HEREDITARY CANCER GENE PANEL DG 2.15 (206 genes)

Releasedate: 31-01-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
A2ML1	130.8	100	99.7	No OMIM phenotype Noonan-like syndrome (Vissers et al. 2015) Noonan syndrome (van Trier (2015) Int J Pediatr Otorhinolaryngol, epub) Otitis media, susceptibility to (Santos-Cortez (2015) Nat Genet 47,917)
ACD	135.2	100	98.2	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
AIP	154.2	99.9	99	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102000
ALK	128.8	99.4	98.1	{Neuroblastoma, susceptibility to, 3}, 613014
ANKRD26	81	88.7	76.8	Thrombocytopenia 2, 188000
APC	159	99.9	98.9	Adenoma, periampullary, somatic, 0 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	136.5	99.8	97.5	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ASXL1	159.8	99.1	97.7	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ATM	109.7	99	94	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0

				{Breast cancer, susceptibility to}, 114480
ATR	138.3	99.4	96.9	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
AXIN2	114.5	99.7	98.9	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BAP1	111	85.1	82.7	Tumor predisposition syndrome, 614327
BARD1	150.3	100	99.7	{Breast cancer, susceptibility to}, 114480
BLM	116.3	99.4	96.5	Bloom syndrome, 210900
BMPR1A	98	99.7	94.2	Juvenile polyposis syndrome, infantile form, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BRAF	74.4	87.6	77.2	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
BRCA1	177.5	98.9	96.9	Fanconi anemia, complementation group S, 617883 {Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	102.7	99	97.4	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	117.8	99.8	97.7	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054

BUB1	136.4	99.8	97.9	Colorectal cancer with chromosomal instability, somatic, 0
BUB1B	136.5	98.6	97.9	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
BUB3	146.1	98.6	97.8	No OMIM phenotype Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544)
CBL	129.8	96.9	95.7	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CDC73	102.7	99.8	97.7	Hyperparathyroidism, familial primary, 145000 Hyperparathyroidism-jaw tumor syndrome, 145001 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266
CDH1	125.3	99.1	98.4	Blepharocheilodontic syndrome 1, 119580 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	100	99.6	{Melanoma, cutaneous malignant, 3}, 609048
CDKN1A	145.9	100	100	No OMIM phenotype Multiple endocrine neoplasia 1 (Agarwal (2009) J Clin Endocrinol Metab 94, 1826) {Cancer, association with} (Mousses (1995) Hum Mol Genet 4, 1089) {Breast cancer, association with} (Staalesen (2006) Clin Cancer Res 12, 6000)
CDKN1B	93.2	100	99.5	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	21.1	68.1	51.8	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732
CDKN2A	79.6	92.2	91.4	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, 0 Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601

CDKN2B	85.9	100	99.8	No OMIM phenotype Renal cell carcinoma (Jafri (2015) Cancer Discov 5, 723) Multiple endocrine neoplasia 1 (Agarwal (2009) J Clin Endocrinol Metab 94, 1826) ?Melanoma (Foley (2015) EBioMedicine 2,74) ?Parathyroid adenoma (Costa-Guda (2013) Horm
CDKN2C	153.6	100	100	No OMIM phenotype MEN-1-like
CEBPA	46.7	75.5	65.1	?Leukemia, acute myeloid, 601626 Leukemia, acute myeloid, somatic, 601626
CHEK2	100.8	82.4	78.7	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast and colorectal cancer, susceptibility to}, 0 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807
CREBBP	123.5	99.4	96.7	Rubinstein-Taybi syndrome 1, 180849
CTC1	119	100	99.8	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTNNA1	125.6	99.9	99	Macular dystrophy, patterned, 2, 608970 Gastric cancer
CTR9	154.2	100	99.8	No OMIM phenotype Wilms tumor (Hanks (2014) Nat Commun 5, 4398)
CYLD	119.9	98.1	93	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
DDB2	162.4	100	99.7	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDX11	113.9	86	81	Warsaw breakage syndrome, 613398
DICER1	145.4	99.5	98.1	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200 Rhabdomyosarcoma, embryonal, 2, 180295
DIS3L2	158.5	99.8	99	Perlman syndrome, 267000
DKC1	111.9	99.6	98.1	Dyskeratosis congenita, X-linked, 305000
DNAJC21	125.7	99.8	98.5	Bone marrow failure syndrome 3, 617052

