Gene	OMIM phenotype	OMIM Number
ACD	?Dyskeratosis congenita, autosomal dominant 6,	616553
	?Dyskeratosis congenita, autosomal recessive 7	
ADA2	Sneddon syndrome	182410
	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome	615688
AK2	Reticular dysgenesis	267500
ALAS2	Anemia, sideroblastic, 1	300751
	Protoporphyria, erythropoietic, X-linked	300752
ANKRD26	Thrombocytopenia 2	188000
APC	Adenoma, periampullary, somatic	175100
	Adenomatous polyposis coli	175100
	Brain tumor-polyposis syndrome 2	175100
	Colorectal cancer, somatic	114500
	Desmoid disease, hereditary	135290
	Gardner syndrome	175100
	Gastric adenocarcinoma and proximal polyposis of the stomach	619182
	Gastric cancer, somatic	613659
	Hepatoblastoma, somatic	11455
ATG2B	NA	NA
ATM	Ataxia-telangiectasia	208900
	Lymphoma, B-cell non-Hodgkin, somatic	NA
	Lymphoma, mantle cell, somatic	NA
	T-cell prolymphocytic leukemia, somatic	NA
	{Breast cancer, susceptibility to}	114480
ATR	?Cutaneous telangiectasia and cancer syndrome, familial	614564
	Seckel syndrome 1	210600
BLM	Bloom syndrome	210900
BRAF	Adenocarcinoma of lung, somatic	211980
	Cardiofaciocutaneous syndrome	115150
	Colorectal cancer, somatic	114500
	LEOPARD syndrome 3	613707
	Melanoma, malignant, somatic	155600
	Nonsmall cell lung cancer, somatic	211980
	Noonan syndrome 7	613706
BRCA1	Fanconi anemia, complementation group S	617883
	{Breast-ovarian cancer, familial, 1}	604370
	{Pancreatic cancer, susceptibility to, 4}	614320
BRCA2	Fanconi anemia, complementation group D1	605724
	Wilms tumor	194070
	{Breast cancer, male, susceptibility to}	114480
	{Breast-ovarian cancer, familial, 2}	612555
	{Glioblastoma 3}	613029
	{Medulloblastoma}	155255
	{Pancreatic cancer 2}	613347
	{Prostate cancer}	176807
RIP1 (FANCJ	Fanconi anemia, complementation group J	609054
	{Breast cancer, early-onset, susceptibility to}	114480
CBL	?Juvenile myelomonocytic leukemia	607785
	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	613563
CDKN2A	{Melanoma and neural system tumor syndrome}	155755
	{Melanoma, cutaneous malignant, 2}	155601
	{Melanoma-pancreatic cancer syndrome}	606719
CEBPA	?Leukemia, acute myeloid	601626
	Leukemia, acute myeloid, somatic	601626
CHEK2	Li-Fraumeni syndrome 2	609265
	Osteosarcoma, somatic	259500
	{Breast cancer, susceptibility to}	114480
	{Colorectal cancer, susceptibility to}	114500
	{Prostate cancer, familial, susceptibility to}	176807
CSF3R	?Neutrophilia, hereditary	162830
	Neutropenia, severe congenital, 7, autosomal recessive	617014
CTC1	Cerebroretinal microangiopathy with calcifications and cysts	612199
DDX11	Warsaw breakage syndrome	613398
DDX41	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}	616871
DKC1	Dyskeratosis congenita, X-linked	305000
DNAJC21	Bone marrow failure syndrome 3	617052
FL1 / EFTUD	Shwachman-Diamond syndrome 2	617941
ELANE	Neutropenia, cyclic	162800
	Neutropenia, severe congenital 1, autosomal dominant	202700
EPCAM	Colorectal cancer, hereditary nonpolyposis, type 8	613244
	Diarrhea 5, with tufting enteropathy, congenital	613217
ERCC4	Fanconi anemia, complementation group Q	615272
	Xeroderma pigmentosum, group F	278760
	Xeroderma pigmentosum, type F/Cockayne syndrome	278760
	XFE progeroid syndrome	610965
ERCC6L2	Bone marrow failure syndrome 2	615715
ESCO2	Juberg-Hayward syndrome	216100
	Roberts-SC phocomelia syndrome	268300
ETV6	Leukemia, acute myeloid, somatic	601626
	Thrombocytopenia 5	616216
FANCA	Fanconi anemia, complementation group A	227650
FANCB	Fanconi anemia, complementation group B	300514
FANCC	Fanconi anemia, complementation group C	227645
FANCD2	Fanconi anemia, complementation group D2	227646
FANCE	Fanconi anemia, complementation group E	600901
FANCF	Fanconi anemia, complementation group F	603467
FANCG	Fanconi anemia, complementation group G	614082
FANCI	Fanconi anemia, complementation group I	609053
FANCL	Fanconi anemia, complementation group L	614083
FANCM	?Premature ovarian failure 15	618096
	Spermatogenic failure 28	618086
G6PC3	Dursun syndrome	612541
	Neutropenia, severe congenital 4, autosomal recessive	612541
GATA1	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities	300835
	Leukemia, megakaryoblastic, with or without Down syndrome, somatic	190685
	Thrombocytopenia with beta-thalassemia, X-linked	314050
	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia	300367
GATA2	Emberger syndrome	614038
	Immunodeficiency 21	614172

GATA2	{Leukemia, acute myeloid, susceptibility to}	601626
	{Myelodysplastic syndrome, susceptibility to}	614286
GFI1	?Neutropenia, nonimmune chronic idiopathic, of adults	607847
	Neutropenia, severe congenital 2, autosomal dominant	613107
GSKIP	NA	NA
HAVCR2	T-cell lymphoma, subcutaneous panniculitis-like	618398
HAX1	Neutropenia, severe congenital 3, autosomal recessive	610738
HRAS	Bladder cancer, somatic	109800
	Congenital myopathy with excess of muscle spindles	218040
	Costello syndrome	218040
	Nevus sebaceous or woolly hair nevus, somatic	162900
	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic	163200
	Spitz nevus or nevus spilus, somatic	137550
	Thyroid carcinoma, follicular, somatic	188470
IKZF1	Immunodeficiency, common variable, 13	616873
JAK2	Erythrocytosis, somatic	133100
	Leukemia, acute myeloid, somatic	601626
	Myelofibrosis, somatic	254450
	Polycythemia vera, somatic	263300
	Thrombocythemia 3	614521
	{Budd-Chiari syndrome, somatic}	600880
KDM1A	Cleft palate, psychomotor retardation, and distinctive facial features	616728
KRAS	Arteriovenous malformation of the brain, somatic	108010
	Bladder cancer, somatic	109800
	Breast cancer, somatic	114480
	Cardiofaciocutaneous syndrome 2	615278
	Gastric cancer, somatic	613659
	Leukemia, acute myeloid, somatic	601626
	Lung cancer, somatic	211980
	Noonan syndrome 3	609942
	Oculoectodermal syndrome, somatic	600268
	Pancreatic carcinoma, somatic	260350
	RAS-associated autoimmune leukoproliferative disorder	614470
	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic	163200
LIG4	LIG4 syndrome	606593
	{Multiple myeloma, resistance to}	254500
MAP2K1	Cardiofaciocutaneous syndrome 3	615279
	Melorheostosis, isolated, somatic mosaic	155950
MAP2K2	Cardiofaciocutaneous syndrome 4	615280
MBD4	NA	NA
MECOM	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2	616738
MLH1	Colorectal cancer, hereditary nonpolyposis, type 2	609310
	Mismatch repair cancer syndrome 1	276300
	Muir-Torre syndrome	158320
MPL	Myelofibrosis with myeloid metaplasia, somatic	254450
	Thrombocythemia 2	601977
	Thrombocytopenia, congenital amegakaryocytic	604498
MSH2	Colorectal cancer, hereditary nonpolyposis, type 1	120435
	Mismatch repair cancer syndrome 2	619096
	Muir-Torre syndrome	158320
MSH6	Colorectal cancer, hereditary nonpolyposis, type 5	614350
	Mismatch repair cancer syndrome 3	619097
	{Endometrial cancer, familial}	608089
NAF1	NA	NA
NBN	Aplastic anemia	609135
	Leukemia, acute lymphoblastic	613065
	Nijmegen breakage syndrome	251260
NF1	Leukemia, juvenile myelomonocytic	607785
	Neurofibromatosis, familial spinal	162210
	Neurofibromatosis, type 1	162200
	Neurofibromatosis-Noonan syndrome	601321
	Watson syndrome	193520
NF2	Meningioma, NF2-related, somatic	607174
	Neurofibromatosis, type 2	101000
	Schwannomatosis, somatic	162091
NHP2	Dyskeratosis congenita, autosomal recessive 2	613987
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291
NOP10	Dyskeratosis congenita, autosomal recessive 1	224230
NPM1	Leukemia, acute myeloid, somatic	601626
NRAS	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic	614470
	Colorectal cancer, somatic	114500
	Epidermal nevus, somatic	162900
	Melanocytic nevus syndrome, congenital, somatic	137550
	Neurocutaneous melanosis, somatic	249400
	Noonan syndrome 6	613224
	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic	163200
	Thyroid carcinoma, follicular, somatic	188470
PALB2	Fanconi anemia, complementation group N	610832
	{Breast cancer, susceptibility to}	114480
	{Pancreatic cancer, susceptibility to, 3}	613348
PARN	Dyskeratosis congenita, autosomal recessive 6	616353
	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4	616371
PAX5	{Leukemia, acute lymphoblastic, susceptibility to, 3}	615545
PMS2	Colorectal cancer, hereditary nonpolyposis, type 4	614337
	Mismatch repair cancer syndrome 4	619101
POT1	{Glioma susceptibility 9}	616568
