

Pediatric Oncopredisposition

Gene panel

Gene panel information

Gene panel	Pediatric Oncopredisposition
Version	4
Total genes	197
Activation date	Thursday 21 march 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
A2ML1	99.75 %	610627	{Otitis media, susceptibility to}, 166760 (3), Autosomal dominant
ABCB11	99.58 %	603201	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive
ACD	99.7 %	609377	?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant
AIP	99.84 %	605555	Pituitary adenoma 1, multiple types, 102200 (3), Somatic mutation, Autosomal dominant; Pituitary adenoma predisposition, 102200 (3), Somatic mutation, Autosomal dominant
ALK	99.52 %	105590	{Neuroblastoma, susceptibility to, 3}, 613014 (3)
AMER1	99.95 %	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
APC	99.98 %	611731	Colorectal cancer, somatic, 114500 (3); Brain tumor-polypoid syndrome 2, 175100 (3), Autosomal dominant; Desmoid disease, hereditary, 135290 (3), Autosomal dominant; Adenoma, periampullary, somatic, 175100 (3); Hepatoblastoma, somatic, 114550 (3); Gastric cancer, somatic, 613659 (3); Gastric adenocarcinoma and proximal polyposis of the stomach, 619182 (3), Autosomal dominant; Gardner syndrome, 175100 (3), Autosomal dominant; Adenomatous polyposis coli, 175100 (3), Autosomal dominant
ASXL1	99.93 %	612990	Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant
ATM	99.88 %	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
ATRX	99.57 %	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive; Alpha-thalassemia/impaired intellectual development syndrome, 301040 (3), X-linked dominant
BAP1	99.86 %	603089	Kury-Isidor syndrome, 619762 (3), Autosomal dominant; Tumor predisposition syndrome 1, 614327 (3), Autosomal dominant; {Uveal melanoma, susceptibility to, 2}, 606661 (3), Autosomal dominant
BLM	99.87 %	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BMPR1A	98.88 %	601299	Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant
BRAF	99.53 %	164757	Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Non-small cell lung cancer, somatic, 211980 (3)

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Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
BRCA1	99.73 %	113705	Fanconi anemia, complementation group S, 617883 (3), Autosomal recessive; {Breast-ovarian cancer, familial, 1}, 604370 (3), Autosomal dominant; {Pancreatic cancer, susceptibility to, 4}, 614320 (3)
BRCA2	99.97 %	600185	Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant; {Prostate cancer}, 176807 (3), Somatic mutation, Autosomal dominant; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant; {Breast cancer, male, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; {Pancreatic cancer 2}, 613347 (3); Wilms tumor, 194070 (3), Somatic mutation, Autosomal dominant
BRIP1	99.53 %	605882	Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant
BUB1B	99.99 %	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
CASP10	99.48 %	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
CBL	99.82 %	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
CD27	99.95 %	186711	Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive
CD70	99.89 %	602840	Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive
CDC73	98.96 %	607393	Hyperparathyroidism, familial primary, 145000 (3), Autosomal dominant; Parathyroid adenoma with cystic changes, 145001 (3), Autosomal dominant; Parathyroid carcinoma, 608266 (3); Hyperparathyroidism-jaw tumor syndrome, 145001 (3), Autosomal dominant
CDH1	99.69 %	192090	Ovarian cancer, somatic, 167000 (3); Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); Breast cancer, lobular, somatic, 114480 (3)
CDK4	99.45 %	123829	{Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant
CDKN1B	99.99 %	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant
CDKN1C	99.99 %	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CDKN2A	99.76 %	600160	{Melanoma and neural system tumor syndrome}, 155755 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 2}, 155601 (3), Autosomal dominant; {Melanoma-pancreatic cancer syndrome}, 606719 (3), Autosomal dominant
CEBPA	99.48 %	116897	Leukemia, acute myeloid, somatic, 601626 (3); ?Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant
CEP57	99.81 %	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
CLPB	99.82 %	616254	Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant
CREBBP	99.79 %	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CTC1	99.9 %	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTLA4	99.88 %	123890	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100 (3), Autosomal dominant

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Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CTR9	99.9 %	609366	No OMIM phenotypes
CYLD	99.49 %	605018	Brooke-Spiegler syndrome, 605041 (3), Autosomal dominant; Cylindromatosis, familial, 132700 (3), Autosomal dominant; Trichoepithelioma, multiple familial, 1, 601606 (3), Autosomal dominant; ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132 (3), Autosomal dominant
DDB2	99.98 %	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
DICER1	99.85 %	606241	Pleuropulmonary blastoma, 601200 (3), Autosomal dominant; Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 (3), Autosomal dominant; GLOW syndrome, somatic mosaic, 618272 (3); Rhabdomyosarcoma, embryonal, 2, 180295 (3)
DIS3L2	99.75 %	614184	Perlman syndrome, 267000 (3), Autosomal recessive
DKC1	99.67 %	300126	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DNAJC21	99.71 %	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
DOCK8	99.75 %	611432	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700 (3), Autosomal recessive
EFL1	98.86 %	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
EGLN1	99.26 %	606425	Erythrocytosis, familial, 3, 609820 (3), Autosomal dominant; [Hemoglobin, high altitude adaptation], 609070 (3), Autosomal dominant
EGLN2	99.14 %	606424	No OMIM phenotypes
ELANE	99.92 %	130130	Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant
ELP1	99.78 %	603722	{Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant; Dysautonomia, familial, 223900 (3), Autosomal recessive
EPAS1	99.04 %	603349	Erythrocytosis, familial, 4, 611783 (3), Autosomal dominant
EPCAM	99.85 %	185535	Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive; Lynch syndrome 8, 613244 (3), Autosomal dominant
ERCC2	99.66 %	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
ERCC3	99.76 %	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC4	99.73 %	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive
ERCC5	99.95 %	133530	Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6L2	99.92 %	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
ETV6	99.79 %	600618	Thrombocytopenia 5, 616216 (3), Autosomal dominant; Leukemia, acute myeloid, somatic, 601626 (3)
EZH2	99.77 %	601573	Weaver syndrome, 277590 (3), Autosomal dominant
FAH	97.69 %	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
FANCA	99.94 %	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
FANCB	99.51 %	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
FANCC	99.91 %	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
FANCD2	99.24 %	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive

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FANCE	99.85 %	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive
FANCF	99.98 %	613897	Fanconi anemia, complementation group F, 603467 (3), Autosomal recessive
FANCG	99.83 %	602956	Fanconi anemia, complementation group G, 614082 (3), Autosomal recessive
FANCI	99.87 %	611360	Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive
FANCL	99.3 %	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive
FAS	99.94 %	134637	Squamous cell carcinoma, burn scar-related, somatic (3); Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant
FBXW7	99.84 %	606278	Developmental delay, hypotonia, and impaired language, 620012 (3), Autosomal dominant
FH	99.84 %	136850	Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive
G6PC3	99.69 %	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
GATA1	99.78 %	305371	Anemia, congenital, nonspherocytic hemolytic, 9, 301083 (3), X-linked recessive; Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595 (3); Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive; Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive
GATA2	99.91 %	137295	{Leukemia, acute myeloid, susceptibility to}, 601626 (3), Somatic mutation, Autosomal dominant; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
GPC3	99.69 %	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
GPC4	97.57 %	300168	Keipert syndrome, 301026 (3), X-linked recessive
GPR161	99.38 %	612250	{Medulloblastoma predisposition syndrome}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant
HAVCR2	99.87 %	606652	T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive
HAX1	99.76 %	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HRAS	99.97 %	190020	Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant
IKZF1	99.58 %	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
ITK	99.59 %	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
KDM3B	99.79 %	609373	Diets-Jongmans syndrome, 618846 (3), Autosomal dominant
KRAS	97.26 %	190070	Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3)

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Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
L2HGDH	99.76 %	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LIG4	99.99 %	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
LZTR1	99.3 %	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
MAP2K1	99.43 %	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAP2K2	99.93 %	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
MAX	99.8 %	154950	Polydactyly-macrocephaly syndrome, 620712 (3), Autosomal dominant; {Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
MDH2	98.1 %	154100	Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive
MDM4	99.45 %	602704	?Bone marrow failure syndrome 6, 618849 (3), Autosomal dominant
MEN1	99.91 %	613733	Lipoma, somatic (3); Angiofibroma, somatic (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant; Carcinoid tumor of lung (3); Adrenal adenoma, somatic (3); Parathyroid adenoma, somatic (3)
MLH1	99.69 %	120436	Lynch syndrome 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 1, 276300 (3), Autosomal recessive
MRAS	99.91 %	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
MSH2	99.77 %	609309	Lynch syndrome 1, 120435 (3), Autosomal dominant; Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 2, 619096 (3), Autosomal recessive
MSH6	99.86 %	600678	Lynch syndrome 5, 614350 (3), Autosomal dominant; Mismatch repair cancer syndrome 3, 619097 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Somatic mutation, Autosomal dominant
MYSM1	95.13 %	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
NAF1	99.83 %	617868	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365 (3), Autosomal dominant
NBN	99.79 %	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NF1	99.83 %	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
NF2	99.9 %	607379	Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, vestibular, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 101000 (3)
NHP2	100 %	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
NOP10	99.98 %	606471	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400 (3), Autosomal dominant; ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425 (3), Autosomal recessive; ?Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NRAS	99.69 %	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NSD1	99.93 %	606681	Sotos syndrome, 117550 (3), Autosomal dominant

