HEREDITARY CANCER GENE PANEL

Gene symbol	*Depth (reads)*	*Coverage (avg %)*	*OMIM disease*	*Description*
ALK	99	86	613014	{Neuroblastoma, susceptibility to, 3}
APC	99	99	175100	Adenomatous polyposis coli
ATM	99	93	208900	Ataxia-telangiectasia
ATR	99	94	614564	Cutaneous telangiectasia and cancer syndrome familial
BAP1	99	86	614327	Tumor predisposition syndrome
BLM	99	89	210900	Bloom syndrome
BMPR1A	99	40	174900	Juvenile polyposis syndrome infantile form
BRCA1	99	97	604370	{Breast-ovarian cancer, familial, 1}
BRCA2	99	97	605724	Fanconi anemia complementation group D1
BRIP1	99	95	114480	Breast cancer early-onset
BUB1	99	88	200	Colorectal cancer with chromosomal instability
BUB1B	99	94	114500	Colorectal cancer somatic
CDC73	99	93	145000	Hyperparathyroidism familial primary
CDH1	99	83	608089	Endometrial carcinoma somatic
CDK4	99	90	609048	{Melanoma, cutaneous malignant, 3}
CDKN2A	99	66	155755	Melanoma and neural system tumor syndrome
CENPJ	99	96	608393	Microcephaly 6 primary autosomal recessive
CHEK2	99	86	609265	Li-Fraumeni syndrome
CREBBP	99	92	180849	Rubinstein-Taybi syndrome
CYLD	99	96	605041	Brooke-Spiegler syndrome
DDB2	99	85	278740	Xeroderma pigmentosum group E DDB-negative subtype
DICER1	99	93	138800	Goiter multinodular 1 with or without Sertoli-Leydig cell tumors
DKC1	99	94	305000	Dyskeratosis congenita X-linked
EGFR	99	86	211980	Adenocarcinoma of lung response to tyrosine kinase inhibitor in
ELANE	99	83	162800	Neutropenia cyclic
ERCC1	99	96	610758	Cerebrooculofacioskeletal syndrome 4
ERCC2	99	89	610756	Cerebrooculofacioskeletal syndrome 2
ERCC3	99	93	601675	Trichothiodystrophy

ERCC4	99	92	615272	Fanconi anemia complementation group Q
ERCC5	99	94	278780	Xeroderma pigmentosum group G
ERCC6	99	92	214150	Cerebrooculofacioskeletal syndrome 1
EXO1	99	93	200	-
EXT1	99	90	215300	Chondrosarcoma
EXT2	99	80	133701	Exostoses multiple type 2
FANCA	99	89	227650	Fanconi anemia complementation group A
FANCB	99	96	300514	Fanconi anemia, complementation group B
FANCC	99	91	227645	Fanconi anemia complementation group C
FANCD2	99	84	227646	Fanconi anemia complementation group D2
FANCE	99	91	600901	Fanconi anemia complementation group E
FANCF	99	100	603467	Fanconi anemia complementation group F
FANCG	99	86	614082	Fanconi anemia, complementation group G
FANCI	99	92	609053	Fanconi anemia complementation group I
FANCL	99	92	614083	Fanconi anemia, complementation group L
FANCM	99	94	614087	Fanconi anemia complementation group M
FH	99	80	606812	Fumarase deficiency
FLCN	99	91	135150	Birt-Hogg-Dube syndrome
G6PC3	99	86	612541	Dursun syndrome
GFI1	99	86	607847	Neutropenia nonimmune chronic idiopathic of adults
GPC3	99	94	312870	Simpson-Golabi-Behmel syndrome type 1
GREM1	99	83	601228	mixed polyposis syndrome
HAX1	99	100	610738	Neutropenia severe congenital 3 autosomal recessive
HNF1A	99	91	612520	Diabetes mellitus insulin-dependent 20
KIT	99	90	606764	Gastrointestinal stromal tumor familial
KLLN	99	93	615107	Cowden syndrome 4
MAX	99	94	171300	{Pheochromocytoma, susceptibility to}
MEN1	99	85	200	Adrenal adenoma
MLH1	99	88	609310	Colorectal cancer hereditary nonpolyposis type 2
MLH3	99	94	614385	Colon cancer hereditary nonpolyposis type 7
MPL	99	88	254450	Myelofibrosis with myeloid metaplasia somatic
MSH2	100	91	120435	Colorectal cancer hereditary nonpolyposis type 1
MSH6	100	98	614350	Colorectal cancer hereditary nonpolyposis type 5

MUTYH	100	94	608456	Adenomas multiple colorectal
NBN	100	94	251260	Nijmegen breakage syndrome
NBN	100	94	613065	Leukemia, acute lymphoblastic
NBN	100	94	609135	Aplastic anemia
NF1	100	85	607785	Leukemia juvenile myelomonocytic
NF2	100	94	607174	Meningioma NF2-related somatic
NHP2	100	46	613987	Dyskeratosis congenita, autosomal recessive 2
NOP10	99	100	224230	Dyskeratosis congenita, autosomal recessive 1
NSD1	99	92	130650	Beckwith-Wiedemann syndrome
OGG1	99	91	144700	Renal cell carcinoma clear cell somatic
PALB2	99	95	610832	Fanconi anemia complementation group N
PHOX2B	99	96	613013	Neuroblastoma with Hirschsprung disease
PMS1	99	97	200	-
PMS2	99	36	614337	Colorectal cancer hereditary nonpolyposis type 4
PMS2CL	99	27	200	-
PRF1	99	100	603553	Hemophagocytic lymphohistiocytosis familial 2
PRKAR1A	99	78	101800	Acrodysostosis 1 with or without hormone resistance
PTCH1	99	48	605462	Basal cell carcinoma somatic
PTEN	99	65	153480	Bannayan-Riley-Ruvalcaba syndrome
PTPRJ	99	87	114500	Colon cancer somatic
RB1	99	95	109800	Bladder cancer somatic
RECQL4	99	97	218600	Baller-Gerold syndrome
RET	99	85	209880	Central hypoventilation syndrome congenital
RPL11	99	61	612562	Diamond-Blackfan anemia 7
RPL35A	99	21	612528	Diamond-Blackfan anemia 5
RPL5	99	30	612561	Diamond-Blackfan anemia 6
RPS10	99	29	613308	Diamond-Blackfan anemia 9
RPS19	99	44	105650	Diamond-Blackfan anemia 1
RPS24	99	58	610629	Diamond-blackfan anemia 3
RPS26	99	26	613309	Diamond-Blackfan anemia 10
RPS7	99	22	612563	Diamond-Blackfan anemia 8
RUNX1	99	83	601626	Leukemia acute myeloid
SBDS	99	70	260400	Shwachman-Bodian-Diamond syndrome

SDHA	99	18	613642	Cardiomyopathy dilated 1GG
SDHAF2	99	83	200	Paragangliomas 2
SDHB	99	87	612359	Cowden syndrome 2
SDHC	99	88	606764	Gastrointestinal stromal tumor
SDHD	99	13	114900	Carcinoid tumors intestinal
SMAD4	99	93	139210	Myhre syndrome
SMAD4	99	93	260350	Pancreatic cancer, somatic
SMARCB1	99	84	614608	Mental retardation autosomal dominant 15
STK11	99	89	200	Melanoma malignant
SUFU	99	89	155255	Medulloblastoma desmoplastic
TERC	99	100	127550	Dyskeratosis congenita autosomal dominant 1
TERT	99	89	613989	{Dyskeratosis congenita}
TERT	99	89	614742	{Bone marrow failure, telomere-related, 1}
TERT	99	89	601626	{Leukemia, acute myeloid}
TINF2	99	92	613990	Dyskeratosis congenita autosomal dominant 3
TMEM127	99	100	171300	{Pheochromocytoma, susceptibility to}
TP53	99	95	202300	Adrenal cortical carcinoma
TSC1	99	87	607341	Focal cortical dysplasia Taylor balloon cell type
TSC2	99	92	606690	Lymphangioleiomyomatosis somatic
VHL	99	95	263400	Erythrocytosis familial 2
WAS	99	85	300299	Neutropenia severe congenital X-linked
WRN	99	96	277700	Werner syndrome
WT1	99	91	194080	Denys-Drash syndrome
XPA	99	89	278700	Xeroderma pigmentosum group A
XPC	99	90	278720	Xeroderma pigmentosum group C

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors