

HEREDITARY CANCER GENE PANEL DG 2.5/2.6

<i>Gene</i>	<i>Median coverage</i>	<i>% covered > 10x</i>	<i>% covered > 20x</i>	<i>Associated phenotype description and OMIM disease ID</i>
ALK	121.9	98%	96%	{Neuroblastoma, susceptibility to, 3}, 613014
APC	152.5	99%	97%	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	96.4	100%	98%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ATM	112	98%	95%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic
ATR	137.4	97%	95%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564
BAP1	121.5	98%	97%	Tumor predisposition syndrome, 614327
BARD1	143.3	100%	99%	{Breast cancer, susceptibility to}, 114480
BLM	115.5	98%	92%	Bloom syndrome, 210900
BMPR1A	98.6	99%	89%	Polyposis, juvenile intestinal, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Juvenile polyposis syndrome, infantile form, 174900
BRCA1	183	98%	95%	{Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320

BRCA2	96.1	98%	95%	{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 {Pancreatic cancer 2},613347
BRIP1	120.7	99%	97%	?Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BUB1	134.3	99%	97%	Colorectal cancer with chromosomal instability,somatic
BUB1B	143.9	98%	97%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BUB3	134.6	97%	97%	No OMIM phenotype Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)
CDC73	96.6	100%	95%	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	107.7	99%	98%	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	113.3	100%	97%	{Melanoma, cutaneous malignant, 3}, 609048
CDKN2A	56.4	93%	91%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CENPJ	144.5	99%	96%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676

CHEK2	99	82%	80%	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	119.9	99%	96%	Rubinstein-Taybi syndrome, 180849
CTC1	93.8	100%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTNNA1	127.1	100%	98%	Macular dystrophy, patterned, 608970 Gastric cancer, diffuse (Majewski (2012) J Pathol epub)
CYLD	116.2	98%	93%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
DDB2	148.5	100%	98%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	83.4	79%	70%	Warsaw breakage syndrome, 613398
DICER1	147.8	99%	97%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800
DKC1	77.4	99%	92%	Dyskeratosis congenita, X-linked, 305000
EGFR	143.2	97%	97%	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	93.3	100%	93%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ERCC1	74.9	100%	99%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	120	100%	99%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	94.3	100%	99%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC4	144.3	100%	99%	Xeroderma pigmentosum, group F, 278760 XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	133.9	100%	96%	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780

ERCC6	165.9	100%	100%	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
ETV6	114.9	100%	100%	Leukemia,acute myeloid,somatic,601626 Thrombocytopenia 5,616216
EXO1	154.6	100%	96%	No OMIM phenotype Colorectal cancer, non-polyposis (Wu (2001) Gastroenterology 120,1580)
EXT1	94.5	100%	98%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	170.1	99%	97%	Exostoses, multiple, type 2, 133701
EZH2	131.8	99%	96%	Weaver syndrome,277590
FAN1	162.2	100%	100%	Interstitial nephritis,karyomegalic,614817
FANCA	107	98%	95%	Fanconi anemia, complementation group A, 227650
FANCB	49.3	89%	75%	Fanconi anemia, complementation group B, 300514
FANCC	108.3	100%	94%	Fanconi anemia, complementation group C, 227645
FANCD2	125.5	98%	96%	Fanconi anemia, complementation group D2, 227646
FANCE	92.2	84%	84%	Fanconi anemia, complementation group E, 600901
FANCF	120.8	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	116.2	100%	98%	Fanconi anemia, complementation group G, 614082
FANCI	158.1	98%	96%	Fanconi anemia, complementation group I, 609053
FANCL	73.4	100%	92%	Fanconi anemia, complementation group L, 614083
FANCM	98.4	98%	94%	Fanconi anemia, complementation group M, 614087
FH	160.3	90%	87%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLCN	142.8	100%	98%	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500
G6PC3	107.7	100%	100%	Dursun syndrome,612541 Neutropenia,severe congenital 4,autosomal recessive,612541