	{Melanoma, cutaneous malignant, susceptibility to, 10}	615848
PRF1	Aplastic anemia	609135
	Hemophagocytic lymphohistiocytosis, familial, 2	603553
	Lymphoma, non-Hodgkin	605027
PTPN11	LEOPARD syndrome 1	151100
	Leukemia, juvenile myelomonocytic, somatic	607785
	Metachondromatosis	156250
	Noonan syndrome 1	163950
RAD51	Fanconi anemia, complementation group R	617244
	Mirror movements 2	614508
	{Breast cancer, susceptibility to}	114480
RAD51C	Fanconi anemia, complementation group O	613390
	{Breast-ovarian cancer, familial, susceptibility to, 3}	613399
	Cardiomyopathy, dilated, 1NN	615916

RAF1	LEOPARD syndrome 2	611554
	Noonan syndrome 5	611553
RBBP6	NA	NA
RBM8A	Thrombocytopenia-absent radius syndrome	274000
RECQL	NA	NA
RECQL4	Baller-Gerold syndrome	218600
	RAPADILINO syndrome	266280
	Rothmund-Thomson syndrome, type 2	268400
RMRP	Anauxetic dysplasia 1	607095
	Cartilage-hair hypoplasia	250250
	Metaphyseal dysplasia without hypotrichosis	250460
RPL11	Diamond-Blackfan anemia 7	612562
RPL15	?Diamond-Blackfan anemia 12	615550
RPL18	?Diamond-Blackfan anemia 18	618310
RPL23	NA	NA
RPL26	?Diamond-Blackfan anemia 11	614900
RPL27	?Diamond-Blackfan anemia 16	617408
RPL31	NA	NA
RPL35	?Diamond-Blackfan anemia 19	618312
RPL35A	Diamond-Blackfan anemia 5	612528
RPL36	NA	NA
RPL5	Diamond-Blackfan anemia 6	612561
RPS10	Diamond-Blackfan anemia 9	613308
RPS17	Diamond-Blackfan anemia 4	612527
RPS19	Diamond-Blackfan anemia 1	105650
RPS24	Diamond-blackfan anemia 3	610629
RPS26	Diamond-Blackfan anemia 10	613309
RPS27	?Diamond-Blackfan anemia 17	617409
RPS28	Diamond Blackfan anemia 15 with mandibulofacial dysostosis	606164
RPS29	Diamond-Blackfan anemia 13	615909
RPS7	Diamond-Blackfan anemia 8	612563
RIT1	Noonan syndrome 8	615355
RTEL1	Dyskeratosis congenita, autosomal dominant 4	615190
	Dyskeratosis congenita, autosomal recessive 5	615190
	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3	616373
RUNX1	Leukemia, acute myeloid	601626
	Platelet disorder, familial, with associated myeloid malignancy	601399
SAMD9	MIRAGE syndrome	617053
	Monosomy 7 myelodysplasia and leukemia syndrome 2	619041
	Tumoral calcinosis, familial, normophosphatemic	610455
SAMD9L	Ataxia-pancytopenia syndrome	159550
	Monosomy 7 myelodysplasia and leukemia syndrome 1	252270
	Spinocerebellar ataxia 49	619806
SBDS	Shwachman-Diamond syndrome	260400
	{Aplastic anemia, susceptibility to}	609135
SH2B3	Erythrocytosis, somatic	133100
	Myelofibrosis, somatic	254450
	Thrombocythemia, somatic	187950
SLX4	Fanconi anemia, complementation group P	613951
SOS1	?Fibromatosis, gingival, 1	135300
	Noonan syndrome 4	610733
SRP54	Neutropenia, severe congenital, 8, autosomal dominant	618752
SRP72	Bone marrow failure syndrome 1	614675
TERC	Dyskeratosis congenita, autosomal dominant 1	127550
	{Aplastic anemia}	614743
	{Pulmonary fibrosis, idiopathic, susceptibility to}	614743
TERT	Dyskeratosis congenita, autosomal dominant 2	613989
	Dyskeratosis congenita, autosomal recessive 4	613989
	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1	614742
	{Leukemia, acute myeloid}	601626
	{Melanoma, cutaneous malignant, 9}	615134
TET2	Immunodeficiency 75	619126
	Myelodysplastic syndrome, somatic	614286
TINF2	Dyskeratosis congenita, autosomal dominant 3	613990
	Revesz syndrome	268130
TP53	Bone marrow failure syndrome 5	618165
	Breast cancer, somatic	114480
	Hepatocellular carcinoma, somatic	114550
	Li-Fraumeni syndrome	151623
	Nasopharyngeal carcinoma, somatic	607107
	Pancreatic cancer, somatic	260350
	{Adrenocortical carcinoma, pediatric}	202300
	{Basal cell carcinoma 7}	614740
	{Choroid plexus papilloma}	260500
	{Colorectal cancer}	114500
	{Glioma susceptibility 1}	137800
	{Osteosarcoma}	259500
UBE2T	Fanconi anemia, complementation group T	616435
USB1	Poikiloderma with neutropenia	604173
VPS45	Neutropenia, severe congenital, 5, autosomal recessive	615285
WAS	Wiskott-Aldrich syndrome	301000
WIPF1	Wiskott-Aldrich syndrome 2	614493
WRAP53	Dyskeratosis congenita, autosomal recessive 3	613988
XRCC2	?Fanconi anemia, complementation group U	617247
	?Premature ovarian failure 17	619146
	Spermatogenic failure 50	619145