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NYNRIN	99.96 %	620129	<i>No OMIM phenotypes</i>
PALB2	99.33 %	610355	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442 (3), Autosomal dominant; {Pancreatic cancer, susceptibility to, 3}, 613348 (3), Autosomal dominant; Fanconi anemia, complementation group N, 610832 (3), Autosomal recessive
PARN	99.09 %	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 (3), Autosomal dominant
PAX5	98.57 %	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3)
PHOX2B	99.93 %	603851	{Neuroblastoma, susceptibility to, 2}, 613013 (3); Neuroblastoma with Hirschsprung disease, 613013 (3); Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880 (3), Autosomal dominant
PIK3CA	99.63 %	171834	Hemifacial myohyperplasia, somatic, 606773 (3); CLOVE syndrome, somatic, 612918 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Cerebral cavernous malformations 4, somatic, 619538 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Macrodactyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Gastric cancer, somatic, 613659 (3); Non-small cell lung cancer, somatic, 211980 (3); Nevus, epidermal, somatic mosaic, 162900 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3)
PMS2	92.98 %	600259	Lynch syndrome 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive
POLD1	99.66 %	174761	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; Immunodeficiency 120, 620836 (3), Autosomal recessive; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant
POLE	99.91 %	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive
POLH	99.77 %	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
POT1	99.83 %	606478	Tumor predisposition syndrome 3, 615848 (3), Autosomal dominant; ?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368 (3), Autosomal recessive; ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367 (3), Autosomal dominant
PPP1CB	99.51 %	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PRF1	99.89 %	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
PRKAR1A	96.1 %	188830	Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant
PTCH1	99.87 %	601309	Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTEN	99.87 %	601728	{Glioma susceptibility 2}, 613028 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant

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PTPN11	99.84 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
RAF1	99.91 %	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant
RB1	99.74 %	614041	Small cell cancer of the lung, somatic, 182280 (3); Bladder cancer, somatic, 109800 (3); Retinoblastoma, trilateral, 180200 (3), Somatic mutation, Autosomal dominant; Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Somatic mutation, Autosomal dominant
RECQL4	99.91 %	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
REST	100 %	600571	Deafness, autosomal dominant 27, 612431 (3), Autosomal dominant; {Wilms tumor 6, susceptibility to}, 616806 (3), Autosomal dominant; Fibromatosis, gingival, 5, 617626 (3), Autosomal dominant
RET	99.85 %	164761	{Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant
RIT1	99.11 %	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RMRP	98.59 %	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
RPL11	98.91 %	604175	Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant
RPL15	99.92 %	604174	Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant
RPL18	99.78 %	604179	?Diamond-Blackfan anemia 18, 618310 (3), Autosomal dominant
RPL26	99.94 %	603704	?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant
RPL27	99.38 %	607526	?Diamond-Blackfan anemia 16, 617408 (3), Autosomal dominant
RPL31	98.15 %	617415	No OMIM phenotypes
RPL35	99.54 %	618315	?Diamond-Blackfan anemia 19, 618312 (3), Autosomal dominant
RPL35A	99.98 %	180468	Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant
RPL36	99.99 %	617893	No OMIM phenotypes
RPL5	99.93 %	603634	Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant
RPS10	99.88 %	603632	Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant
RPS15A	98.96 %	603674	?Diamond-Blackfan anemia 20, 618313 (3), Autosomal dominant
RPS17	99.96 %	180472	Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant
RPS19	99.99 %	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
RPS24	99.95 %	602412	Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant
RPS26	100 %	603701	Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant
RPS27	98.5 %	603702	?Diamond-Blackfan anemia 17, 617409 (3), Autosomal dominant
RPS28	99.91 %	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant
RPS29	99.81 %	603633	Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant
RPS7	99.94 %	603658	Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant
RRAS	99.69 %	165090	No OMIM phenotypes

Pediatric Oncopredisposition

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RRAS2	99.71 %	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant
RTEL1	99.92 %	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 (3), Autosomal dominant
RUNX1	99.92 %	151385	Platelet disorder, familial, with associated myeloid malignancy, 601399 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant
SAMD9	99.94 %	610456	Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant
SAMD9L	99.81 %	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant; ?Spinocerebellar ataxia 49, 619806 (3), Autosomal dominant; Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 (3), Autosomal dominant
SBDS	99.32 %	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
SDHA	97.97 %	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Pheochromocytoma/paraganglioma syndrome 5, 614165 (3), Autosomal dominant
SDHAF2	99.87 %	613019	Pheochromocytoma/paraganglioma syndrome 2, 601650 (3), Autosomal dominant
SDHB	93.61 %	185470	Pheochromocytoma/paraganglioma syndrome 4, 115310 (3), Autosomal dominant; Mitochondrial complex II deficiency, nuclear type 4, 619224 (3), Autosomal recessive; Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Paraganglioma and gastric stromal sarcoma, 606864 (3)
SDHC	98.25 %	602413	Pheochromocytoma/paraganglioma syndrome 3, 605373 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases
SDHD	91.13 %	602690	Pheochromocytoma/paraganglioma syndrome 1, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive
SETBP1	99.98 %	611060	Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant
SH2B3	99.86 %	605093	Thrombocythemia, somatic, 187950 (3); Myelofibrosis, somatic, 254450 (3); Erythrocytosis, somatic, 133100 (3)
SH2D1A	99.61 %	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
SHOC2	99.83 %	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
SLX4	99.88 %	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
SMAD4	99.95 %	600993	Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant
SMARCA4	99.81 %	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; ?Otosclerosis 12, 620792 (3), Autosomal dominant
SMARCB1	99.8 %	601607	Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant

Pediatric Oncopredisposition

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SMARCE1	99.26 %	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
SOS1	99.57 %	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
SOS2	99.81 %	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SPRTN	99.91 %	616086	Ruijs-Aalfs syndrome, 616200 (3), Autosomal recessive
SRP54	99.84 %	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
SRP72	99.71 %	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant
STAT3	99.7 %	102582	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
STK11	99.95 %	602216	Melanoma, malignant, somatic, 155600 (3); Pancreatic cancer, somatic, 260350 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3)
SUFU	99.9 %	607035	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Basal cell nevus syndrome 2, 620343 (3); {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant
TERC	99.98 %	602322	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant
TERT	99.86 %	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant
TINF2	99.94 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TMEM127	99.76 %	613403	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
TP53	99.83 %	191170	{Basal cell carcinoma 7}, 614740 (3), Autosomal dominant; {Adrenocortical carcinoma, pediatric}, 202300 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Li-Fraumeni syndrome, 151623 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3); Nasopharyngeal carcinoma, somatic, 607107 (3); {Osteosarcoma}, 259500 (3), Somatic mutation; {Choroid plexus papilloma}, 260500 (3), Autosomal dominant; {Colorectal cancer}, 114500 (3), Somatic mutation, Autosomal dominant; {Glioma susceptibility 1}, 137800 (3), Somatic mutation, Autosomal dominant; Bone marrow failure syndrome 5, 618165 (3), Autosomal dominant
TRIM28	99.94 %	601742	Wilms tumor 7, 621332 (3), Autosomal dominant
TRIM37	97.98 %	605073	Mulibrey nanism, 253250 (3), Autosomal recessive
TRIP13	99.98 %	604507	Oocyte/zygote/embryo maturation arrest 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive
TSC1	99.91 %	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangiomyomatosis, 606690 (3)
TSC2	99.88 %	191092	Lymphangiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
TSR2	99.78 %	300945	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive

Pediatric Oncopredisposition

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
UBE2T	99.23 %	610538	Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive
USB1	99.94 %	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
VHL	99.98 %	608537	Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
WAS	96.84 %	300392	Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive
WRAP53	99.62 %	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
WT1	97.92 %	607102	Mesothelioma, somatic, 156240 (3); Meacham syndrome, 608978 (3), Autosomal dominant; Frasier syndrome, 136680 (3), Somatic mutation, Autosomal dominant; Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Denys-Drash syndrome, 194080 (3), Somatic mutation, Autosomal dominant; Wilms tumor, type 1, 194070 (3), Somatic mutation, Autosomal dominant
XPA	99.78 %	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPC	99.93 %	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
ZCCHC8	99.87 %	616381	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

- (1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
- (2) the disorder has been placed on the map by linkage; no mutation has been found
- (3) the molecular basis for the disorder is known; a mutation has been found in the gene
- (4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.