pediatrische oncopredispositie panel

versie v1 (195 genen) Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
A2ML1	610627	{Otitis media, susceptibility to}, 166760 (3), Autosomal dominant
ACD	609377	?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant
AKT1	164730	Breast cancer, somatic, 114480 (3); Cowden syndrome 6, 615109 (3); Proteus syndrome, somatic, 176920 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3)
ALK	105590	{Neuroblastoma, susceptibility to, 3}, 613014 (3)
ANKRD26	610855	Thrombocytopenia 2, 188000 (3), Autosomal dominant
APC	611731	Adenoma, periampullary, somatic (3); Desmoid disease, hereditary, 135290 (3), Autosomal dominant; Adenomatous polyposis coli, 175100 (3), Autosomal dominant; Gardner syndrome, 175100 (3), Autosomal dominant; Hepatoblastoma, somatic, 114550 (3); Colorectal cancer, somatic, 114500 (3); Brain tumor-polyposis syndrome 2, 175100 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3)
ASXL1	612990	Bohring-Opitz syndrome, 605039 (3), Autosomal dominant; Myelodysplastic syndrome, somatic, 614286 (3)
ATM	607585	Lymphoma, mantle cell, somatic (3); 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)
ATP7B	606882	Wilson disease, 277900 (3), Autosomal recessive
BAP1	603089	Tumor predisposition syndrome, 614327 (3), Autosomal dominant
BLM	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BMPR1A	601299	Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis syndrome, infantile form, 174900 (3), Autosomal dominant
BRAF	164757	Nonsmall cell lung cancer, somatic (3); Melanoma, malignant, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Colorectal cancer, somatic (3); Adenocarcinoma of lung, somatic, 211980 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant

BRCA1	113705	Fanconi anemia, complementation group S, 617883 (3), Autosomal recessive; {Pancreatic cancer, susceptibility to, 4}, 614320 (3); {Breast-ovarian cancer, familial, 1}, 604370 (3), Autosomal dominant, Multifactorial
BRCA2	600185	{Pancreatic cancer 2}, 613347 (3); {Breast cancer, male, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; {Glioblastoma 3}, 613029 (3), Autosomal recessive; Wilms tumor, 194070 (3), Autosomal dominant, Somatic mutation; Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Autosomal recessive, Autosomal dominant, Somatic mutation; {Prostate cancer}, 176807 (3), Autosomal dominant, Somatic mutation; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant
BRIP1	605882	Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
BUB1B	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
CBL	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation
CDC73	607393	Parathyroid adenoma with cystic changes, 145001 (3), Autosomal dominant; Hyperparathyroidism-jaw tumor syndrome, 145001 (3), Autosomal dominant; Parathyroid carcinoma, 608266 (3); Hyperparathyroidism, familial primary, 145000 (3), Autosomal dominant
CDK4	123829	{Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant
CDKN1B	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant
CDKN1C	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CDKN2A	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	116897	Leukemia, acute myeloid, somatic, 601626 (3); ?Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation
CREBBP	600140	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant; Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTR9	609366	No OMIM phenotype

CYLD	605018	Cylindromatosis, familial, 132700 (3), Autosomal dominant; Brooke-Spiegler syndrome, 605041 (3), Autosomal dominant; Trichoepithelioma, multiple familial, 1, 601606 (3), Autosomal dominant
DDB2	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
DICER1	606241	GLOW syndrome, somatic mosaic, 618272 (3); Rhabdomyosarcoma, embryonal, 2, 180295 (3); Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 (3), Autosomal dominant; Pleuropulmonary blastoma, 601200 (3), Autosomal dominant
DIS3L2	614184	Perlman syndrome, 267000 (3), Autosomal recessive
DKC1	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DNAJC21	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
EFL1	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
EP300	602700	Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant; Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3)
EPAS1	603349	Erythrocytosis, familial, 4, 611783 (3)
EPCAM	185535	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 (3); Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive
EPO	133170	Erythrocytosis, familial, 5, 617907 (3), Autosomal dominant; {Microvascular complications of diabetes 2}, 612623 (3); ?Diamond-Blackfan anemia-like, 617911 (3), Autosomal recessive
ERCC2	126340	Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
ERCC3	133510	Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive; Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive
ERCC4	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive
ERCC5	133530	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive; Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive
ERCC6L2	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
ETV6	600618	Leukemia, acute myeloid, somatic, 601626 (3); Thrombocytopenia 5, 616216 (3), Autosomal dominant

EXT1	608177	Exostoses, multiple, type 1, 133700 (3), Autosomal dominant; Chondrosarcoma, 215300 (3), Somatic mutation
EXT2	608210	Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant
EZH2	601573	Weaver syndrome, 277590 (3), Autosomal dominant
FAH	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
FANCA	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
FANCB	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
FANCC	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
FANCD2	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
FANCE	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive
FANCF	613897	Fanconi anemia, complementation group F, 603467 (3)
FANCG	602956	Fanconi anemia, complementation group G, 614082 (3)
FANCI	611360	Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive
FANCL	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive
FANCM	609644	Spermatogenic failure 28, 618086 (3), Autosomal recessive; ?Premature ovarian failure 15, 618096 (3), Autosomal recessive
G6PC	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
GALNT14	608225	No OMIM phenotype
GATA1	305371	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3); Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive; Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive
GATA2	137295	Emberger syndrome, 614038 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3); Immunodeficiency 21, 614172 (3), Autosomal dominant; {Leukemia, acute myeloid, susceptibility to}, 601626 (3), Autosomal dominant, Somatic mutation
GBE1	607839	Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive; Glycogen storage disease IV, 232500 (3), Autosomal recessive
GFI1B	604383	Bleeding disorder, platelet-type, 17, 187900 (3), Autosomal recessive, Autosomal dominant

GNAS	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
GPC4	300168	Keipert syndrome, 301026 (3), X-linked recessive
HOXA11	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 (3), Autosomal dominant
HRAS	190020	Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant
JAK2	147796	Myelofibrosis, somatic, 254450 (3); Thrombocythemia 3, 614521 (3), Autosomal dominant, Somatic mutation; Polycythemia vera, somatic, 263300 (3); {Budd-Chiari syndrome, somatic}, 600880 (3); Leukemia, acute myeloid, somatic, 601626 (3); Erythrocytosis, somatic, 133100 (3)
KIT	164920	Gastrointestinal stromal tumor, familial, 606764 (3), Autosomal dominant, Isolated cases; Mastocytosis, cutaneous, 154800 (3), Autosomal dominant; Germ cell tumors, somatic, 273300 (3); Leukemia, acute myeloid, somatic, 601626 (3); Mastocytosis, systemic, somatic, 154800 (3); Piebaldism, 172800 (3), Autosomal dominant
KRAS	190070	Oculoectodermal syndrome, somatic, 600268 (3); Leukemia, acute myeloid, somatic, 601626 (3); Breast cancer, somatic, 114480 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Bladder cancer, somatic, 109800 (3); Pancreatic carcinoma, somatic, 260350 (3); Lung cancer, somatic, 211980 (3); Gastric cancer, somatic, 137215 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant
L2HGDH	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LRMDA	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive

LZTR1	600574	{Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant; Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant
MAD2L2	604094	?Fanconi anemia, complementation group V, 617243 (3), Autosomal recessive
MAP2K1	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAPK1	176948	No OMIM phenotype
MAX	154950	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
MDM4	602704	?Bone marrow failure syndrome 6, 618849 (3), Autosomal dominant
MECOM	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant
MEN1	613733	Angiofibroma, somatic (3); Adrenal adenoma, somatic (3); Parathyroid adenoma, somatic (3); Lipoma, somatic (3); Carcinoid tumor of lung (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant
MLH1	120436	Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Colorectal cancer, hereditary nonpolyposis, type 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant
MPL	159530	Myelofibrosis with myeloid metaplasia, somatic, 254450 (3); Thrombocytopenia, congenital amegakaryocytic, 604498 (3), Autosomal recessive; Thrombocythemia 2, 601977 (3), Autosomal dominant, Somatic mutation
MRAS	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
MSH2	609309	Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Muir-Torre syndrome, 158320 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3), Autosomal dominant
MSH6	600678	Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Autosomal dominant, Somatic mutation; Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant
MTAP	156540	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250 (3), Autosomal dominant
MUTYH	604933	Gastric cancer, somatic, 613659 (3); Adenomas, multiple colorectal, 608456 (3), Autosomal recessive
MYSM1	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
NBN	602667	Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive; Aplastic anemia, 609135 (3)

NF1	613113	Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Watson syndrome, 193520 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant
NF2	607379	Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, somatic, 162091 (3); Neurofibromatosis, type 2, 101000 (3), Autosomal dominant
NHP2	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
NKX2-1	600635	{Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
NOP10	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NRAS	164790	Epidermal nevus, somatic, 162900 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Colorectal cancer, somatic, 114500 (3); ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Noonan syndrome 6, 613224 (3), Autosomal dominant
NSD1	606681	Sotos syndrome 1, 117550 (3), Autosomal dominant
OCA2	611409	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive; Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive
PALB2	610355	{Pancreatic cancer, susceptibility to, 3}, 613348 (3); Fanconi anemia, complementation group N, 610832 (3); {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
PARN	604212	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant; Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive
PAX5	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3)
PDE11A	604961	Pigmented nodular adrenocortical disease, primary, 2, 610475 (3), Autosomal dominant
PDE8B	603390	Striatal degeneration, autosomal dominant, 609161 (3), Autosomal dominant; Pigmented nodular adrenocortical disease, primary, 3, 614190 (3)

PDGFRA	173490	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 (3); Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3), Isolated cases, Somatic mutation
PDGFRB	173410	Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant
PHOX2B	603851	Neuroblastoma with Hirschsprung disease, 613013 (3); Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 (3), Autosomal dominant; {Neuroblastoma, susceptibility to, 2}, 613013 (3)
PIK3CA	171834	Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); CLAPO syndrome, somatic, 613089 (3); Cowden syndrome 5, 615108 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Macrodactyly, somatic, 155500 (3); Keratosis, seborrheic, somatic, 182000 (3); Gastric cancer, somatic, 613659 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Nevus, epidermal, somatic, 162900 (3); CLOVE syndrome, somatic, 612918 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
PMS2	600259	Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3)
POLH	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
POT1	606478	{Glioma susceptibility 9}, 616568 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant
PPP1CB	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
PRKAR1A	188830	Adrenocortical tumor, somatic (3); Myxoma, intracardiac, 255960 (3), Autosomal dominant; Carney complex, type 1, 160980 (3), Autosomal dominant; Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant
PTCH1	601309	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTCH2	603673	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3)

PTEN	601728	Prostate cancer, somatic, 176807 (3); {Glioma susceptibility 2}, 613028 (3); Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant
PTPN11	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
RAD51	179617	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; Mirror movements 2, 614508 (3), Autosomal dominant; Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant
RAD51C	602774	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive
RAF1	164760	LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3), Autosomal dominant; Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant
RASA2	601589	No OMIM phenotype
RB1	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
RECQL4	603780	RAPADILINO syndrome, 266280 (3), Autosomal recessive; Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2,, 268400 (3), Autosomal recessive
REST	600571	{Wilms tumor 6, susceptibility to}, 616806 (3); Fibromatosis, gingival, 5, 617626 (3), Autosomal dominant; ?Deafness, autosomal dominant 27, 612431 (3), Autosomal dominant
RET	164761	Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant
RFWD3	614151	?Fanconi anemia, complementation group W, 617784 (3), Autosomal recessive
RIT1	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RMRP	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive
RPL11	604175	Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant

RPL15	604174	?Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant
RPL18	604179	?Diamond-Blackfan anemia 18, 618310 (3), Autosomal dominant
RPL26	603704	?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant
RPL27	607526	?Diamond-Blackfan anemia 16, 617408 (3), Autosomal dominant
RPL35	618315	?Diamond-Blackfan anemia 19, 618312 (3), Autosomal dominant
RPL35A	180468	Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant
RPL5	603634	Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant
RPS10	603632	Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant
RPS15A	603674	?Diamond-Blackfan anemia 20, 618313 (3), Autosomal dominant
RPS17	180472	Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant
RPS19	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
RPS24	602412	Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant
RPS26	603701	Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant
RPS27	603702	?Diamond-Blackfan anemia 17, 617409 (3), Autosomal dominant
RPS28	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant
RPS29	603633	Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant
RPS7	603658	Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant
RRAS	165090	No OMIM phenotype
RRAS2	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant
RTEL1	608833	Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant
RUNX1	151385	Platelet disorder, familial, with associated myeloid malignancy, 601399 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation
SAMD9	610456	MIRAGE syndrome, 617053 (3), Autosomal dominant; Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive
SAMD9L	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant
SBDS	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Paragangliomas 5, 614165 (3), Autosomal dominant; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive
SDHAF2	613019	Paragangliomas 2, 601650 (3), Autosomal dominant
SDHB	185470	Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Pheochromocytoma, 171300 (3), Autosomal dominant; Paragangliomas 4, 115310 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3)

SDHC	602413	Paragangliomas 3, 605373 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases
SDHD	602690	Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
SEC23B	610512	?Cowden syndrome 7, 616858 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type II, 224100 (3), Autosomal recessive
SETBP1	611060	Mental retardation, autosomal dominant 29, 616078 (3), Autosomal dominant; Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant
SH2D1A	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
SHOC2	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
SLC24A5	609802	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive
SLX4	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
SMAD4	600993	Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant; Myhre syndrome, 139210 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3)
SMARCA4	603254	{Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant
SMARCB1	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	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
SOS1	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
SOS2	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SRP72	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant
STK11	602216	Testicular tumor, somatic, 273300 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Melanoma, malignant, somatic (3); Pancreatic cancer, somatic, 260350 (3)

SUFU	607035	Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Medulloblastoma, desmoplastic, 155255 (3), Autosomal recessive, Autosomal dominant, Somatic mutation; {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive
TERC	602322	{Aplastic anemia}, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant; {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743 (3), Autosomal dominant
TERT	187270	{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; {Dyskeratosis congenita, autosomal recessive 4}, 613989 (3), Autosomal recessive, Autosomal dominant; {Dyskeratosis congenita, autosomal dominant 2}, 613989 (3), Autosomal recessive, Autosomal dominant
THPO	600044	Thrombocythemia 1, 187950 (3), Autosomal dominant
TINF2	604319	Revesz syndrome, 268130 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant
TMEM127	613403	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
TP53	191170	{Adrenocortical carcinoma, pediatric}, 202300 (3), Autosomal dominant; {Glioma susceptibility 1}, 137800 (3), Autosomal dominant, Somatic mutation; {Basal cell carcinoma 7}, 614740 (3), Autosomal dominant; Bone marrow failure syndrome 5, 618165 (3), Autosomal dominant; {Colorectal cancer}, 114500 (3), Autosomal dominant, Somatic mutation; Nasopharyngeal carcinoma, somatic, 607107 (3); Breast cancer, somatic, 114480 (3); {Osteosarcoma}, 259500 (3), Somatic mutation; {Choroid plexus papilloma}, 260500 (3), Autosomal dominant; Li-Fraumeni syndrome, 151623 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); Pancreatic cancer, somatic, 260350 (3)
TRIM37	605073	Mulibrey nanism, 253250 (3), Autosomal recessive
TSC1	605284	Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, 606690 (3)
TSC2	191092	Tuberous sclerosis-2, 613254 (3), Autosomal dominant; ?Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, somatic, 606690 (3)
TSR2	300945	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive

TYR	606933	Waardenburg syndrome/albinism, digenic, 103470 (3); Albinism, oculocutaneous, type IB, 606952 (3), Autosomal recessive; [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 (3), Autosomal dominant; Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive; [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3), Autosomal dominant
TYRP1	115501	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive
UBE2T	610538	Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive
VHL	608537	Pheochromocytoma, 171300 (3), Autosomal dominant; Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Hemangioblastoma, cerebellar, somatic (3)
WAS	300392	Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive; Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive
WRAP53	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
WRN	604611	Werner syndrome, 277700 (3), Autosomal recessive
WT1	607102	Mesothelioma, somatic, 156240 (3); Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Meacham syndrome, 608978 (3); Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant
XPA	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPC	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
XRCC2	600375	?Fanconi anemia, complementation group U, 617247 (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: Sep 01, 2020

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.