012) J Pathol epub)*
CTR9	*177.1*	*100%*	*100%*	*No OMIM phenotype* *Wilms tumor (Hanks (2014) Nat Commun 5, 4398)*
CYLD	*132.2*	*99%*	*95%*	*Brooke-Spiegler syndrome, 605041* *Cylindromatosis, familial, 132700* *Trichoepithelioma, multiple familial, 1, 601606*
DDB2	*181.7*	*100%*	*99%*	*Xeroderma pigmentosum, group E, DDB-negative subtype, 278740*
DDX11	*104.7*	*79%*	*74%*	*Warsaw breakage syndrome, 613398*
DICER1	*172*	*99%*	*98%*	*Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800* *Pleuropulmonary blastoma, 601200* *Rhabdomyosarcoma, embryonal, 2, 180295*
DKC1	*122.5*	*100%*	*98%*	*Dyskeratosis congenita, X-linked, 305000*
DNAJC21	*150.2*	*99%*	*98%*	*Bone marrow failure syndrome 3, 617052*
EGFR	*178.5*	*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	*115.1*	*99%*	*98%*	*Neutropenia, cyclic, 162800* *Neutropenia, severe congenital 1, autosomal dominant, 202700*
ERCC1	*102*	*99%*	*96%*	*Cerebrooculofacioskeletal syndrome 4, 610758*
ERCC2	*143*	*100%*	*99%*	*Cerebrooculofacioskeletal syndrome 2, 610756* *Trichothiodystrophy 1, photosensitive, 601675* *Xeroderma pigmentosum, group D, 278730*
ERCC3	*120.8*	*100%*	*99%*	*Trichothiodystrophy 2, photosensitive, 616390* *Xeroderma pigmentosum, group B, 610651*
ERCC4	*165.7*	*99%*	*99%*	*Fanconi anemia, complementation group Q, 615272* *Xeroderma pigmentosum, group F, 278760* *Xeroderma pigmentosum, type F/Cockayne syndrome, 278760* *?XFE progeroid syndrome, 610965*
ERCC5	*151.3*	*100%*	*99%*	*Cerebrooculofacioskeletal syndrome 3, 616570*

				Xeroderma pigmentosum, group G, 278780 *Xeroderma pigmentosum, group G/Cockayne syndrome, 278780*
ERCC6	*196.3*	*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	*149.1*	*99%*	*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	*152.2*	*100%*	*100%*	*Leukemia, acute myeloid, somatic, 601626* *Thrombocytopenia 5, 616216*
EXT1	*105.5*	*99%*	*97%*	*Chondrosarcoma, 215300* *Exostoses, multiple, type 1, 133700*
EXT2	*178.8*	*99%*	*99%*	*Exostoses, multiple, type 2, 133701* *?Seizures, scoliosis, and macrocephaly syndrome, 616682*
EZH2	*166.9*	*99%*	*98%*	*Weaver syndrome, 277590*
FAN1	*168.9*	*100%*	*99%*	*Interstitial nephritis, karyomegalic, 614817*
FANCA	*129.9*	*99%*	*98%*	*Fanconi anemia, complementation group A, 227650*
FANCB	*84.3*	*97%*	*90%*	*Fanconi anemia, complementation group B, 300514*
FANCC	*118.8*	*99%*	*98%*	*Fanconi anemia, complementation group C, 227645*
FANCD2	*156.1*	*99%*	*96%*	*Fanconi anemia, complementation group D2, 227646*
FANCE	*127.7*	*88%*	*85%*	*Fanconi anemia, complementation group E, 600901*
FANCF	*179.5*	*100%*	*100%*	*Fanconi anemia, complementation group F, 603467*
FANCG	*158*	*100%*	*99%*	*Fanconi anemia, complementation group G, 614082*
FANCI	*174.9*	*99%*	*98%*	*Fanconi anemia, complementation group I, 609053*
FANCL	*105.1*	*99%*	*97%*	*Fanconi anemia, complementation group L, 614083*
FANCM	*117*	*99%*	*96%*	*No OMIM phenotype* *Fanconi anemia, complementation group M, 614087*

FAS	*294.7*	*100%*	*99%*	*Autoimmune lymphoproliferative syndrome, type IA, 601859* *Squamous cell carcinoma, burn scar-related, somatic* *{Autoimmune lymphoproliferative syndrome}, 601859*
