

<b>Pediatrie oncopredispositie panel</b>		
<b>versie</b>	v3 (178 genen)	Centrum voor Medische Genetica Gent
<b>Gene</b>	<b>OMIM gene ID</b>	<b>Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern</b>
<i>A2ML1</i>	610627	{Otitis media, susceptibility to}, 166760 (3), Autosomal dominant
<i>ABCB11</i>	603201	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive
<i>ACD</i>	609377	?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal dominant, Autosomal recessive; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal dominant, Autosomal recessive
<i>AIP</i>	605555	Pituitary adenoma 1, multiple types, 102200 (3), Autosomal dominant, Somatic mutation; Pituitary adenoma predisposition, 102200 (3), Autosomal dominant, Somatic mutation
<i>ALK</i>	105590	{Neuroblastoma, susceptibility to, 3}, 613014 (3)
<i>APC</i>	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
<i>ASXL1</i>	612990	Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant
<i>ATM</i>	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
<i>BAP1</i>	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
<i>BLM</i>	604610	Bloom syndrome, 210900 (3), Autosomal recessive
<i>BMPR1A</i>	601299	Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant
<i>BRAF</i>	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)

<i>BRCA1</i>	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)
<i>BRCA2</i>	600185	Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Autosomal dominant, Somatic mutation, Autosomal recessive; {Prostate cancer}, 176807 (3), Autosomal dominant, Somatic mutation; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant; {Breast cancer, male, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; {Pancreatic cancer 2}, 613347 (3); Wilms tumor, 194070 (3), Autosomal dominant, Somatic mutation
<i>BRIP1</i>	605882	Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
<i>BUB1B</i>	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
<i>CBL</i>	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation
<i>CD27</i>	186711	Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive
<i>CD70</i>	602840	Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive
<i>CDC73</i>	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
<i>CDH1</i>	192090	Ovarian cancer, somatic, 167000 (3); Blepharochelodontic 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), Autosomal dominant, Somatic mutation
<i>CDK4</i>	123829	{Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant
<i>CDKN1B</i>	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant

<i>CDKN1C</i>	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
<i>CDKN2A</i>	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
<i>CEBPA</i>	116897	Leukemia, acute myeloid, somatic, 601626 (3); ?Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation
<i>CEP57</i>	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
<i>CREBBP</i>	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
<i>CTC1</i>	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<i>CTLA4</i>	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
<i>CTR9</i>	609366	No OMIM phenotype
<i>DDB2</i>	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
<i>DICER1</i>	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)
<i>DIS3L2</i>	614184	Perlman syndrome, 267000 (3), Autosomal recessive
<i>DKC1</i>	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
<i>DNAJC21</i>	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
<i>EFL1</i>	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
<i>EGLN1</i>	606425	Erythrocytosis, familial, 3, 609820 (3), Autosomal dominant; [Hemoglobin, high altitude adaptation], 609070 (3), Autosomal dominant
<i>EGLN2</i>	606424	No OMIM phenotype
<i>ELP1</i>	603722	Dysautonomia, familial, 223900 (3), Autosomal recessive; Medulloblastoma, 155255 (3), Autosomal dominant, Somatic mutation, Autosomal recessive
<i>EPAS1</i>	603349	Erythrocytosis, familial, 4, 611783 (3), Autosomal dominant
<i>EPCAM</i>	185535	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 (3), Autosomal dominant; Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive
<i>ERCC2</i>	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal

		recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
<i>ERCC3</i>	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
<i>ERCC4</i>	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
<i>ERCC5</i>	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
<i>ERCC6L2</i>	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
<i>ETV6</i>	600618	Thrombocytopenia 5, 616216 (3), Autosomal dominant; Leukemia, acute myeloid, somatic, 601626 (3)
<i>EZH2</i>	601573	Weaver syndrome, 277590 (3), Autosomal dominant
<i>FAH</i>	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
<i>FANCA</i>	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
<i>FANCB</i>	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
<i>FANCC</i>	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
<i>FANCD2</i>	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
<i>FANCE</i>	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive
<i>FANCF</i>	613897	Fanconi anemia, complementation group F, 603467 (3), Autosomal recessive
<i>FANCG</i>	602956	Fanconi anemia, complementation group G, 614082 (3), Autosomal recessive
<i>FANCI</i>	611360	Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive
<i>FANCL</i>	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive
<i>FAS</i>	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
<i>FBXW7</i>	606278	No OMIM phenotype
<i>FH</i>	136850	Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive

<i>GATA1</i>	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
<i>GATA2</i>	137295	{Leukemia, acute myeloid, susceptibility to}, 601626 (3), Autosomal dominant, Somatic mutation; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
<i>GPC3</i>	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
<i>GPC4</i>	300168	Keipert syndrome, 301026 (3), X-linked recessive
<i>GPR161</i>	612250	No OMIM phenotype
<i>HAVCR2</i>	606652	T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive
<i>HRAS</i>	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
<i>IKZF1</i>	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
<i>ITK</i>	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
<i>KRAS</i>	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)
<i>L2HGDH</i>	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<i>LIG4</i>	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
<i>LZTR1</i>	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
<i>MAP2K1</i>	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)

<i>MAP2K2</i>	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
<i>MAX</i>	154950	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
<i>MDH2</i>	154100	Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive
<i>MDM4</i>	602704	?Bone marrow failure syndrome 6, 618849 (3), Autosomal dominant
<i>MEN1</i>	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)
<i>MLH1</i>	120436	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 1, 276300 (3), Autosomal recessive
<i>MRAS</i>	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
<i>MSH2</i>	609309	Muir-Torre syndrome, 158320 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3), Autosomal dominant; Mismatch repair cancer syndrome 2, 619096 (3), Autosomal recessive
<i>MSH6</i>	600678	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant; Mismatch repair cancer syndrome 3, 619097 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Autosomal dominant, Somatic mutation
<i>MYSM1</i>	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
<i>NBN</i>	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
<i>NF1</i>	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
<i>NF2</i>	607379	Neurofibromatosis, type 2, 101000 (3), Autosomal dominant; Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, somatic, 162091 (3)
<i>NHP2</i>	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
<i>NOP10</i>	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
<i>NRAS</i>	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)

<i>NSD1</i>	606681	Sotos syndrome, 117550 (3), Autosomal dominant
<i>PALB2</i>	610355	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; {Pancreatic cancer, susceptibility to, 3}, 613348 (3); Fanconi anemia, complementation group N, 610832 (3)
<i>PARN</i>	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant
<i>PAX5</i>	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3)
<i>PHOX2B</i>	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
<i>PIK3CA</i>	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); Macroductyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Nevus, epidermal, somatic, 162900 (3); Gastric cancer, somatic, 613659 (3); Non-small cell lung cancer, somatic, 211980 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3)
<i>PMS2</i>	600259	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive
<i>POLD1</i>	174761	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant
<i>POLE</i>	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive
<i>POLH</i>	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
<i>POT1</i>	606478	{Glioma susceptibility 9}, 616568 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant
<i>PPP1CB</i>	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
<i>PRF1</i>	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)

<i>PRKAR1A</i>	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
<i>PTCH1</i>	601309	Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant; Basal cell nevus syndrome, 109400 (3), Autosomal dominant
<i>PTEN</i>	601728	{Glioma susceptibility 2}, 613028 (3); {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
<i>PTPN11</i>	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)
<i>RAF1</i>	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3)
<i>RB1</i>	614041	Small cell cancer of the lung, somatic, 182280 (3); Bladder cancer, somatic, 109800 (3); Retinoblastoma, trilateral, 180200 (3), Autosomal dominant, Somatic mutation; Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Autosomal dominant, Somatic mutation
<i>RECQL4</i>	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
<i>REST</i>	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
<i>RET</i>	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
<i>RIT1</i>	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
<i>RMRP</i>	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
<i>RPL11</i>	604175	Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant
<i>RPL15</i>	604174	?Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant
<i>RPL18</i>	604179	?Diamond-Blackfan anemia 18, 618310 (3), Autosomal dominant



