HEREDITARY CANCER GENE PANEL DG 2.3.x

Gene	Median coverage	% covered >10x	% covered >20x	Associated Phenotype description and OMIM ID
ALK	99,5	100%	98%	{Neuroblastoma, susceptibility to, 3}, 613014
APC	151,1	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 Gardner syndrome, 175100
ARMC5	99,3	99%	95%	macronodular adrenal hyperplasia with Cushings syndrome
ATM	120,3	100%	99%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic
ATR	126,2	100%	99%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564
BAP1	89,5	100%	95%	Tumor predisposition syndrome, 614327
BARD1	123,2	100%	100%	{Breast cancer, susceptibility to}, 114480
BLM	119,5	100%	100%	Bloom syndrome, 210900
BMPR1A	62,7	75%	65%	Polyposis, juvenile intestinal, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Juvenile polyposis syndrome, infantile form, 174900
BRCA1	153,3	98%	97%	{Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320

BRCA2	156,4	100%	100%	{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 {Pre-B-cell acute lymphoblastic leukemia} Pancreatic cancer, 613347
BRIP1	138,8	100%	100%	?Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BUB1	116,2	99%	98%	Colorectal cancer with chromosomal instability,somatic
BUB1B	122,3	100%	100%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BUB3	111,9	100%	100%	No OMIM phenotype Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)
CDC73	148,7	100%	100%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	117,1	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	122,6	97%	91%	{Melanoma, cutaneous malignant, 3}, 609048
CDKN2A	85,1	93%	93%	{Melanoma, cutaneous malignant, 2}, 155601 Melanoma and neural system tumor syndrome, 155755 Pancreatic cancer/melanoma syndrome, 606719 Orolaryngeal cancer, multiple, -3
CENPJ	140,1	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676

CHEK2	55,4	63%	58%	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	84,2	99%	98%	Rubinstein-Taybi syndrome, 180849
CTC1	105,2	100%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTNNA1	97,5	99%	97%	No OMIM phenotype Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CYLD	115,7	100%	100%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
DDB2	96,7	98%	97%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	11,3	24%	11%	Warsaw breakage syndrome, 613398
DICER1	125,9	100%	99%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800
DKC1	99,3	100%	100%	Dyskeratosis congenita, X-linked, 305000
EGFR	103,3	100%	98%	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
ELANE	113,9	94%	89%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ERCC1	81,9	100%	97%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	91,4	99%	95%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	130,7	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC4	150	100%	95%	Xeroderma pigmentosum, group F, 278760 XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	126,5	98%	97%	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780

ERCC6	158,4	99%	98%	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 {Lung cancer, susceptibility to}, 211980
EXO1	116,3	94%	94%	No OMIM phenotype Colorectal cancer, non-polyposis (Wu (2001) Gastroenterology 120,1580)
EXT1	109,6	100%	97%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	126,3	100%	96%	Exostoses, multiple, type 2, 133701
FANCA	91,1	100%	98%	Fanconi anemia, complementation group A, 227650
FANCB	134,7	100%	99%	Fanconi anemia, complementation group B, 300514
FANCC	82,5	100%	98%	Fanconi anemia, complementation group C, 227645
FANCD2	99,3	87%	86%	Fanconi anemia, complementation group D2, 227646
FANCE	90,7	95%	89%	Fanconi anemia, complementation group E, 600901
FANCF	146,4	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	137,6	100%	100%	Fanconi anemia, complementation group G, 614082
FANCI	131,3	100%	100%	Fanconi anemia, complementation group I, 609053
FANCL	103,2	100%	99%	Fanconi anemia, complementation group L, 614083
FANCM	120,3	100%	99%	Fanconi anemia, complementation group M, 614087
FH	94,6	97%	91%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLCN	115,2	100%	99%	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500
G6PC3	117	100%	100%	Dursun syndrome,612541 Neutropenia,severe congenital 4,autosomal recessive,612541
GDNF	185	100%	96%	Central hypoventilation syndrome, 209880 {Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711
GFI1	63,9	100%	95%	Neutropenia, severe congenital 2, autosomal dominant, 613107 Neutropenia, nonimmune chronic idiopathic, of adults, 607847

GPC3	110,8	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GREM1	139,8	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)
HAX1	143,7	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HNF1A	94,2	100%	97%	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	108,1	100%	99%	No OMIM phenotype Prostate cancer, increased risk (Lin (2013) Prostate 73, 169)
KIF1B	133,5	100%	100%	Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIT	117	99%	98%	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mast cell disease, 154800 Leukemia, acute myeloid, 601626 Germ cell tumors, 273300
KLLN	101,5	100%	100%	Cowden syndrome 4, 615107
LZTR1	94,7	100%	99%	{Schwannomatosis-2, susceptibility to}, 615670
MAX	89,5	96%	96%	{Pheochromocytoma, susceptibility to}, 171300
MDH2	99,9	92%	91%	No OMIM phenotype
MEN1	115,6	100%	94%	Multiple endocrine neoplasia 1, 131100 Carcinoid tumor of lung Parathyroid adenoma, somatic Lipoma, somatic Angiofibroma, somatic Adrenal adenoma, somatic
MET	135,1	100%	99%	papillary renal cell cancer

MLH1	106	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MPL	122,9	100%	98%	Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977 Myelofibrosis with myeloid metaplasia, somatic, 254450
MRE11A	90,1	99%	99%	Ataxia-telangiectasia-like disorder, 604391
MSH2	103,3	97%	96%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300
MSH6	152,7	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MUC5B	47,9	73%	62%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
MUTYH	120,2	100%	100%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600
NBN	120,8	98%	95%	Aplastic anemia,609135 Leukemia,acute lymphoblastic,613065 Nijmegen breakage syndrome,251260
NF1	86,7	83%	81%	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Melanoma, desmoplastic neurotrophic Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	88,3	100%	95%	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, 162091
NHP2	54,8	100%	95%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	170,2	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NSD1	131,5	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650
OGG1	105,9	100%	98%	Renal cell carcinoma, clear cell, somatic, 144700

PALB2	146,4	98%	97%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PARK2	73,1	96%	94%	Lung cancer
PHOX2B	62,9	94%	82%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013
PMS2	70	56%	56%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
POLD1	73,5	94%	91%	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	111,2	100%	99%	{Colorectal cancer, susceptibility to, 12}, 615083 FILS syndrome, 615139
PRF1	100,8	100%	98%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKAR1A	99,4	95%	85%	Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Thyroid carcinoma, papillary, somatic, 188550 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800
PTCH1	85,6	97%	94%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTEN	141,4	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 PTEN hamartoma tumor syndrome VATER association with macrocephaly and ventriculomegaly, 276950 {Prostate cancer, somatic}, 176807 Thyroid carcinoma, follicular, somatic, 188470 Malignant melanoma, somatic, 155600 Endometrial carcinoma, somatic, 608089 Squamous cell carcinoma, head and neck, somatic, 275355

PTPRJ	121	97%	97%	Colon cancer, somatic, 114500
RAD50	122,5	100%	99%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	102,1	100%	100%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD51D	81,9	100%	94%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RB1	111,3	100%	98%	Retinoblastoma, 180200 Osteosarcoma, somatic, 259500 Bladder cancer, somatic, 109800 Small cell cancer of the lung, somatic, 182280 Retinoblastoma, trilateral, 180200
RECQL	106,5	99%	98%	No OMIM phenotype
RECQL4	94,2	98%	95%	Rothmund-Thomson syndrome, 268400 RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600
RET	95,3	95%	93%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIB, 162300 Central hypoventilation syndrome, congenital, 209880 Pheochromocytoma, 171300 Renal agenesis, 191830 {Hirschsprung disease, susceptibility to, 1}, 142623
RINT1	139,8	98%	98%	No OMIM phenotype
RPL11	76,5	100%	95%	Diamond-Blackfan anemia 7, 612562
RPL35A	26,3	89%	57%	Diamond-Blackfan anemia 5, 612528
RPL5	35,3	84%	56%	Diamond-Blackfan anemia 6, 612561
RPS10	41	85%	66%	Diamond-Blackfan anemia 9, 613308
RPS17	0,6	0%	0%	Diamond-Blackfan anemia 4, 612527
RPS19	42,8	66%	45%	Diamond-Blackfan anemia 1, 105650
RPS24	95,7	96%	92%	Diamond-blackfan anemia 3, 610629
RPS26	38,2	63%	62%	Diamond-Blackfan anemia 10, 613309
RPS7	20,1	87%	33%	Diamond-Blackfan anemia 8, 612563
RTEL1	78,4	99%	89%	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190

RUNX1	64,8	92%	84%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SBDS	77,3	98%	89%	Shwachman-Bodian-Diamond syndrome, 260400
SDHA	10,4	28%	16%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SDHAF2	94	95%	94%	Paragangliomas 2, 601650
SDHB	97	100%	100%	Paragangliomas 4, 115310 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764
SDHC	30,6	53%	43%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	43,2	55%	37%	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	106	99%	96%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SFTPA1	22,6	45%	41%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
SFTPA2	29,7	45%	43%	Pulmonary fibrosis, idiopathic, 178500
SLX4	146,7	99%	96%	Fanconi anemia, complementation group P, 613951
SMAD4	137,5	100%	98%	Pancreatic cancer Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210
SMARCA4	81,5	97%	92%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609

SMARCB1	124,7	100%	100%	Rhabdoid tumors, somatic, 609322 Rhabdoid predisposition syndrome 1, 609322 Mental retardation, autosomal dominant 15, 614608
SQSTM1	85,6	100%	96%	Paget disease of bone, 602080
STK11	70,5	99%	95%	Peutz-Jeghers syndrome, 175200 Melanoma, malignant, somatic Pancreatic cancer, 260350 Testicular tumor, somatic, 273300
SUFU	97,9	97%	90%	Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
TERT	110,4	99%	94%	{Bone marrow failure, telomere-related, 1}, 614742 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989 {Coronary artery disease} {Pulmonary fibrosis, telomere-related, 1}, 614742 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134
TINF2	191,8	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMEM127	76,9	95%	89%	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	103,3	95%	92%	Osteolysis, familial expansile, 174810 Paget disease of bone, 602080 Osteopetrosis, autosomal recessive 7, 612301
TP53	86,5	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 Breast cancer, 114480 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800

TSC1	96,9	99%	96%	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	87,7	99%	97%	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690
VHL	121,2	100%	100%	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400
WAS	64,1	100%	98%	Wiskott-Aldrich syndrome, 301000 Thrombocytopenia, X-linked, 313900 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900
WRAP53	142	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	144,3	100%	99%	Werner syndrome, 277700
WT1	70,3	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	83,3	100%	97%	Xeroderma pigmentosum, group A, 278700
XPC	123,8	100%	98%	Xeroderma pigmentosum, group C, 278720
XRCC2	191,2	100%	100%	No OMIM phenotype Breast cancer (Park (2012) Am J Hum Genet 90, 734)

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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 : June 30th, 2015
This list is accurate for all panel versions starting with DG 2.3. (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 Ad 2. OMIM phenotype descriptions between {} signify risk factors