GDNF	138.3	98%	94%	Central hypoventilation syndrome, 209880 {Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711
GFI1	88.8	97%	91%	Neutropenia, severe congenital 2, autosomal dominant, 613107 Neutropenia, nonimmune chronic idiopathic, of adults, 607847
GPC3	72	93%	85%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GREM1	111.3	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)
HABP2	136.9	100%	99%	{?Thyroid cancer,nonmedullary,5},616535 {Venous thromboembolism,susceptibility to},188050
HAX1	123.9	100%	99%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HNF1A	124.6	99%	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%	97%	No OMIM phenotype Prostate cancer, increased risk (Lin (2013) Prostate 73, 169)
IPMK	83.7	92%	67%	No OMIM phenotype Small intestinal carcinoid (Sei (2015) Gastroenterology 149,67)
KIF1B	149.8	99%	98%	Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIT	153.8	99%	98%	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mast cell disease, 154800 Leukemia, acute myeloid, 601626 Germ cell tumors, 273300
KLLN	81.4	100%	100%	Cowden syndrome 4, 615107
LZTR1	125.8	98%	95%	{Schwannomatosis-2, susceptibility to}, 615670
MAX	82	100%	94%	{Pheochromocytoma, susceptibility to}, 171300

MDH2	105	98%	96%	No OMIM phenotype Paraganglioma (Cascon (2015) J Natl Cancer Inst 107,djv053)
MEN1	110.9	96%	89%	Multiple endocrine neoplasia 1, 131100 Carcinoid tumor of lung Parathyroid adenoma, somatic Lipoma, somatic Angiofibroma, somatic Adrenal adenoma, somatic
MET	192.4	100%	99%	?Deafness,autosomal recessive 97,616705 Hepatocellular carcinoma,childhood type,somatic,114550 Renal cell carcinoma,papillary,1,familial and somatic,605074
MLH1	162.3	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MPL	127.5	98%	92%	Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977 Myelofibrosis with myeloid metaplasia, somatic, 254450
MRE11A	53.1	95%	84%	Ataxia-telangiectasia-like disorder, 604391
MSH2	109.1	96%	88%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300
MSH6	159.6	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MUC5B	88.8	86%	82%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
MUTYH	135	100%	99%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600
NBN	74.1	99%	96%	Aplastic anemia,609135 Leukemia,acute lymphoblastic,613065 Nijmegen breakage syndrome,251260
NF1	122.7	91%	86%	Neurofibromatosis, type 1, 162200
NF2	96.7	100%	100%	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, 162091

NHP2	66.3	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	159.4	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NSD1	142.2	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650
OGG1	117.5	100%	98%	Renal cell carcinoma, clear cell, somatic, 144700
PALB2	148.7	100%	100%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PARK2	116	98%	96%	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PARN	123.6	100%	97%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PHOX2B	85.6	91%	87%	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 {Neuroblastoma, susceptibility to, 2}, 613013 Neuroblastoma with Hirschsprung disease, 613013
PMS2	79.5	82%	79%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
PMS2CL				Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
POLD1	89	94%	92%	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	130.6	99%	98%	{Colorectal cancer, susceptibility to, 12}, 615083 FELS syndrome, 615139
PPM1D	157.1	100%	99%	Breast cancer, 114480
PRF1	113.4	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKAR1A	90.8	98%	90%	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	116.3	98%	96%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoencephaly-7, 610828
PTEN	142.2	100%	100%	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	170.5	97%	95%	Colon cancer, somatic, 114500
RAD50	91.4	93%	86%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	148.2	100%	100%	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RAD51D	157.4	100%	98%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RB1	77.5	91%	76%	Retinoblastoma, 180200 Osteosarcoma, somatic, 259500 Bladder cancer, somatic, 109800 Small cell cancer of the lung, somatic, 182280 Retinoblastoma, trilateral, 180200
RECQL	136.1	100%	92%	No OMIM phenotype Breast cancer (Cybulski (2015) Nat Genet 47,643)
RECQL4	121.3	97%	95%	Rothmund-Thomson syndrome, 268400 RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600

