# Pediatric Oncopredisposition

Gene panel

## Gene panel information

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

## Genes

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **A2ML1** | 99.97 % | 610627 | {Otitis media, susceptibility to}, 166760 (3), Autosomal dominant |
| **ABCB11** | 99.86 % | 603201 | Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive |
| **ACD** | 100 % | 609377 | ?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant |
| **AIP** | 99.99 % | 605555 | Pituitary adenoma 1, multiple types, 102200 (3), Somatic mutation, Autosomal dominant; Pituitary adenoma predisposition, 102200 (3), Somatic mutation, Autosomal dominant |
| **ALK** | 99.93 % | 105590 | {Neuroblastoma, susceptibility to, 3}, 613014 (3) |
| **AMER1** | 100 % | 300647 | Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant |
| **APC** | 99.97 % | 611731 | Colorectal cancer, somatic, 114500 (3); Brain tumor-polyposis 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** | 100 % | 612990 | Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant |
| **ATM** | 99.83 % | 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** | 99.44 % | 300032 | Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive |
| **BAP1** | 99.99 % | 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** | 99.8 % | 604610 | Bloom syndrome, 210900 (3), Autosomal recessive |
| **BMPR1A** | 99.58 % | 601299 | Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant |
| **BRAF** | 99.78 % | 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); Nonsmall cell lung cancer, somatic, 211980 (3) |

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **BRCA1** | 98.33 % | 113705 | Fanconi anemia, complementation group S, 617883 (3), Autosomal recessive; {Breast-ovarian cancer, familial, 1}, 604370 (3), Multifactorial, Autosomal dominant; {Pancreatic cancer, susceptibility to, 4}, 614320 (3) |
| **BRCA2** | 99.99 % | 600185 | Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Autosomal recessive, Somatic mutation, 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** | 99.39 % | 605882 | Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant |
| **BUB1B** | 100 % | 602860 | Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive |
| **CASP10** | 99.85 % | 601762 | Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3) |
| **CBL** | 99.95 % | 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** | 99.99 % | 602840 | Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive |
| **CDC73** | 99.6 % | 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** | 99.98 % | 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); {Prostate cancer, susceptibility to}, 176807 (3), Somatic mutation, Autosomal dominant |
| **CDK4** | 100 % | 123829 | {Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant |
| **CDKN1B** | 100 % | 600778 | Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant |
| **CDKN1C** | 100 % | 600856 | IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant |
| **CDKN2A** | 100 % | 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** | 100 % | 116897 | Leukemia, acute myeloid, somatic, 601626 (3); ?Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant |
| **CEP57** | 99.92 % | 607951 | Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive |
| **CLPB** | 99.97 % | 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** | 99.97 % | 600140 | Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant |
| **CTC1** | 100 % | 613129 | Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive |

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **CTLA4** | 99.99 % | 123890 | Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Celiac disease, susceptibility to, 3}, 609755 (3); {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant |
| **CTR9** | 99.98 % | 609366 | *No OMIM phenotypes* |
| **CYLD** | 99.46 % | 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** | 100 % | 600811 | Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive |
| **DICER1** | 99.96 % | 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** | 99.9 % | 614184 | Perlman syndrome, 267000 (3), Autosomal recessive |
| **DKC1** | 99.59 % | 300126 | ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive |
| **DNAJC21** | 99.67 % | 617048 | Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive |
| **DOCK8** | 99.86 % | 611432 | Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive |
| **EFL1** | 99.83 % | 617538 | Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive |
| **EGLN1** | 99.86 % | 606425 | Erythrocytosis, familial, 3, 609820 (3), Autosomal dominant; [Hemoglobin, high altitude adaptation], 609070 (3), Autosomal dominant |
| **EGLN2** | 99.97 % | 606424 | *No OMIM phenotypes* |
| **ELANE** | 100 % | 130130 | Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant |
| **ELP1** | 99.96 % | 603722 | {Medulloblastoma}, 155255 (3), Autosomal recessive, Somatic mutation, Autosomal dominant; Dysautonomia, familial, 223900 (3), Autosomal recessive |
| **EPAS1** | 99.99 % | 603349 | Erythrocytosis, familial, 4, 611783 (3), Autosomal dominant |
| **EPCAM** | 99.89 % | 185535 | Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive; Lynch syndrome 8, 613244 (3), Autosomal dominant |
| **ERCC2** | 99.98 % | 126340 | Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive |
| **ERCC3** | 99.9 % | 133510 | Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive |
| **ERCC4** | 99.92 % | 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** | 99.99 % | 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** | 99.94 % | 615667 | Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive |
| **ETV6** | 99.99 % | 600618 | Thrombocytopenia 5, 616216 (3), Autosomal dominant; Leukemia, acute myeloid, somatic, 601626 (3) |
| **EZH2** | 99.89 % | 601573 | Weaver syndrome, 277590 (3), Autosomal dominant |
| **FAH** | 99.98 % | 613871 | Tyrosinemia, type I, 276700 (3), Autosomal recessive |

