HEREDITARY CANCER GENE PANEL DG 2.4.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM ID*
ALK	89.8	99%	96%	{Neuroblastoma, susceptibility to, 3}, 613014
APC	144.1	100%	99%	Adenomatous polyposis coli, 175100 Gastric cancer, somatic, 613659 Adenoma, periampullary, somatic Hepatoblastoma, somatic, 114550 Desmoid disease, hereditary, 135290 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100
ARMC5	95.8	100%	98%	macronodular adrenal hyperplasia with Cushings syndrome
ATM	111.1	99%	99%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic
ATR	113.1	100%	99%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564
BAP1	89.6	99%	97%	Tumor predisposition syndrome, 614327
BARD1	114.6	100%	99%	{Breast cancer, susceptibility to}, 114480
BLM	114.8	100%	98%	Bloom syndrome, 210900
BMPR1A	59.2	79%	64%	Polyposis, juvenile intestinal, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Juvenile polyposis syndrome, infantile form, 174900
BRCA1	143.5	98%	96%	{Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320

BRCA2	142.7	99%	99%	{Breast-ovarian cancer, familial, 2}, 612555 Fanconi anemia, complementation group D1, 605724 Prostate cancer, 176807 {Breast cancer, male, susceptibility to}, 114480 Wilms tumor, 194070 {Medulloblastoma}, 155255 {Glioblastoma 3}, 613029
BRIP1	120.9	100%	99%	?Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BUB1	103.9	99%	98%	Colorectal cancer with chromosomal instability,somatic
BUB1B	111	100%	99%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BUB3	106.1	100%	100%	No OMIM phenotype Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)
CDC73	138.9	100%	100%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid carcinoma, 608266
CDH1	105.2	100%	100%	Endometrial carcinoma, somatic, 608089 Ovarian carcinoma, somatic, 167000 {Breast cancer, lobular}, 114480 Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 {Prostate cancer, susceptibility to}, 176807
CDK4	120.9	96%	88%	{Melanoma, cutaneous malignant, 3}, 609048
CDKN2A	97.6	93%	93%	{Melanoma, cutaneous malignant, 2}, 155601 Melanoma and neural system tumor syndrome, 155755 Pancreatic cancer/melanoma syndrome, 606719
CENPJ	127.2	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676
CHEK2	53	60%	56%	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807 {Breast and colorectal cancer, susceptibility to}

CREBBP	78	99%	97%	Rubinstein-Taybi syndrome, 180849
CTC1	96.7	99%	96%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTNNA1	90.9	99%	94%	No OMIM phenotype Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CYLD	113.6	100%	99%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
DDB2	90.8	100%	98%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	10.3	16%	12%	Warsaw breakage syndrome, 613398
DICER1	117.9	100%	99%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800
DKC1	91.1	100%	98%	Dyskeratosis congenita, X-linked, 305000
EGFR	97	100%	99%	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980
ELANE	109.7	99%	92%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ERCC1	80.4	98%	93%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	87.2	99%	93%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	121.5	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC4	140.8	99%	93%	Xeroderma pigmentosum, group F, 278760 XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	113.8	98%	97%	Xeroderma pigmentosum, group G, 278780
ERCC6	141.3	98%	97%	Cockayne syndrome, type B, 133540 Cerebrooculofacioskeletal syndrome 1, 214150 De Sanctis-Cacchione syndrome, 278800 {Macular degeneration, age-related, susceptibility to 5}, 613761 UV-sensitive syndrome 1, 600630
EXO1	105.6	94%	94%	No OMIM phenotype Colorectal cancer, non-polyposis (Wu (2001) Gastroenterology 120,1580)