RET	137.7	99%	97%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIB, 162300 Central hypoventilation syndrome, congenital, 209880 Pheochromocytoma, 171300 {Hirschsprung disease, susceptibility to},142623
RINT1	178.7	100%	98%	No OMIM phenotype Breast cancer (Park (2014) Cancer Discov 4, 804)
RPL11	89.3	100%	95%	Diamond-Blackfan anemia 7, 612562
RPL35A	64.6	95%	80%	Diamond-Blackfan anemia 5, 612528
RPL5	31	71%	50%	Diamond-Blackfan anemia 6, 612561
RPS10	112.9	93%	89%	Diamond-Blackfan anemia 9, 613308
RPS17	40.8	78%	68%	Diamond-Blackfan anemia 4, 612527
RPS19	78.4	100%	97%	Diamond-Blackfan anemia 1, 105650
RPS24	105.6	99%	87%	Diamond-blackfan anemia 3, 610629
RPS26	71.8	79%	69%	Diamond-Blackfan anemia 10, 613309
RPS7	91.1	79%	70%	Diamond-Blackfan anemia 8, 612563
RTEL1	100	98%	92%	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190
RUNX1	106.5	96%	92%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SBDS	170.6	100%	99%	Shwachman-Bodian-Diamond syndrome, 260400
SDHA	96.8	81%	73%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SDHAF2	142.1	94%	94%	Paragangliomas 2, 601650
SDHB	124.7	100%	100%	Paragangliomas 4, 115310 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764
SDHC	94	99%	92%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864

				Gastrointestinal stromal tumor, 606764
SDHD	51.2	59%	59%	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	118.9	100%	97%	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282 Familial colorectal cancer,type X (Schulz (2014) Nat Commun 5,5191)
SFTPA1	127.7	100%	97%	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
SFTPA2	134.2	100%	100%	Pulmonary fibrosis, idiopathic, 178500
SLX4	101.3	100%	99%	Fanconi anemia, complementation group P, 613951
SMAD4	114.6	99%	99%	Pancreatic cancer,260350 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210
SMAD9	140.8	100%	99%	Pulmonary hypertension,primary,615342 Polyposis & gastrointestinal ganglioneuromas (Ngeow (2015) Gastroenterology 149,886)
SMARCA4	131.9	99%	94%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609
SMARCB1	202.6	100%	100%	Rhabdoid tumors, somatic, 609322 Rhabdoid predisposition syndrome 1, 609322 Mental retardation, autosomal dominant 15, 614608
SQSTM1	97.5	97%	92%	Paget disease of bone, 602080
STK11	112.3	100%	97%	Peutz-Jeghers syndrome, 175200 Melanoma, malignant, somatic Pancreatic cancer, 260350 Testicular tumor, somatic, 273300
SUFU	125.3	97%	96%	Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
TERC				Dyskeratosis congenita,autosomal dominant 1,127550 {Aplastic anemia},614743 {Pulmonary fibrosis,idiopathic,susceptibility to},614743

TERT	119	98%	89%	{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	156.3	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMEM127	90	96%	92%	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	127.3	92%	89%	Osteolysis, familial expansile, 174810 Paget disease of bone, 602080 Osteopetrosis, autosomal recessive 7, 612301
TP53	86.8	98%	91%	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	127.2	99%	97%	Tuberous sclerosis-1, 191100 Lymphangioliomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	111.2	99%	98%	Tuberous sclerosis-2, 613254 Lymphangioliomyomatosis, somatic, 606690
VHL	91.1	98%	75%	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400

WAS	35.9	81%	62%	Wiskott-Aldrich syndrome, 301000 Thrombocytopenia, X-linked, 313900 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900
WRAP53	127.4	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	128.3	97%	94%	Werner syndrome, 277700
WT1	84.4	92%	85%	Wilms tumor, type 1, 194070 Denys-Drash syndrome, 194080 Nephrotic syndrome, type 4, 256370 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240
XPA	46.6	95%	84%	Xeroderma pigmentosum, group A, 278700
XPC	148.7	100%	100%	Xeroderma pigmentosum, group C, 278720
XRCC2	167.4	86%	86%	No OMIM phenotype Breast cancer (Park (2012) Am J Hum Genet 90, 734)

Gene symbols used follow HGNC 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 10th, 2016.

This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 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