FH	*183.5*	*93%*	*89%*	*Fumarase deficiency, 606812* *Leiomyomatosis and renal cell cancer, 150800*
FLCN	*176.6*	*100%*	*99%*	*Birt-Hogg-Dube syndrome, 135150* *Colorectal cancer, somatic, 114500* *Pneumothorax, primary spontaneous, 173600* *Renal carcinoma, chromophobe, somatic, 144700*
G6PC3	*143.5*	*100%*	*100%*	*Dursun syndrome, 612541* *Neutropenia, severe congenital 4, autosomal recessive, 612541*
GDNF	*213.9*	*99%*	*98%*	*Central hypoventilation syndrome, 209880* *{Hirschsprung disease, susceptibility to, 3}, 613711* *{Pheochromocytoma, modifier of}, 171300*
GFI1	*99.4*	*99%*	*96%*	*Neutropenia, nonimmune chronic idiopathic, of adults, 607847* *Neutropenia, severe congenital 2, autosomal dominant, 613107*
GPC3	*106.4*	*98%*	*94%*	*Simpson-Golabi-Behmel syndrome, type 1, 312870* *Wilms tumor, somatic, 194070*
GREM1	*113.3*	*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	*152.8*	*100%*	*100%*	*Deafness, autosomal dominant 28, 608641* *Ectodermal dysplasia/short stature syndrome, 616029*
HABP2	*148.6*	*100%*	*99%*	*{?Thyroid cancer, nonmedullary, 5}, 616535* *{Venous thromboembolism, susceptibility to}, 188050*
HAX1	*157.7*	*100%*	*100%*	*Neutropenia, severe congenital 3, autosomal recessive, 610738*
HNF1A	*164.4*	*99%*	*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	*159.1*	*99%*	*98%*	*No OMIM phenotype* *{Prostate cancer, increased risk} (Lin (2013) Prostate 73, 169)*

IPMK	*116.2*	*98%*	*91%*	*No OMIM phenotype* *Small intestinal carcinoid (Sei (2015) Gastroenterology 149,67)*
KIF1B	*186.4*	*99%*	*99%*	*Pheochromocytoma, 171300* *?Charcot-Marie-Tooth disease, type 2A1, 118210* *{Neuroblastoma, susceptibility to, 1}, 256700*
KIT	*195.4*	*100%*	*99%*	*Gastrointestinal stromal tumor, familial, 606764* *Germ cell tumors, 273300* *Leukemia, acute myeloid, 601626* *Mast cell disease, 154800* *Piebaldism, 172800*
KLLN	*143.7*	*100%*	*100%*	*Cowden syndrome 4, 615107*
LIG4	*207.5*	*100%*	*99%*	*LIG4 syndrome, 606593* *{Multiple myeloma, resistance to}, 254500*
LZTR1	*166.5*	*100%*	*99%*	*Noonan syndrome 10, 616564* *{Schwannomatosis-2, susceptibility to}, 615670*
MAX	*92.9*	*99%*	*96%*	*{Pheochromocytoma, susceptibility to}, 171300*
MDH2	*121.1*	*98%*	*97%*	*Epileptic encephalopathy, early infantile, 51, 617339*
MEN1	*140.1*	*99%*	*97%*	*Adrenal adenoma, somatic* *Angiofibroma, somatic* *Carcinoid tumor of lung* *Lipoma, somatic* *Multiple endocrine neoplasia 1, 131100* *Parathyroid adenoma, somatic*
MET	*223.1*	*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	*173.3*	*100%*	*100%*	*Tietz albinism-deafness syndrome, 103500* *Waardenburg syndrome, type 2A, 193510* *Waardenburg syndrome/ocular albinism, digenic, 103470* *{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456*
MLH1	*187.4*	*100%*	*99%*	*Colorectal cancer, hereditary nonpolyposis, type 2, 609310* *Mismatch repair cancer syndrome, 276300* *Muir-Torre syndrome, 158320*
MPL	*163.9*	*99%*	*97%*	*Myelofibrosis with myeloid metaplasia, somatic, 254450*

				Thrombocythemia 2, 601977 *Thrombocytopenia, congenital amegakaryocytic, 604498*
MRE11A	*64.3*	*97%*	*89%*	*Ataxia-telangiectasia-like disorder, 604391*
MSH2	*137.3*	*99%*	*96%*	*Colorectal cancer, hereditary nonpolyposis, type 1, 120435* *Mismatch repair cancer syndrome, 276300* *Muir-Torre syndrome, 158320*
MSH3	*142.5*	*99%*	*97%*	*Endometrial carcinoma, somatic, 608089* *Familial adenomatous polyposis 4,617100*
MSH6	*190.9*	*100%*	*99%*	*Colorectal cancer, hereditary nonpolyposis, type 5, 614350* *Endometrial cancer, familial, 608089* *Mismatch repair cancer syndrome, 276300*
MUC5B	*109.8*	*87%*	*82%*	*{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500*
MUTYH	*178*	*100%*	*99%*	*Adenomas, multiple colorectal, 608456* *Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600* *Gastric cancer, somatic, 613659*
NBN	*106*	*99%*	*98%*	*Aplastic anemia, 609135* *Leukemia, acute lymphoblastic, 613065* *Nijmegen breakage syndrome, 251260*
NF1	*146.2*	*93%*	*91%*	*Leukemia, juvenile myelomonocytic, 607785* *Neurofibromatosis, familial spinal, 162210* *Neurofibromatosis, type 1, 162200* *Neurofibromatosis-Noonan syndrome, 601321* *Watson syndrome, 193520*
NF2	*113.8*	*100%*	*99%*	*Meningioma, NF2-related, somatic, 607174* *Neurofibromatosis, type 2, 101000* *Schwannomatosis, 162091*
NHP2	*101.6*	*100%*	*99%*	*Dyskeratosis congenita, autosomal recessive 2, 613987*
NOP10	*159.6*	*100%*	*99%*	*Dyskeratosis congenita, autosomal recessive 1, 224230*
NSD1	*181.1*	*100%*	*100%*	*Beckwith-Wiedemann syndrome, 130650* *Leukemia, acute myeloid, 601626* *Sotos syndrome 1, 117550*
NTHL1	*118.4*	*98%*	*94%*	*Familial adenomatous polyposis 3, 616415*
OGG1	*162.2*	*100%*	*99%*	*Renal cell carcinoma, clear cell, somatic, 144700*
PALB2	*180*	*100%*	*99%*	*Fanconi anemia, complementation group N, 610832* *{Breast cancer, susceptibility to}, 114480*

				{Pancreatic cancer, susceptibility to, 3}, 613348
PARK2	*142.8*	*99%*	*99%*	*Adenocarcinoma of lung, somatic, 211980* *Adenocarcinoma, ovarian, somatic, 167000* *Parkinson disease, juvenile, type 2, 600116* *{Leprosy, susceptibility to}, 607572*
PARN	*151.5*	*100%*	*99%*	*Dyskeratosis congenita, autosomal recessive 6, 616353* *Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371*
PAX5	*127.9*	*98%*	*96%*	*{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545*
PHOX2B	*106.6*	*94%*	*88%*	*Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880* *Neuroblastoma with Hirschsprung disease, 613013* *{Neuroblastoma, susceptibility to, 2}, 613013*
PMS2	*109.8*	*83%*	*81%*	*Colorectal cancer, hereditary nonpolyposis, type 4, 614337* *Mismatch repair cancer syndrome, 276300*
PMS2CL	*NC*	*NC*	*NC*	*No OMIM phenotype*
POLD1	*117.3*	*95%*	*92%*	*Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381* *{Colorectal cancer, susceptibility to, 10}, 612591*
POLE	*158.9*	*99%*	*99%*	*FILS syndrome, 615139* *{Colorectal cancer, susceptibility to, 12}, 615083*
POT1	*125.8*	*99%*	*98%*	*{Glioma susceptibility 9}, 616568* *{Melanoma, cutaneous malignant, susceptibility to, 10}, 615848*
POU6F2	*143.1*	*98%*	*98%*	*{Wilms tumor susceptibility-5}, 601583*
PPM1D	*183.5*	*100%*	*99%*	*Breast cancer, 114480*
PRF1	*128.8*	*100%*	*99%*	*Aplastic anemia, 609135* *Hemophagocytic lymphohistiocytosis, familial, 2, 603553* *Lymphoma, non-Hodgkin, 605027*
PRKAR1A	*104.2*	*97%*	*92%*	*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	*127.7*	*98%*	*96%*	*Basal cell carcinoma, somatic, 605462* *Basal cell nevus syndrome, 109400* *Holoprosencephaly-7, 610828*
PTEN	*169.6*	*100%*	*99%*	*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	*119.9*	*94%*	*89%*	*Nijmegen breakage syndrome-like disorder, 613078*
RAD51C	*165.7*	*100%*	*99%*	*Fanconi anemia, complementation group O, 613390* *{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399*
RAD51D	*178.4*	*100%*	*99%*	*{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291*
RB1	*97.9*	*94%*	*83%*	*Bladder cancer, somatic, 109800* *Osteosarcoma, somatic, 259500* *Retinoblastoma, 180200* *Retinoblastoma, trilateral, 180200* *Small cell cancer of the lung, somatic, 182280*
RECQL	*164.7*	*99%*	*97%*	*No OMIM phenotype* *Breast cancer (Cybulski (2015) Nat Genet 47,643)*
RECQL4	*152.3*	*99%*	*98%*	*Baller-Gerold syndrome, 218600* *RAPADILINO syndrome, 266280* *Rothmund-Thomson syndrome, 268400*
REST	*150.7*	*100%*	*100%*	*{Wilms tumor 6, susceptibility to}, 616806*
RET	*169*	*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	*225.7*	*99%*	*98%*	*No OMIM phenotype* *?Breast cancer (Park (2014) Cancer Discov 4, 804)*
RNF43	*144.5*	*99%*	*99%*	*Sessile serrated polyposis cancer syndrome, 617108*
RPL11	*111.8*	*100%*	*99%*	*Diamond-Blackfan anemia 7, 612562*

RPL35A	*90.3*	*97%*	*88%*	*Diamond-Blackfan anemia 5, 612528*
RPL5	*48.5*	*86%*	*71%*	*Diamond-Blackfan anemia 6, 612561*
RPS10	*131.7*	*97%*	*91%*	*Diamond-Blackfan anemia 9, 613308*
RPS17	*51.1*	*83%*	*70%*	*Diamond-Blackfan anemia 4, 612527*
RPS19	*97.5*	*99%*	*94%*	*Diamond-Blackfan anemia 1, 105650*
RPS24	*131.5*	*97%*	*91%*	*Diamond-blackfan anemia 3, 610629*
RPS26	*95.3*	*91%*	*78%*	*Diamond-Blackfan anemia 10, 613309*
RPS7	*118.7*	*84%*	*68%*	*Diamond-Blackfan anemia 8, 612563*
RTEL1	*137.2*	*99%*	*97%*	*Dyskeratosis congenita, autosomal dominant 4, 615190* *Dyskeratosis congenita, autosomal recessive 5, 615190* *Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373*
RUNX1	*111.4*	*97%*	*92%*	*Leukemia, acute myeloid, 601626* *Platelet disorder, familial, with associated myeloid malignancy, 601399*
SBDS	*231.2*	*100%*	*99%*	*Shwachman-Diamond syndrome, 260400* *{Aplastic anemia, susceptibility to}, 609135*
SDHA	*123.2*	*84%*	*79%*	*Cardiomyopathy, dilated, 1GG, 613642* *Leigh syndrome, 256000* *Mitochondrial respiratory chain complex II deficiency, 252011* *Paragangliomas 5, 614165*
SDHAF2	*151.4*	*94%*	*93%*	*Paragangliomas 2, 601650*
SDHB	*146.3*	*100%*	*99%*	*Cowden syndrome 2, 612359* *Gastrointestinal stromal tumor, 606764* *Paraganglioma and gastric stromal sarcoma, 606864* *Paragangliomas 4, 115310* *Pheochromocytoma, 171300*
SDHC	*117.8*	*99%*	*96%*	*Gastrointestinal stromal tumor, 606764* *Paraganglioma and gastric stromal sarcoma, 606864* *Paragangliomas 3, 605373*