EXT1	103.7	97%	95%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	104.7	100%	95%	Exostoses, multiple, type 2, 133701
FAN1	112.7	100%	99%	Interstitial nephritis,karyomegalic,614817
FANCA	87.6	99%	97%	Fanconi anemia, complementation group A, 227650
FANCB	121.9	100%	97%	Fanconi anemia, complementation group B, 300514
FANCC	76.9	100%	94%	Fanconi anemia, complementation group C, 227645
FANCD2	92.2	87%	86%	Fanconi anemia, complementation group D2, 227646
FANCE	86.4	97%	92%	Fanconi anemia, complementation group E, 600901
FANCF	143.4	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	120.8	99%	96%	Fanconi anemia, complementation group G, 614082
FANCI	118.1	100%	100%	Fanconi anemia, complementation group I, 609053
FANCL	93.8	100%	100%	Fanconi anemia, complementation group L, 614083
FANCM	117.3	100%	99%	Fanconi anemia, complementation group M, 614087
FH	85.3	96%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLCN	110.5	100%	97%	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500
G6PC3	115.8	100%	99%	Dursun syndrome,612541 Neutropenia,severe congenital 4,autosomal recessive,612541
GDNF	146.5	100%	97%	Central hypoventilation syndrome, 209880 {Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711
GFI1	71.2	100%	98%	Neutropenia, severe congenital 2, autosomal dominant, 613107 Neutropenia, nonimmune chronic idiopathic, of adults, 607847
GPC3	96	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GREM1	117.8	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	87.7	100%	100%	{?Thyroid cancer,nonmedullary,5},616535 {Venous thromboembolism,susceptibility to},188050
HAX1	128.2	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HNF1A	89.1	100%	96%	MODY, type III, 600496 {Diabetes mellitus, noninsulin-dependent, 2}, 125853 {Diabetes mellitus, insulin-dependent}, 222100 Hepatic adenoma, somatic, 142330 Renal cell carcinoma, 144700 Diabetes mellitus, insulin-dependent, 20, 612520
HOXB13	114.4	100%	100%	No OMIM phenotype Prostate cancer, increased risk (Lin (2013) Prostate 73, 169)
KIF1B	120.2	100%	100%	Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIT	106.6	98%	96%	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mast cell disease, 154800 Leukemia, acute myeloid, 601626 Germ cell tumors, 273300
KLLN	109.6	100%	100%	Cowden syndrome 4, 615107
LZTR1	87.7	100%	99%	{Schwannomatosis-2, susceptibility to}, 615670
MAX	92.7	96%	95%	{Pheochromocytoma, susceptibility to}, 171300
MDH2	99.5	98%	97%	No OMIM phenotype pheochromocytoma (Cascón A et al, J Natl Cancer Inst. (2015) 11;107)
MEN1	106.3	100%	96%	Multiple endocrine neoplasia 1, 131100 Carcinoid tumor of lung Parathyroid adenoma, somatic Lipoma, somatic Angiofibroma, somatic Adrenal adenoma, somatic
MET	125.1	100%	99%	papillary renal cell cancer
MLH1	98.7	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320

MPL	112	100%	99%	Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977 Myelofibrosis with myeloid metaplasia, somatic, 254450
MRE11A	90.5	99%	99%	Ataxia-telangiectasia-like disorder, 604391
MSH2	98.8	98%	97%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300
MSH6	144.7	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MUC5B	44.6	68%	59%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
MUTYH	110.4	100%	99%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600
NBN	115.9	98%	97%	Aplastic anemia,609135 Leukemia,acute lymphoblastic,613065 Nijmegen breakage syndrome,251260
NF1	78.6	82%	81%	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	82	100%	98%	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, 162091
NHP2	47	100%	93%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	169.5	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NSD1	119.6	100%	99%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650
OGG1	99.4	99%	98%	Renal cell carcinoma, clear cell, somatic, 144700
PALB2	122.1	99%	97%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348

PARK2	61.6	100%	93%	Lung cancer
PHOX2B	64.1	100%	85%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 {Neuroblastoma, susceptibility to, 2}, 613013 Neuroblastoma with Hirschsprung disease, 613013
PMS2	68.3	56%	55%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
POLD1	78.3	94%	91%	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	103.8	100%	98%	{Colorectal cancer, susceptibility to, 12}, 615083 FILS syndrome, 615139
PRF1	84.5	100%	97%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKAR1A	94.7	91%	83%	Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Thyroid carcinoma, papillary, somatic, 188550 Pigmented nodular adrenocortical disease, primary, 1, 610489
PTCH1	86.9	99%	95%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTEN	122.4	99%	94%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 {Meningioma}, 607174 {Glioma susceptibility 2}, 613028 Macrocephaly/autism syndrome, 605309
PTPRJ	105.8	97%	97%	Colon cancer, somatic, 114500
RAD50	113.2	100%	99%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	93.4	100%	100%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD51D	81.7	96%	91%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291

RB1	103.6	98%	98%	Retinoblastoma, 180200 Osteosarcoma, somatic, 259500 Bladder cancer, somatic, 109800 Small cell cancer of the lung, somatic, 182280 Retinoblastoma, trilateral, 180200
RECQL	102.8	99%	95%	No OMIM phenotype Breast cancer (Cybulski et al, Nat Genet. (2015) 47:643-646)
RECQL4	91.5	98%	96%	Rothmund-Thomson syndrome, 268400 RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600
RET	88.5	97%	94%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIB, 162300 Central hypoventilation syndrome, congenital, 209880 Pheochromocytoma, 171300 Renal agenesis, 191830
RINT1	125.6	99%	98%	No OMIM phenotype Breast cancer (Park DJ et al, Cancer Discov. (2014) 4:804-815)
RPL11	69.4	94%	87%	Diamond-Blackfan anemia 7, 612562
RPL35A	26.5	75%	50%	Diamond-Blackfan anemia 5, 612528
RPL5	30.4	79%	60%	Diamond-Blackfan anemia 6, 612561
RPS10	39.3	90%	74%	Diamond-Blackfan anemia 9, 613308
RPS17	0	0%	0%	Diamond-Blackfan anemia 4, 612527
RPS19	39.8	65%	45%	Diamond-Blackfan anemia 1, 105650
RPS24	89.5	96%	94%	Diamond-blackfan anemia 3, 610629
RPS26	34.5	64%	62%	Diamond-Blackfan anemia 10, 613309
RPS7	19.4	81%	47%	Diamond-Blackfan anemia 8, 612563
RTEL1	73.3	99%	92%	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190
RUNX1	63.7	97%	89%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SBDS	88.2	98%	93%	Shwachman-Bodian-Diamond syndrome, 260400

SDHA	8.6	34%	12%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SDHAF2	100	94%	94%	Paragangliomas 2, 601650
SDHB	94.2	100%	100%	Paragangliomas 4, 115310 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764
SDHC	32	55%	49%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	40.7	42%	33%	Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300 Carcinoid tumors, intestinal, 114900 Merkel cell carcinoma, somatic Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 3, 615106
SEMA4A	101.8	99%	97%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SFTPA1	20.7	43%	39%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
SFTPA2	23.2	50%	43%	Pulmonary fibrosis, idiopathic, 178500
SLX4	128.5	97%	95%	Fanconi anemia, complementation group P, 613951
SMAD4	119.9	100%	98%	Pancreatic cancer Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210
SMAD9	98.9	100%	98%	Pulmonary hypertension,primary,615342
SMARCA4	83.2	98%	92%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609
SMARCB1	120.7	100%	100%	Rhabdoid tumors, somatic, 609322 Rhabdoid predisposition syndrome 1, 609322 Mental retardation, autosomal dominant 15, 614608

SQSTM1	83.9	99%	96%	Paget disease of bone, 602080
STK11	73.3	99%	95%	Peutz-Jeghers syndrome, 175200 Pancreatic cancer, 260350 Testicular tumor, somatic, 273300
SUFU	89.6	98%	90%	Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
TERT	103.7	100%	99%	{Bone marrow failure, telomere-related, 1}, 614742 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989 {Pulmonary fibrosis, telomere-related, 1}, 614742
TINF2	176.7	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMEM127	61.8	99%	88%	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	100.8	95%	93%	Osteolysis, familial expansile, 174810 Paget disease of bone, 602080 Osteopetrosis, autosomal recessive 7, 612301
TP53	77.6	94%	94%	Colorectal cancer, 114500 Li-Fraumeni syndrome, 151623 Hepatocellular carcinoma, 114550 Osteosarcoma, 259500 Choroid plexus papilloma, 260500 Nasopharyngeal carcinoma, 607107 Pancreatic cancer, 260350 Adrenal cortical carcinoma, 202300
TSC1	89.9	99%	97%	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	81.8	98%	95%	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690
VHL	114.9	100%	100%	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400

WAS	60.1	100%	90%	Wiskott-Aldrich syndrome, 301000 Thrombocytopenia, X-linked, 313900 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900
WRAP53	142.8	100%	98%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	126.7	100%	98%	Werner syndrome, 277700
WT1	70.1	100%	99%	Wilms tumor, type 1, 194070 Denys-Drash syndrome, 194080 Nephrotic syndrome, type 4, 256370 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240
XPA	80.9	100%	93%	Xeroderma pigmentosum, group A, 278700
XPC	114.6	99%	97%	Xeroderma pigmentosum, group C, 278720
XRCC2	170	100%	100%	No OMIM phenotype Breast cancer (Hilbers FS et al, J Med Genet. (2012) 49:618-620)

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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association