EGFR	160.8	100	99.1	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 {Nonsmall cell lung cancer, susceptibility to}, 211980
ELANE	80.9	99.7	95.9	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
EPCAM	64.5	93.3	79.8	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
ERCC1	76.6	100	97.3	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	123.7	100	99.7	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	113.2	99.9	98.9	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	139.2	100	99.5	?XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC5	139.8	100	99.4	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	191.3	100	99.9	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	133.7	100	99.5	?Ovarian dysgenesis 8, 618187 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
ETV6	140.1	100	99.9	Leukemia, acute myeloid, somatic, 601626

				Thrombocytopenia 5, 616216
EXT1	105.4	99.9	98.5	Chondrosarcoma, 215300 Exostoses, multiple, type 1, 133700
EXT2	163.5	99.9	99.1	?Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EZH2	139.5	99.8	97.6	Weaver syndrome, 277590
FAN1	143.9	100	99.9	Interstitial nephritis, karyomegalic, 614817 Colorectal cancer
FANCA	123.3	99.8	98.5	Fanconi anemia, complementation group A, 227650
FANCB	68.4	96.7	87.9	Fanconi anemia, complementation group B, 300514
FANCC	121.6	99.4	97.1	Fanconi anemia, complementation group C, 227645
FANCD2	127.6	98.7	95.5	Fanconi anemia, complementation group D2, 227646
FANCE	108	85.9	84.6	Fanconi anemia, complementation group E, 600901
FANCF	166.8	100	100	Fanconi anemia, complementation group F, 603467
FANCG	147.7	100	100	Fanconi anemia, complementation group G, 614082
FANCI	152.1	99.5	97.5	Fanconi anemia, complementation group I, 609053
FANCL	87.8	99.4	94.7	Fanconi anemia, complementation group L, 614083
FANCM	96.8	99.2	94.3	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAS	272.1	100	99.3	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0 {Autoimmune lymphoproliferative syndrome}, 601859
FH	146.4	91.7	87.6	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FLCN	160.5	100	99.5	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
G6PC3	123.7	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA2	119.6	99.9	98.5	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286

GDNF	185.9	99.9	98.8	Central hypoventilation syndrome, 209880 {Hirschsprung disease, susceptibility to, 3}, 613711 {Pheochromocytoma, modifier of}, 171300
GFI1	83.1	99	92.9	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, autosomal dominant, 613107
GPC3	85.1	98.6	92.6	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GREM1	95.1	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	134.6	100	100	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
HABP2	136.9	100	99.6	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HAX1	136.5	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HNF1A	156.7	100	99.4	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	127.8	99.9	97.7	{Prostate cancer, hereditary, 9}, 610997
IPMK	95.5	96.9	83.9	No OMIM phenotype Small intestinal carcinoid (Sei (2015) Gastroenterology 149,67)
KIF1B	154.8	100	99.5	?Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700

KIT	153	100	99.7	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, 601626 Mastocytosis, cutaneous, 154800 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KLLN	117.1	100	100	Cowden syndrome 4, 615107
KRAS	64.7	99.9	98.7	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LIG4	165.6	100	99.6	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
LZTR1	134	100	99.4	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
MAP2K1	92.3	99.8	95.6	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	107.9	97.6	89.2	Cardiofaciocutaneous syndrome 4, 615280
MAX	86.6	99.4	96.4	{Pheochromocytoma, susceptibility to}, 171300
MDH2	123.3	98	97.9	Epileptic encephalopathy, early infantile, 51, 617339
MEN1	123.2	99.5	96.3	Adrenal adenoma, somatic, 0 Angiofibroma, somatic, 0 Carcinoid tumor of lung, 0 Lipoma, somatic, 0 Multiple endocrine neoplasia 1, 131100

				Parathyroid adenoma, somatic, 0
MET	184.7	100	99.6	?Deafness, autosomal recessive 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 Renal cell carcinoma, papillary, 1, familial and somatic, 605074 {Osteofibrous dysplasia, susceptibility to}, 607278
MITF	155.5	100	99.9	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MLH1	162	100	99.7	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MPL	136.7	99.6	97.5	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MRE11	51.2	95.3	82.3	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	113.4	98.6	93.1	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSH3	113.4	99	95	Endometrial carcinoma, somatic, 608089 Familial adenomatous polyposis 4, 617100
MSH6	171.1	100	99.5	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300
MUC5B	98.5	87.7	82.4	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
MUTYH	165	100	99.9	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659

NBN	80.6	99.1	94.6	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NF1	125.9	92.3	89.3	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NF2	100.2	100	99.9	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, somatic, 162091
NHP2	111	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	160.5	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	70.3	88.7	77.9	Leukemia, acute myeloid, somatic, 601626
NRAS	188.4	100	100	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NSD1	155.2	100	99.9	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NTHL1	98.1	99.1	95.7	Familial adenomatous polyposis 3, 616415
OGG1	128.5	100	99.6	Renal cell carcinoma, clear cell, somatic, 144700
PALB2	152.6	100	99.7	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PARN	128.4	99.9	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	118.8	98.3	95.4	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545

PDGFB	95.1	100	100	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PHOX2B	92.9	93	87.2	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 Neuroblastoma with Hirschsprung disease, 613013 {Neuroblastoma, susceptibility to, 2}, 613013
PIK3CA	120.7	99.9	99.1	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PMS2	95.1	83.5	80.7	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMS2CL				No OMIM phenotype
POLD1	101.2	93.9	90.8	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLE	144.1	100	99.5	FILS syndrome, 615139 {Colorectal cancer, susceptibility to, 12}, 615083
POLH	140.7	100	99.8	Xeroderma pigmentosum, variant type, 278750
POT1	90.7	99.6	96	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
POU6F2	142.4	100	99.9	{Wilms tumor susceptibility-5}, 601583
PPM1D	166.7	100	99.8	Breast cancer, somatic, 114480 Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450

PRF1	122.5	91.2	90.8	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRKAR1A	90.7	99.1	93.9	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PRKN	98.6	79.6	78.8	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PRSS1	190.8	100	99.9	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044
PTCH1	114.6	98.4	95.9	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	120.1	99.4	97.5	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTEN	143.2	99.6	96	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTPN11	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
RAD50	99	92.6	86.2	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	143.4	100	98.9	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399

RAD51D	153.9	100	99.5	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RAF1	127.3	100	99.7	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RB1	88	90.1	76.3	Bladder cancer, somatic, 109800 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200 Retinoblastoma, trilateral, 180200 Small cell cancer of the lung, somatic, 182280
RECQL	127.4	99.7	96.1	No OMIM phenotype Breast cancer (Cybulski (2015) Nat Genet 47,643)
RECQL4	149.6	99.2	96.5	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
REST	128.6	98.5	98.4	Fibromatosis, gingival, 5, 617626 {Wilms tumor 6, susceptibility to}, 616806
RET	141	99.7	97.8	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, protection against}, 142623 {Hirschsprung disease, susceptibility to, 1}, 142623
RHBDF2	97.7	99.5	97	Tylosis with esophageal cancer, 148500
RINT1	171	99.7	97.5	No OMIM phenotype ?Breast cancer (Park (2014) Cancer Discov 4, 804)
RIT1	165.6	100	100	Noonan syndrome 8, 615355
RNF43	126.5	100	99.6	Sessile serrated polyposis cancer syndrome, 617108
RPL11	99.8	100	99.5	Diamond-Blackfan anemia 7, 612562
RPL35A	83.9	99.2	91	Diamond-Blackfan anemia 5, 612528
RPL5	43.8	81.8	69.3	Diamond-Blackfan anemia 6, 612561
RPS10	140.1	99.8	97.1	Diamond-Blackfan anemia 9, 613308