SDHD	*59.2*	*63%*	*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	*153.4*	*99%*	*99%*	*Cone-rod dystrophy 10, 610283* *Retinitis pigmentosa 35, 610282*
SFTPA1	*186.3*	*99%*	*99%*	*{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500*
SFTPA2	*182.9*	*100%*	*100%*	*Pulmonary fibrosis, idiopathic, 178500*
SH2B3	*113.1*	*95%*	*84%*	*Erythrocytosis, somatic, 133100* *Myelofibrosis, somatic, 254450* *Thrombocythemia, somatic, 187950*
SLX4	*129.5*	*100%*	*99%*	*Fanconi anemia, complementation group P, 613951*
SMAD4	*136.5*	*99%*	*99%*	*Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050* *Myhre syndrome, 139210* *Pancreatic cancer, somatic, 260350* *Polyposis, juvenile intestinal, 174900*
SMAD9	*149.8*	*100%*	*100%*	*Pulmonary hypertension,primary,615342* *Polyposis & gastrointestinal ganglioneuromas (Ngeow (2015) Gastroenterology 149,886)*
SMARCA4	*165.4*	*100%*	*99%*	*Coffin-Siris syndrome 4, 614609* *{Rhabdoid tumor predisposition syndrome 2}, 613325*
SMARCB1	*265.7*	*100%*	*100%*	*Coffin-Siris syndrome 3, 614608* *Rhabdoid tumors, somatic, 609322* *{Rhabdoid predisposition syndrome 1}, 609322* *{Schwannomatosis-1, susceptibility to}, 162091*
SQSTM1	*142.3*	*99%*	*96%*	*Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437* *Paget disease of bone 3, 167250*
STK11	*127.1*	*99%*	*97%*	*Melanoma, malignant, somatic* *Pancreatic cancer, 260350* *Peutz-Jeghers syndrome, 175200* *Testicular tumor, somatic, 273300*
SUFU	*146*	*99%*	*97%*	*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	*124.9*	*99%*	*97%*	*No OMIM phenotype* *Melanoma (Aoude (2015) J Natl Cancer Inst 107)*
TERT	*150*	*96%*	*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	*208.7*	*100%*	*100%*	*Dyskeratosis congenita, autosomal dominant 3, 613990* *Revesz syndrome, 268130*
TMEM127	*110.1*	*97%*	*93%*	*{Pheochromocytoma, susceptibility to}, 171300*
TNFRSF11A	*152.6*	*94%*	*91%*	*Osteolysis, familial expansile, 174810* *Osteopetrosis, autosomal recessive 7, 612301* *{Paget disease of bone 2, early-onset}, 602080*
TP53	*100.3*	*99%*	*97%*	*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	*140.4*	*99%*	*97%*	*Lymphangioleiomyomatosis, 606690* *Tuberous sclerosis-1, 191100*
TSC2	*150.1*	*99%*	*99%*	*Lymphangioleiomyomatosis, somatic, 606690* *Tuberous sclerosis-2, 613254*
USB1	*157.7*	*99%*	*97%*	*Poikiloderma with neutropenia,604173*
VHL	*126*	*97%*	*90%*	*Erythrocytosis, familial, 2, 263400* *Hemangioblastoma, cerebellar, somatic* *Pheochromocytoma, 171300* *Renal cell carcinoma, somatic, 144700* *von Hippel-Lindau syndrome, 193300*
WAS	*72*	*89%*	*80%*	*Neutropenia, severe congenital, X-linked, 300299* *Thrombocytopenia, X-linked, 313900* *Thrombocytopenia, X-linked, intermittent, 313900* *Wiskott-Aldrich syndrome, 301000*

WRAP53	*175.2*	*100%*	*100%*	*Dyskeratosis congenita, autosomal recessive 3, 613988*
WRN	*149.6*	*99%*	*96%*	*Werner syndrome, 277700*
WT1	*94.2*	*96%*	*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	*69.2*	*97%*	*89%*	*Xeroderma pigmentosum, group A, 278700*
XPC	*176.3*	*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 2.9

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