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|---|---|---|---|
| **FANCA** | 100 % | 607139 | Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive |
| **FANCB** | 99.24 % | 300515 | Fanconi anemia, complementation group B, 300514 (3), X-linked recessive |
| **FANCC** | 99.98 % | 613899 | Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive |
| **FANCD2** | 99.86 % | 613984 | Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive |
| **FANCE** | 99.99 % | 613976 | Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive |
| **FANCF** | 100 % | 613897 | Fanconi anemia, complementation group F, 603467 (3), Autosomal recessive |
| **FANCG** | 100 % | 602956 | Fanconi anemia, complementation group G, 614082 (3), Autosomal recessive |
| **FANCI** | 99.96 % | 611360 | Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive |
| **FANCL** | 99.67 % | 608111 | Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive |
| **FAS** | 99.99 % | 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** | 99.9 % | 606278 | Developmental delay, hypotonia, and impaired language, 620012 (3), Autosomal dominant |
| **FH** | 99.95 % | 136850 | Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive |
| **G6PC3** | 99.98 % | 611045 | Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive |
| **GATA1** | 99.97 % | 305371 | Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (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; Hemolytic anemia due to elevated adenosine deaminase, 301083 (3), X-linked recessive |
| **GATA2** | 99.99 % | 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** | 99.6 % | 300037 | Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive |
| **GPC4** | 99.89 % | 300168 | Keipert syndrome, 301026 (3), X-linked recessive |
| **GPR161** | 99.92 % | 612250 | {Medulloblastoma predisposition syndrome}, 155255 (3), Autosomal recessive, Somatic mutation, Autosomal dominant |
| **HAVCR2** | 99.93 % | 606652 | T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive |
| **HAX1** | 100 % | 605998 | Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive |
| **HRAS** | 100 % | 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** | 99.92 % | 603023 | Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant |
| **ITK** | 99.91 % | 186973 | Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive |
| **KDM3B** | 99.98 % | 609373 | Diets-Jongmans syndrome, 618846 (3), Autosomal dominant |

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **KRAS** | 99.13 % | 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) |
| **L2HGDH** | 99.92 % | 609584 | L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive |
| **LIG4** | 100 % | 601837 | LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation |
| **LZTR1** | 99.46 % | 600574 | Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant |
| **MAP2K1** | 99.98 % | 176872 | Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3) |
| **MAP2K2** | 99.99 % | 601263 | Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant |
| **MAX** | 99.96 % | 154950 | {Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant |
| **MDH2** | 99.54 % | 154100 | Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive |
| **MDM4** | 99.43 % | 602704 | ?Bone marrow failure syndrome 6, 618849 (3), Autosomal dominant |
| **MEN1** | 99.98 % | 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** | 99.64 % | 120436 | Lynch syndrome 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 1, 276300 (3), Autosomal recessive |
| **MRAS** | 99.97 % | 608435 | Noonan syndrome 11, 618499 (3), Autosomal dominant |
| **MSH2** | 99.23 % | 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** | 99.97 % | 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** | 94.16 % | 612176 | Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive |
| **NAF1** | 99.94 % | 617868 | Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365 (3), Autosomal dominant |
| **NBN** | 99.93 % | 602667 | Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive |
| **NF1** | 99.88 % | 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** | 100 % | 607379 | Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, vestibular, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 101000 (3) |
| **NHP2** | 99.96 % | 606470 | Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive |
| **NOP10** | 99.99 % | 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 |

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **NRAS** | 99.66 % | 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** | 99.98 % | 606681 | Sotos syndrome, 117550 (3), Autosomal dominant |
| **NYNRIN** | 100 % | 620129 | *No OMIM phenotypes* |
| **PALB2** | 99.71 % | 610355 | {Breast-ovarian cancer, familial, susceptibility to, 5}, 620442 (3), Autosomal dominant; {Pancreatic cancer, susceptibility to, 3}, 613348 (3); Fanconi anemia, complementation group N, 610832 (3) |
| **PARN** | 99.75 % | 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** | 99.82 % | 167414 | {Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3) |
| **PHOX2B** | 99.98 % | 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** | 99.74 % | 171834 | 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); Nevus, epidermal, somatic, 162900 (3); Gastric cancer, somatic, 613659 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3) |
| **PMS2** | 70.47 % | 600259 | Lynch syndrome 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive |
| **POLD1** | 99.96 % | 174761 | Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant |
| **POLE** | 99.99 % | 174762 | {Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive |
| **POLH** | 99.85 % | 603968 | Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive |
| **POT1** | 99.91 % | 606478 | {Glioma susceptibility 9}, 616568 (3), Autosomal dominant; ?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368 (3), Autosomal recessive; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant; ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367 (3), Autosomal dominant |
| **PPP1CB** | 99.89 % | 600590 | Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant |
| **PRF1** | 100 % | 170280 | Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3) |
| **PRKAR1A** | 100 % | 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** | 99.99 % | 601309 | Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant |

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **PTEN** | 99.89 % | 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 |
| **PTPN11** | 99.98 % | 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** | 99.97 % | 164760 | Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant |
| **RB1** | 99.84 % | 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** | 100 % | 603780 | Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive |
| **REST** | 99.99 % | 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** | 99.97 % | 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** | 99.78 % | 609591 | Noonan syndrome 8, 615355 (3), Autosomal dominant |
| **RMRP** | 100 % | 157660 | Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive |
| **RPL11** | 99.81 % | 604175 | Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant |
| **RPL15** | 31.77 % | 604174 | Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant |
| **RPL18** | 100 % | 604179 | ?Diamond-Blackfan anemia 18, 618310 (3), Autosomal dominant |
| **RPL26** | 30.55 % | 603704 | ?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant |
| **RPL27** | 99.83 % | 607526 | ?Diamond-Blackfan anemia 16, 617408 (3), Autosomal dominant |
| **RPL31** | 99.88 % | 617415 | *No OMIM phenotypes* |
| **RPL35** | 99.99 % | 618315 | ?Diamond-Blackfan anemia 19, 618312 (3), Autosomal dominant |
| **RPL35A** | 97.55 % | 180468 | Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant |
| **RPL36** | 100 % | 617893 | *No OMIM phenotypes* |
| **RPL5** | 28.81 % | 603634 | Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant |
| **RPS10** | 0 % | 603632 | Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant |
| **RPS15A** | 22.14 % | 603674 | ?Diamond-Blackfan anemia 20, 618313 (3), Autosomal dominant |
| **RPS17** | 100 % | 180472 | Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant |
| **RPS19** | 100 % | 603474 | Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant |
| **RPS24** | 91.48 % | 602412 | Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant |
| **RPS26** | 8.99 % | 603701 | Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant |
| **RPS27** | 27.45 % | 603702 | ?Diamond-Blackfan anemia 17, 617409 (3), Autosomal dominant |
| **RPS28** | 100 % | 603685 | Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant |

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **RPS29** | 99.96 % | 603633 | Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant |
| **RPS7** | 88.5 % | 603658 | Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant |
| **RRAS** | 99.98 % | 165090 | *No OMIM phenotypes* |
| **RRAS2** | 99.94 % | 600098 | Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant |
| **RTEL1** | 100 % | 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** | 100 % | 151385 | Platelet disorder, familial, with associated myeloid malignancy, 601399 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant |
| **SAMD9** | 99.93 % | 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** | 99.95 % | 611170 | Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant; Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 (3), Autosomal dominant; Spinocerebellar ataxia 49, 619806 (3), Autosomal dominant |
| **SBDS** | 99.93 % | 607444 | {Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive |
| **SDHA** | 99.98 % | 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; Paragangliomas 5, 614165 (3), Autosomal dominant |
| **SDHAF2** | 99.96 % | 613019 | Paragangliomas 2, 601650 (3), Autosomal dominant |
| **SDHB** | 97.32 % | 185470 | Paragangliomas 4, 115310 (3), Autosomal dominant; Mitochondrial complex II deficiency, nuclear type 4, 619224 (3), Autosomal recessive; Gastrointestinal stromal tumor, 606764 (3), Isolated cases, Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3) |
| **SDHC** | 99.67 % | 602413 | Paragangliomas 3, 605373 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Gastrointestinal stromal tumor, 606764 (3), Isolated cases, Autosomal dominant |
| **SDHD** | 82.93 % | 602690 | Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive; Pheochromocytoma, 171300 (3), Autosomal dominant |
| **SETBP1** | 100 % | 611060 | Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant |
| **SH2B3** | 99.95 % | 605093 | Thrombocythemia, somatic, 187950 (3); Myelofibrosis, somatic, 254450 (3); Erythrocytosis, somatic, 133100 (3) |
| **SH2D1A** | 98.98 % | 300490 | Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive |
| **SHOC2** | 99.96 % | 602775 | Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant |
| **SLX4** | 100 % | 613278 | Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive |
| **SMAD4** | 99.97 % | 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** | 99.99 % | 603254 | Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant |

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **SMARCB1** | 99.99 % | 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 |
| **SMARCE1** | 99.87 % | 603111 | {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant |
| **SOS1** | 99.68 % | 182530 | Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant |
| **SOS2** | 99.39 % | 601247 | Noonan syndrome 9, 616559 (3), Autosomal dominant |
| **SPRTN** | 99.98 % | 616086 | Ruijs-Aalfs syndrome, 616200 (3), Autosomal recessive |
| **SRP54** | 99.83 % | 604857 | Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant |
| **SRP72** | 99.91 % | 602122 | Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant |
| **STAT3** | 99.97 % | 102582 | Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant |
| **STK11** | 100 % | 602216 | Melanoma, malignant, somatic, 155600 (3); Pancreatic cancer, somatic, 260350 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3) |
| **SUFU** | 100 % | 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), Autosomal recessive, Somatic mutation, Autosomal dominant |
| **TERC** | 98.59 % | 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** | 100 % | 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** | 100 % | 604319 | Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant |
| **TMEM127** | 99.99 % | 613403 | {Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant |
| **TP53** | 99.98 % | 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** | 100 % | 601742 | *No OMIM phenotypes* |
| **TRIM37** | 98.19 % | 605073 | Mulibrey nanism, 253250 (3), Autosomal recessive |
| **TRIP13** | 100 % | 604507 | Oocyte/zygote/embryo maturation arrest 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive |
| **TSC1** | 99.99 % | 605284 | Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3) |
| **TSC2** | 99.98 % | 191092 | Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant |

| Gene | % coding region covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **TSR2** | 99.96 % | 300945 | ?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive |
| **UBE2T** | 99.89 % | 610538 | Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive |
| **USB1** | 89.62 % | 613276 | Poikiloderma with neutropenia, 604173 (3), Autosomal recessive |
| **VHL** | 100 % | 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** | 99.9 % | 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** | 100 % | 612661 | Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive |
| **WT1** | 99.99 % | 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** | 99.68 % | 611153 | Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive |
| **XPC** | 99.98 % | 613208 | Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive |
| **ZCCHC8** | 99.95 % | 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: **2023-07-31**

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.

* Exome panels: >=20x, HyperCap panels: >=30x