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<i>RPL26</i>	603704	?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant
<i>RPL27</i>	607526	?Diamond-Blackfan anemia 16, 617408 (3), Autosomal dominant
<i>RPL35</i>	618315	?Diamond-Blackfan anemia 19, 618312 (3), Autosomal dominant
<i>RPL35A</i>	180468	Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant
<i>RPL5</i>	603634	Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant
<i>RPS10</i>	603632	Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant
<i>RPS15A</i>	603674	?Diamond-Blackfan anemia 20, 618313 (3), Autosomal dominant
<i>RPS17</i>	180472	Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant
<i>RPS19</i>	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
<i>RPS24</i>	602412	Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant
<i>RPS26</i>	603701	Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant
<i>RPS27</i>	603702	?Diamond-Blackfan anemia 17, 617409 (3), Autosomal dominant
<i>RPS28</i>	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant
<i>RPS29</i>	603633	Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant
<i>RPS7</i>	603658	Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant
<i>RRAS</i>	165090	No OMIM phenotype
<i>RRAS2</i>	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant
<i>RTEL1</i>	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal dominant, Autosomal recessive
<i>RUNX1</i>	151385	Platelet disorder, familial, with associated myeloid malignancy, 601399 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation
<i>SAMD9</i>	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
<i>SAMD9L</i>	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
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
<i>SDHA</i>	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
<i>SDHAF2</i>	613019	Paragangliomas 2, 601650 (3), Autosomal dominant

<i>SDHB</i>	185470	Paragangliomas 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; Pheochromocytoma, 171300 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3)
<i>SDHC</i>	602413	Paragangliomas 3, 605373 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases
<i>SDHD</i>	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
<i>SETBP1</i>	611060	Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant
<i>SH2D1A</i>	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
<i>SHOC2</i>	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
<i>SLX4</i>	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
<i>SMARCA4</i>	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant
<i>SMARCB1</i>	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
<i>SMARCE1</i>	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
<i>SOS1</i>	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
<i>SOS2</i>	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
<i>SRP54</i>	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
<i>SRP72</i>	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant
<i>STK11</i>	602216	Melanoma, malignant, somatic, 155600 (3); Pancreatic cancer, somatic, 260350 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3)
<i>SUFU</i>	607035	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Medulloblastoma, desmoplastic, 155255 (3), Autosomal dominant, Somatic mutation, Autosomal recessive; Basal cell nevus syndrome, 109400 (3), Autosomal dominant

<i>TERC</i>	602322	{Pulmonary fibrosis, idiopathic, susceptibility to}, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant; {Aplastic anemia}, 614743 (3), Autosomal dominant
<i>TERT</i>	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal dominant, Autosomal recessive; {Melanoma, cutaneous malignant, 9}, 615134 (3); Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Autosomal dominant, Somatic mutation
<i>TINF2</i>	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
<i>TMEM127</i>	613403	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
<i>TP53</i>	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), Autosomal dominant, Somatic mutation; {Glioma susceptibility 1}, 137800 (3), Autosomal dominant, Somatic mutation; Bone marrow failure syndrome 5, 618165 (3), Autosomal dominant
<i>TRIM28</i>	601742	No OMIM phenotype
<i>TRIM37</i>	605073	Mulibrey nanism, 253250 (3), Autosomal recessive
<i>TRIP13</i>	604507	Oocyte maturation defect 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive
<i>TSC1</i>	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioliomyomatosis, 606690 (3)
<i>TSC2</i>	191092	Lymphangioliomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
<i>TSR2</i>	300945	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive
<i>UBE2T</i>	610538	Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive
<i>USB1</i>	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive

<i>VHL</i>	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
<i>WAS</i>	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
<i>WRAP53</i>	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
<i>WT1</i>	607102	Mesothelioma, somatic, 156240 (3); Meacham syndrome, 608978 (3), Autosomal dominant; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation
<i>XPA</i>	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
<i>XPC</i>	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.

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

OMIM release used for OMIM disease identifiers and descriptions: August 24, 2022

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.