RPS17	52.4	85	73.7	Diamond-Blackfan anemia 4, 612527
RPS19	82.5	99.7	95.5	Diamond-Blackfan anemia 1, 105650
RPS20	74.7	99.7	94.1	No OMIM phenotype Colorectal cancer, non-polyposis (Nieminen (2014) Gastroenterology 147,595)
RPS24	110.4	92.4	87.2	Diamond-blackfan anemia 3, 610629
RPS26	106.8	94.8	82.1	Diamond-Blackfan anemia 10, 613309
RPS7	93.7	76.9	63.4	Diamond-Blackfan anemia 8, 612563
RTEL1	110.9	99.2	95.1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RUNX1	92	97.2	89.7	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SAMD9	159.1	99.9	99.3	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	165.7	100	99.9	Ataxia-pancytopenia syndrome, 159550
SBDS	212.3	100	99.9	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SDHA	122.2	84.8	80.8	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF2	144.3	94.7	94.3	Paragangliomas 2, 601650
SDHB	120.3	100	99.3	Gastrointestinal stromal tumor, 606444 Paragangliolas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SDHC	100.1	99.8	96.8	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 3, 605373
SDHD	48.4	55.2	50.4	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300

SEMA4A	127.5	99.9	98.9	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SFTPA1	175.5	99.8	98.3	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500
SFTPA2	167.9	100	99.1	Pulmonary fibrosis, idiopathic, 178500
SH2B3	97.5	90.7	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SHOC2	140.4	100	99.4	Noonan-like syndrome with loose anagen hair, 607721
SLX4	114.2	100	99.8	Fanconi anemia, complementation group P, 613951
SMAD4	125.5	100	100	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
SMAD9	132.8	100	100	Pulmonary hypertension, primary, 2, 615342
SMARCA4	143.8	100	99.5	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCB1	214.3	100	100	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid tumor predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMARCE1	73.6	96.5	86.8	Coffin-Siris syndrome 5, 616938 {Meningioma, familial, susceptibility to}, 607174
SOS1	94.3	96.7	90.3	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SPINK1	80.2	100	99.4	Pancreatitis, hereditary, 167800 Tropical calcific pancreatitis, 608189 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPRED1	164.3	98.7	96.7	Legius syndrome, 611431
SQSTM1	109.1	98.6	94.5	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145

				Paget disease of bone 3, 167250
STK11	111.9	99.7	95.8	Melanoma, malignant, somatic, 0 Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
SUFU	122.6	99.9	99	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 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	116.7	100	97.6	No OMIM phenotype Melanoma (Aoude (2015) J Natl Cancer Inst 107) Chronic lymphocytic leukaemia (Speedy (2016) Blood 128,2319)
TERT	138.3	95.3	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	184	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMEM127	107.4	98.2	94.7	{Pheochromocytoma, susceptibility to}, 171300
TNFRSF11A	146.3	93.3	91.4	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080

TP53	92	99.9	98.1	Adrenocortical carcinoma, pediatric, 202300 Bone marrow failure syndrome 5, 618165 Breast cancer, somatic, 114480 Choroid plexus papilloma, 260500 Colorectal cancer, 114500 Hepatocellular carcinoma, somatic, 114550 Li-Fraumeni syndrome, 151623 Nasopharyngeal carcinoma, somatic, 607107 Osteosarcoma, 259500 Pancreatic cancer, somatic, 260350 {Basal cell carcinoma 7}, 614740 {Glioma susceptibility 1}, 137800
TRIP13	141.2	100	100	Mosaic variegated aneuploidy syndrome 3, 617598
TSC1	128.8	99.8	98.8	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	131.2	100	99	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
USB1	125	99.9	98.2	Poikiloderma with neutropenia, 604173
VHL	119.7	92.6	85.3	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
WAS	66.1	88.2	78.7	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WRAP53	154.4	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	123.6	98.3	94.6	Werner syndrome, 277700

WT1	76.5	91.8	81.4	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
XPA	52.9	98.5	88.9	Xeroderma pigmentosum, group A, 278700
XPC	140.7	100	99.7	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 : December 31st, 2018.
This list is accurate for panel version DG 2.15
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors