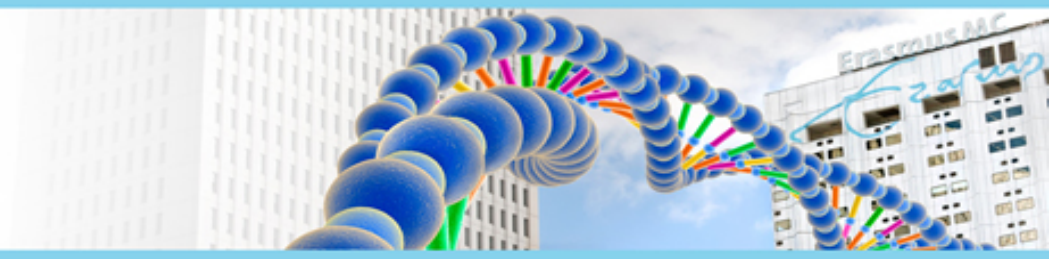
Whole Exome Sequencing
Gene package Oncogenetics, version 2.2, 31-1-2020



Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
A2ML1	No OMIM phenotype	610627	44	100	97	80
ACD	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553	609377	92	100	100	100
AIP	Pituitary adenoma, growth hormone-secreting, 102200 Pituitary adenoma, prolactin-secreting, 600634 Pituitary adenoma, ACTH-secreting, 219090	605555	78	100	100	100
AKT1	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Ovarian cancer, somatic, 167000 {Schizophrenia, susceptibility to}, 181500 Proteus syndrome, somatic, 176920 Cowden syndrome 6, 615109	164730	72	100	100	97
ALK	{Neuroblastoma, susceptibility to, 3}, 613014	105590	74	100	99	93

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APC	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	611731	130	100	100	100
ARMC5	ACTH-independent macronodular adrenal hyperplasia 2, 615954	615549	88	100	100	100
ATM	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic	607585	55	100	97	83
ATR	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564	601215	58	100	96	78
AXIN2	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500	604025	83	100	100	99
BAP1	Tumor predisposition syndrome, 614327	603089	81	100	100	99
BARD1	{Breast cancer, susceptibility to}, 114480	601593	55	100	100	91
BLM	Bloom syndrome, 210900	604610	62	100	98	90
BMPR1A	Polyposis, juvenile intestinal, 174900 Polyposis syndrome, hereditary mixed, 2, 610069 Juvenile polyposis syndrome, infantile form, 174900	601299	61	100	100	98
BRAF	Melanoma, malignant, somatic Colorectal cancer, somatic Adenocarcinoma of lung, somatic, 211980 Nonsmall cell lung cancer, somatic Cardiofaciocutaneous syndrome, 115150 Noonan syndrome 7, 613706 LEOPARD syndrome 3, 613707	164757	53	100	98	84
BRCA1	{Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320	113705	153	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
BRCA2	{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	600185	143	100	100	100
BRIP1	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054	605882	43	100	96	77
BUB1	Colorectal cancer with chromosomal instability, somatic	602452	58	100	98	88
BUB1B	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430	602860	51	100	98	86
BUB3	No OMIM phenotype	603719	48	100	97	85
CASR	Hypocalciuric hypercalcemia, type I, 145980 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypercalciuric hypercalcemia {Calcium, serum level of}	601199	85	100	100	99
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	165360	54	100	100	97
CDC73	Hyperparathyroidism-jaw tumor syndrome, 145001 Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266	607393	56	100	99	84
CDH1	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	192090	60	100	99	95
CDK4	{Melanoma, cutaneous malignant, 3}, 609048	123829	62	100	100	99
CDKN1A	No OMIM phenotype	116899	109	100	100	100
CDKN1B	Multiple endocrine neoplasia, type IV, 610755	600778	126	100	100	100

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CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	56	91	83	74
CDKN2A	{Melanoma, cutaneous malignant, 2}, 155601 Melanoma and neural system tumor syndrome, 155755 Pancreatic cancer/melanoma syndrome, 606719 Orolaryngeal cancer, multiple,	600160	80	100	100	100
CDKN2B	No OMIM phenotype	600431	53	100	100	100
CDKN2C	No OMIM phenotype	603369	66	100	100	100
CEBPA	?Leukemia, acute myeloid, 601626 Leukemia, acute myeloid, somatic, 601626	116897	69	100	87	70
CFTR	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF {Pancreatitis, idiopathic}, 167800 {Hypertrypsinemia, neonatal} {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400	602421	69	100	99	92
CHEK2	Li-Fraumeni syndrome, 609265 Osteosarcoma, somatic, 259500 {Breast cancer, susceptibility to}, 114480 {Prostate cancer, familial, susceptibility to}, 176807 {Breast and colorectal cancer, susceptibility to}	604373	58	100	92	73
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, localisata variant, 226650 Epithelial recurrent erosion dystrophy, 122400	113811	65	100	100	95
CREBBP	Rubinstein-Taybi syndrome, 180849	600140	81	100	99	91
CTNNA1	No OMIM phenotype	116805	65	100	99	93
CYLD	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606	605018	44	100	96	80
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740	600811	67	100	100	95
DICER1	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Rhabdomyosarcoma, embryonal, 2, 180295	606241	60	100	98	90
DIS3L2	Perlman syndrome, 267000	614184	57	100	98	90
DKC1	Dyskeratosis congenita, X-linked, 305000	300126	45	100	96	83

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DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700	611432	49	100	94	81
EGFR	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 ?Inflammatory skin and bowel disease, neonatal, 2, 616069	131550	56	100	100	92
EGLN1	Erythrocytosis, familial, 3, 609820 [Hemoglobin, high altitude adaptation], 609070	606425	81	100	100	93
ELANE	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700	130130	67	100	100	98
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758	126380	52	100	100	95
ERCC2	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 Cerebrooculofacioskeletal syndrome 2, 610756	126340	75	100	100	99
ERCC3	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390	133510	56	100	99	89
ERCC4	Xeroderma pigmentosum, group F, 278760 ?XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760	133520	58	100	99	88
ERCC5	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Cerebrooculofacioskeletal syndrome 3, 616570	133530	65	100	99	94
ERCC6	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	609413	60	100	98	92
EXO1	No OMIM phenotype	606063	51	100	98	88
EXT1	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300	608177	51	100	97	89
EXT2	Exostoses, multiple, type 2, 133701 ?Seizures, scoliosis, and macrocephaly syndrome, 616682	608210	68	100	99	89
FAN1	Interstitial nephritis, karyomegalic, 614817	613534	58	100	100	93
FANCA	Fanconi anemia, complementation group A, 227650	607139	69	100	98	89

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FANCB	Fanconi anemia, complementation group B, 300514	300515	46	100	98	85
FANCC	Fanconi anemia, complementation group C, 227645	613899	47	100	94	75
FANCD2	Fanconi anemia, complementation group D2, 227646	613984	53	100	98	87
FANCE	Fanconi anemia, complementation group E, 600901	613976	80	100	100	92
FANCF	Fanconi anemia, complementation group F, 603467	613897	114	100	100	100
FANCG	Fanconi anemia, complementation group G, 614082	602956	88	100	100	100
FANCI	Fanconi anemia, complementation group I, 609053	611360	52	100	98	87
FANCL	Fanconi anemia, complementation group L, 614083	608111	43	100	94	72
FANCM	No OMIM phenotype	609644	64	100	98	91
FAS	{Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic Autoimmune lymphoproliferative syndrome, type IA, 601859	134637	109	100	99	84
FH	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800	136850	57	98	93	88
FLCN	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500	607273	72	100	100	100
Focad	No OMIM phenotype	614606	44	99	92	75
G6PC3	Neutropenia, severe congenital 4, autosomal recessive, 612541 Dursun syndrome, 612541	611045	72	100	100	100
GATA1	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835	305371	54	100	99	94
GATA2	Immunodeficiency 21, 614172 Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 {Leukemia, acute myeloid, susceptibility to}, 601626	137295	86	100	100	100
GDNF	Central hypoventilation syndrome, 209880 {Pheochromocytoma, modifier of}, 171300 {Hirschsprung disease, susceptibility to, 3}, 613711	600837	66	100	100	100
GFI1	Neutropenia, severe congenital 2, autosomal dominant, 613107 Neutropenia, nonimmune chronic idiopathic, of adults, 607847	600871	90	100	100	100

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GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	42	100	94	79
GREM1	No OMIM phenotype	603054	62	100	100	100
HABP2	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050	603924	57	100	97	87
HAX1	Neutropenia, severe congenital 3, autosomal recessive, 610738	605998	79	100	100	99
HNF1A	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	142410	75	100	100	99
HNF1B	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853 {Renal cell carcinoma}, 144700	189907	77	100	100	99
HOXB13	No OMIM phenotype	604607	115	100	100	100
HRAS	{Bladder cancer, somatic}, 109800 Costello syndrome, 218040 {Thyroid carcinoma, follicular, somatic}, 188470 Congenital myopathy with excess of muscle spindles, 218040 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Spitz nevus or nevus spilus, somatic}, 137550	190020	105	100	100	100
ITK	Lymphoproliferative syndrome 1, 613011	186973	53	100	98	79
KIF1B	?Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700	605995	48	100	98	85
KIT	Piebaldism, 172800 Gastrointestinal stromal tumor, familial, 606764 Mast cell disease, 154800 Leukemia, acute myeloid, 601626 Germ cell tumors, 273300	164920	56	100	100	96
KLLN	Cowden syndrome 4, 615107	612105	77	100	100	100

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KRAS	Lung cancer, somatic, 211980 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 Breast cancer, somatic, 114480 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 RAS-associated autoimmune leukoproliferative disorder, 614470	190070	61	100	97	64
LZTR1	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 10, 616564	600574	80	100	100	100
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	55	100	100	91
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	601263	69	100	92	86
MAX	{Pheochromocytoma, susceptibility to}, 171300	154950	47	100	100	92
MBD4	No OMIM phenotype	603574	72	100	100	95
MC1R	[Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 [Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 [Analgesia from kappa-opioid receptor agonist, female-specific], 613098 {UV-induced skin damage}, 266300 {Albinism, oculocutaneous, type II, modifier of}, 203200 {Melanoma, cutaneous malignant, 5}, 613099	155555	133	100	100	100
MEN1	Multiple endocrine neoplasia 1, 131100 Carcinoid tumor of lung Parathyroid adenoma, somatic Lipoma, somatic Angiofibroma, somatic Adrenal adenoma, somatic	613733	111	100	100	100
MET	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705	164860	47	100	98	88
MITF	Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456	156845	58	100	100	90

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MLH1	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320	120436	122	100	100	100
MLH3	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 Colorectal cancer, somatic, 114500 {Endometrial cancer, susceptibility to}, 608089	604395	53	100	100	97
MPL	Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977 Myelofibrosis with myeloid metaplasia, somatic, 254450	159530	64	100	100	98
MRE11	Ataxia-telangiectasia-like disorder, 604391	600814	43	100	91	72
MSH2	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300	609309	112	100	100	100
MSH3	Endometrial carcinoma, somatic, 608089	600887	55	100	98	87
MSH6	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Endometrial cancer, familial, 608089 Mismatch repair cancer syndrome, 276300	600678	148	100	100	100
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250	156540	51	100	97	78
MUTYH	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600	604933	114	100	100	100
NBN	Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065	602667	52	100	95	76
NF1	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520	613113	41	98	87	67
NF2	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, 162091	607379	84	100	100	100
NHP2	Dyskeratosis congenita, autosomal recessive 2, 613987	606470	66	100	100	100
NOP10	Dyskeratosis congenita, autosomal recessive 1, 224230	606471	121	100	100	100

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NOTCH2	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500	600275	42	100	100	91
NRAS	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200	164790	42	100	100	91
NSD1	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 Beckwith-Wiedemann syndrome, 130650	606681	56	100	99	92
NTHL1	Familial adenomatous polyposis 3, 616415	602656	77	100	100	100
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240	191315	70	100	100	98
OGG1	Renal cell carcinoma, clear cell, somatic, 144700	601982	68	100	100	100
PALB2	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348	610355	53	100	98	91
PALLD	{Pancreatic cancer, susceptibility to, 1}, 606856	608092	48	100	98	84
PAX5	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545	167414	74	100	95	93
PDGFRA	Gastrointestinal stromal tumor, somatic, 606764 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685	173490	55	99	96	87
PHOX2B	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 {Neuroblastoma, susceptibility to, 2}, 613013 Neuroblastoma with Hirschsprung disease, 613013	603851	97	100	100	100

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PIK3CA	Ovarian cancer, somatic, 167000 Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Nonsmall cell lung cancer, somatic, 211980 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108	171834	65	100	99	92
PMS2	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337	600259	106	100	100	99
POLD1	{Colorectal cancer, susceptibility to, 10}, 612591 Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381	174761	67	100	97	93
POLE	{Colorectal cancer, susceptibility to, 12}, 615083 FILS syndrome, 615139	174762	78	100	100	96
POLH	Xeroderma pigmentosum, variant type, 278750	603968	53	100	99	91
POT1	{Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 {Glioma susceptibility 9}, 616568	606478	53	100	98	82
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027 Aplastic anemia, 609135	170280	87	100	100	100
PRKAR1A	Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489 Adrenocortical tumor, somatic, Acrodysostosis 1, with or without hormone resistance, 101800	188830	64	100	100	94
PRSS1	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044	276000	142	100	100	100
PTCH1	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828	601309	70	100	97	92

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PTCH2	Medulloblastoma, 155255 Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400	603673	84	100	100	100
PTEN	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 Malignant melanoma, somatic, 155600 Endometrial carcinoma, somatic, 608089 Squamous cell carcinoma, head and neck, somatic, 275355	601728	85	92	81	77
PTPN11	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250	176876	57	100	96	80
RAD50	Nijmegen breakage syndrome-like disorder, 613078	604040	71	100	100	95
RAD51B	No OMIM phenotype	602948	47	100	99	90
RAD51C	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399	602774	49	100	98	83
RAD51D	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291	602954	64	100	100	98
RAF1	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554 Cardiomyopathy, dilated, 1NN, 615916	164760	54	100	95	87
RASAL1	No OMIM phenotype	604118	62	100	96	89
RB1	Retinoblastoma, 180200 Osteosarcoma, somatic, 259500 Bladder cancer, somatic, 109800 Small cell cancer of the lung, somatic, 182280 Retinoblastoma, trilateral, 180200	614041	104	100	100	99
RECQL	No OMIM phenotype	600537	72	100	98	90

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RECQL4	Rothmund-Thomson syndrome, 268400 RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600	603780	95	100	100	98
REST	{Wilms tumor 6, susceptibility to}, 616806	600571	57	100	100	93
RET	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	164761	92	100	100	99
RHBDF2	Tylosis with esophageal cancer, 148500	614404	77	100	100	100
RINT1	No OMIM phenotype	610089	44	100	96	80
RIT1	Noonan syndrome 8, 615355	609591	53	100	100	94
RMRP	Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460 Anauxetic dysplasia, 607095	157660	No coverage data	0	0	0
RNF43	Sessile serrated polyposis cancer syndrome, 617108	612482	105	100	100	99
RPL11	Diamond-Blackfan anemia 7, 612562	604175	44	100	100	94
RPL15	?Diamond-Blackfan anemia 12, 615550	604174	43	100	100	87
RPL35A	Diamond-Blackfan anemia 5, 612528	180468	73	100	100	100
RPL5	Diamond-Blackfan anemia 6, 612561	603634	50	100	96	84
RPS10	Diamond-Blackfan anemia 9, 613308	603632	46	100	98	86
RPS17	Diamond-Blackfan anemia 4, 612527	180472	74	100	100	90
RPS19	Diamond-Blackfan anemia 1, 105650	603474	72	100	100	95
RPS24	Diamond-blackfan anemia 3, 610629	602412	76	100	100	99
RPS26	Diamond-Blackfan anemia 10, 613309	603701	70	100	100	98
RPS29	Diamond-Blackfan anemia 13, 615909	603633	54	100	100	100
RPS7	Diamond-Blackfan anemia 8, 612563	603658	49	100	100	91
RTEL1	Dyskeratosis congenita, autosomal recessive 5, 615190 Dyskeratosis congenita, autosomal dominant 4, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373	608833	80	100	100	98
RUNX1	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399	151385	71	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SBDS	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135	607444	49	100	100	92
SDHA	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165	600857	93	100	93	83
SDHAF2	Paragangliomas 2, 601650	613019	91	100	100	93
SDHB	Paragangliomas 4, 115310 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764	185470	80	100	100	100
SDHC	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764	602413	113	100	100	100
SDHD	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 Mitochondrial complex II deficiency, 252011	602690	111	100	100	100
SERPINA1	Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Hemorrhagic diathesis due to \'antithrombin\' Pittsburgh, 613490 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963	107400	65	100	100	93
SFTPA1	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500	178630	136	100	100	100
SFTPA2	Pulmonary fibrosis, idiopathic, 178500	178642	127	100	100	100
SH2D1A	Lymphoproliferative syndrome, X-linked, 1, 308240	300490	44	100	99	79
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	602775	48	100	98	85
SLX4	Fanconi anemia, complementation group P, 613951	613278	87	100	100	98
SMAD4	Pancreatic cancer, somatic, 260350 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900	600993	59	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SMAD9	Pulmonary hypertension, primary, 2, 615342	603295	67	100	100	98
SMARCA4	{Rhabdoid tumor predisposition syndrome 2}, 613325 Mental retardation, autosomal dominant 16, 614609	603254	76	100	100	98
SMARCB1	Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 Mental retardation, autosomal dominant 15, 614608 {Schwannomatosis-1, susceptibility to}, 162091	601607	71	100	100	95
SMARCE1	{Meningioma, familial, susceptibility to}, 607174	603111	49	100	100	92
SOS1	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733	182530	57	100	98	84
SPINK1	Pancreatitis, hereditary, 167800 {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 Tropical calcific pancreatitis, 608189	167790	70	100	100	83
SPRED1	Legius syndrome, 611431	609291	44	100	95	84
SRY	46XY sex reversal 1, 400044 46XX sex reversal 1, 400045	480000	20	50	41	35
STAT3	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 615952	102582	60	100	100	91
STK11	Peutz-Jeghers syndrome, 175200 Melanoma, malignant, somatic Pancreatic cancer, 260350 Testicular tumor, somatic, 273300	602216	83	100	100	100
SUFU	Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174 Basal cell nevus syndrome, 109400	607035	60	100	100	97
TERC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743	602322	No coverage data	0	0	0
TERF1	No OMIM phenotype	600951	39	96	79	60
TERF2IP	No OMIM phenotype	605061	65	100	99	90
TERT	{Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134	187270	85	100	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TGFBR1	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800	190181	96	95	93	92
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168	190182	124	100	100	100
TINF2	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130	604319	94	100	100	100
TMEM127	{Pheochromocytoma, susceptibility to}, 171300	613403	61	100	100	96
TNFRSF11A	Osteolysis, familial expansile, 174810 {Paget disease of bone 2, early-onset}, 602080 Osteopetrosis, autosomal recessive 7, 612301	603499	66	95	95	91
TP53	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	191170	99	100	100	100
TRIM37	Mulibrey nanism, 253250	605073	44	100	97	80
TSC1	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341	605284	105	100	100	100
TSC2	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690	191092	104	100	100	100
UROD	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100	613521	70	100	100	98
USB1	Poikiloderma with neutropenia, 604173	613276	47	100	99	85

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
VHL	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400	608537	88	100	100	100
WAS	Wiskott-Aldrich syndrome, 301000 Thrombocytopenia, X-linked, 313900 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900	300392	54	96	84	75
WRAP53	Dyskeratosis congenita, autosomal recessive 3, 613988	612661	95	100	100	100
WRN	Werner syndrome, 277700	604611	47	100	98	84
WT1	Wilms tumor, type 1, 194070 Denys-Drash syndrome, 194080 Nephrotic syndrome, type 4, 256370 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240	607102	80	100	98	96
XPA	Xeroderma pigmentosum, group A, 278700	611153	45	100	92	70
XPC	Xeroderma pigmentosum, group C, 278720	613208	66	100	98	90
XRCC2	No OMIM phenotype	600375	63	100	100	100

- Gene symbols according HGCN
- OMIM release used: 17-3-2016
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 96 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x