Gen	Krankheit
ABCC8+del	Diabetes mellitus, noninsulin-dependent
	Diabetes mellitus, permanent neonatal
	Diabetes mellitus, transient neonatal 2
	Hyperinsulinemic hypoglycemia, familial, 1
	Hypoglycemia of infancy, leucine-sensitive
ACADM (=MCAD)+del	Acyl-CoA dehydrogenase medium chain deficiency (ACADM)
ACADVL (=VLCAD)	Acyl-CoA dehydrogenase very long chain deficiency (ACADVLD)
ACAN	Osteochondritis dissecans, short stature, and early-onset osteoarthritis (OD)
	Spondyloepimetaphyseal dysplasia (SEDM), aggrecan type
	Spondyloepiphyseal dysplasia, Kimberley type (SEDK)
ACP5	Spondyloenchondrodysplasia with immune dysregulation (SPENCDI)
ACTA2	Aortic aneurysm, familial thoracic 6 (AAT6)
	Congenital mydriasis
	Moyamoya disease 5 (MYMY5)
	Multisystemic smooth muscle dysfunction syndrome
ACVR1	Fibrodysplasia ossificans progressiva (FOP)
ADAMTSL2	Geleophysic dysplasia (GLPD)
AGPAT2+del	Lipodystrophy, congenital generalized, type 1 (CGL1)
AKT1	Breast cancer, somatic (BC)
	Colorectal cancer, somatic (CRC) (= Colon cancer)
	Ovarian cancer, somatic (OC)
	Proteus syndrome, somatic (PROTEUSS)
	Schizophrenia, susceptibility to
ALDOB+del	Fructose intolerance, hereditary (HFI)
ALPL (=TNSALP)+del	Hypophosphatasia (HOPS), adult, childhood, infantile or perinatal lethal
	Odontohypophosphatasia
ALX4+del	foramina parietalia permagna (FPP)
	frontonasal dysplasia type 2 (FND2)
	parietal foramina 2 (PFM2)
	Potocki-Shaffer syndrome (PSS) (= 11p11.2 deletion syndrome)
ANTXR1 (=TEM8)	Hemangioma, capillary infantile, susceptibility to (HCI)
ANTXR2 (=CMG2)	infantile systemic hyalinosis (ISH)
	juvenile hyaline fibromatosis (JHF)

APC+del+dup	Adenomatous polyposis coli, familial (FAP)
	Brain tumor-polyposis syndrome 2 (Gardner syndrome (GS))
	Colorectal cancer, somatic (CRC)
	Desmoid disease, hereditary (HDD)
	Gastric cancer, somatic
	Hepatoblastoma, somatic
	Medulloblastoma (MDB), somatic
APOC2+del	Apolipoprotein C-II deficiency
ARSE+del	X-linked recessive chondrodysplasia punctata type 1 (CPXR1)
ARTEMIS (offiz. DCLRE1C)+del	Omenn syndrome (Reticuloendotheliosis, familial, with eosinophilia)
	severe combined immunodeficiency Athabaskan-type (SCIDA)
	severe combined immunodeficiency with sensitivity to ionizing radiation (RS-SCID)
ARX+del+dup	Corpus callosum, agenesis of (ACC), with abnormal genitalia (Proud syndrome)
	Epileptic encephalopathy, early infantile, 1 (EIEE1) (West syndrome, X-linked)
	Hydranencephaly with abnormal genitalia
	Lissencephaly, X-linked 2 (LISX2) (Lissencephaly, X-linked, with ambiguous genitalia (XLAG))
	Mental retardation, X-linked, with or without seizures, ARX-related (MRXARX)
	Partington X-linked mental retardation syndrome (PRTS)
ATL1+del	Neuropathy, hereditary sensory, type ID (HSN1D)
	Spastic paraplegia-3A (SPG3A) (Strumpell-Lorrain syndrome)
ATP6V0A2+del	Cutis laxa, autosomal recessive, type IIA (ARCL2A)
	Wrinkly skin syndrome (WSS)
ATP7A+del+dup	Menkes disease (= Kinky hair disease)
	Occipital horn syndrome (OHS)
	Spinal muscular atrophy, distal, X-linked 3 (SMAX3)
ATRX+del+dup	Alpha-thalassemia myelodysplasia syndrome (ATMDS)
	Alpha-thalassemia/mental retardation syndrome, X-linked (ATRX)
	Mental retardation-hypotonic facies syndrome, X-linked (MRXHF1)
B3GALT6	Ehlers-Danlos syndrome, progeroid type, 2 (EDSP2)
	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures (SEMDJL1)
B3GALTL+del	Peters-plus syndrome (PpS)
B4GALT7	Ehlers-Danlos syndrome, progeroid form (EDSP)
BARX2	Role in chondrogenesis, myoblast fusion, ocular gland branching, muscle growth and regeneration
BCOR+del	Microphthalmia, syndromic 1 (MCOPS1)
	Microphthalmia, syndromic 2 (MCOPS2)

BMP1	Osteogenesis imperfecta,autosomal recessive
BMP2+del+dup	Brachydactyly, type A2 (BDA2)
	HFE hemochromatosis, modifier of
BMP4	Congenital 'healed' cleft lip (CHCL)
	Microphthalmia, syndromic 6 (MCOPS6)
	orofacial cleft type 11 (OFC11)
BMPR1A+del	Juvenile polyposis of stomach
	Juvenile polyposis syndrome (JPS)
	Polyposis syndrome, hereditary mixed, 2 (HMPS2)
BMPR1Adel	Chromosome 10q23 deletion syndrome
	Juvenile polyposis of infancy
BMPR1B	acromesomelic chondrodysplasia with genital anomalies (AMDGA)
	brachydactyly, type A2 (BDA2)
BRAF	Adenocarcinoma of lung, somatic
	Cardiofaciocutaneous syndrome (CFC syndrome)
	Colorectal cancer, somatic (CRC)
	LEOPARD syndrome 3
	Melanoma, malignant, somatic
	Nonsmall cell lung cancer, somatic
	Noonan syndrome 7
BSCL2+del	Lipodystrophy, congenital generalized, type 2 (CGL2) (Berardinelli-Seip congenital lipodystrophy, ty
	Neuronopathy, distal hereditary motor, type VA (HMN5A)
	Spastic paraplegia 17, autosomal dominant (SPG17) (Silver snydrome)
C1NH (offiz. SERPING1)+del	Angioedema, hereditary, type I (HAE1) (C1 esterase inhibitor, deficiency of)
	Angioedema, hereditary, type II (HAE2)
	Complement component 4 (C4), partial deficiency of
C6ST1 (offiz. CHST3)	Larsen syndrome, autosomal recessive
	Spondyloepiphyseal dysplasia with congenital joint dislocations (SED Omani type)
C7orf10	Glutaric aciduria III (GA3) (Glutaryl-CoA oxidase deficiency)
C8B	Complement component 8 deficiency, type II (C8D2)
C16orf57 (offiz. USB1)	Poikiloderma with neutropenia (PN) (Poikiloderma with neutropenia, Clericuzio-type)
CANT1+del	Desbuquois dysplasia (DBQD)
CAV1	Lipodystrophy, congenital generalized, type 3 (CGL3) (Berardinelli-Seip congenital lipodystrophy, ty
	Pulmonary hypertension, primary, 3 (PPH3)
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSL)
CCDC8	3M syndrome-3 (3M3)

CD96	C syndrome (CSYN) (Opitz trigonocephaly syndrome)
	C-like syndrome (CLSYN) (Opitz trigonocephaly-like syndrome)
CDK1 (=CDC2)	Schlüsselrolle bei der Zellzyklus-Kontrolle
CDKL5+del+dup	Angelman syndrome-like
	Epileptic encephalopathy, early infantile, 2 (EIEE2) (Rett syndrome, atypical, CDKL5-related)
CDKN1C+dup	Beckwith-Wiedemann syndrome (BWS)
	IMAGE (Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasia congenita, and anomalies) syndrome
CDMP1 (offiz. GDF5)	Acromesomelic dysplasia, Hunter-Thompson type (AMDH)
	Brachydactyly, type A2 (BDA2) (=Brachymesophalangy II)
	Brachydactyly, type C (BDC) (Brachydactyly, Haws type)
	Chondrodysplasia, Grebe type
	Fibular hypoplasia and complex brachydactyly (Du Pan syndrome (DPS))
	Multiple synostoses syndrome 2 (SYNS2)
	Osteoarthritis susceptibility 5 (OS5)
	Symphalangism, proximal (SYM1) (Cushing symphalangism)
CEBPA	Leukemia, acute myeloid
CEP19	Obesity, morbide
CGKI (offiz. PRKG1)	nitric oxide/cGMP signaling pathway, key mediators
	signal transduction processes in diverse cell types, important components
CHD7 (=KAL5)+del	CHARGE syndrome
	hypogonadotropic hypogonadism, idiopathic (IHH)
	idiopathic scoliosis 3, susceptibility to (IS3)
	Kallmann syndrome 5 (KAL5)
CHRNA3	Lung cancer susceptibility 2 (LNCR2)
	Smoking as a quantitative trait locus 3 (SQTL3)
CHST10	Rolle in der HNK1-Biosynthese (neurodevelopment and synaptic plasticity)
CHST11	Sulfatstoffwechsel
CHST12	Sulfatstoffwechsel
CHST3 (=C6ST1)	Larsen syndrome, autosomal recessive
	Spondyloepiphyseal dysplasia with congenital joint dislocations (= SED Omani type)
CMG2 (offiziell ANTXR2)	infantile systemic hyalinosis (ISH)
	juvenile hyaline fibromatosis (JHF)
C-MLP (offiz. MLP)	Myelofibrosis with myeloid metaplasia, somatic (MMM)
	Thrombocythemia 2 (THCYT2)
	Thrombocytopenia, congenital amegakaryocytic (CAMT)

COL1A1+del	Caffey disease (CAFFD) (infantile cortical hyperostosis)
	Ehlers-Danlos syndrome type I (EDS1) (Ehlers-Danlos syndrome gravis)
	Ehlers-Danlos syndrome type VIIA (EDS7A) (EDS, arthrochalasia type)
	Osteogenesis imperfecta type I (OI1) (OI tarda, OI with blue sclerae)
	Osteogenesis imperfecta type IIA (OI2A) (OI congenita, perinathal lethal form)
	Osteogenesis imperfecta type III (OI3) (OI, progressively deforming, with normal sclerae)
	Osteogenesis imperfecta type IV (OI4) (OI with normal sclerae)
	Osteoporosis, susceptibility to (OSTEOP)
COL1A2+del	Ehlers-Danlos syndrome, cardiac valvular form (EDSCV)
	Ehlers-Danlos syndrome, type VIIB (EDS7B) (EDS, arthrochalasia type)
	Marfan syndrome, atypical
	Osteogenesis imperfecta type IIA (OI2A) (OI congenita, perinathal lethal form)
	Osteogenesis imperfecta type III (OI3) (OI, progressively deforming, with normal sclerae)
	Osteogenesis imperfecta type IV (OI4) (OI with normal sclerae)
	Osteoporosis, postmenopausal
COL2A1+del	Achondrogenesis type II (ACG2) (ACG, Langer-Saldino type)
	Avascular necrosis of the femoral head (ANFH)
	Czech dysplasia (pseudorheumatoid dysplasia, progressive, with hypoplastic toes)
	Epiphyseal dysplasia, multiple, with myopia and deafness (EDMMD)
	Hypochondrogenesis
	Kniest dysplasia (KD)
	Legg-Calve-Perthes disease (LCPD; LCP)
	Osteoarthritis with mild chondrodysplasia (OACD)
	Otospondylomegaepiphyseal dysplasia (OSMED) (Nance-Sweeney chondrodysplasia)
	Platyspondylic lethal skeletal dysplasia, Luton type (PLSD-L)
	Platyspondylic skeletal dysplasia, Torrance type (PLSD-T)
	rhegmatogenous retinal detachment, autosomal dominant (DRRD)
	SED, Namaqualand type
	Spondyloepimetaphyseal dysplasia, Strudwick type (SEMD-STR)
	Spondyloepiphyseal dysplasia congenita (SEDC)
	Spondyloperipheral dysplasia (SPD)
	Stickler sydrome, type I, nonsyndromic ocular (STL1O)
	Stickler syndrome, type I (STL1) (Stickler syndrome, vitreous type 1)
	Vitreoretinopathy with phalangeal epiphyseal dysplasia
	Wagner syndrome type 2 (WGN2)

COL3A1+del	aortic aneurysm, familial abdominal 1 (AAA1)
	Ehlers-Danlos syndrome, type III (EDSIII; EDS3) (EDS, hypermobility type)
	Ehlers-Danlos syndrome, type IV, autosomal dominant (EDS IV; EDS4) (EDS, vascular type)
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps (HANAC)
	Brain small vessel disease with Axenfeld-Rieger anomaly
	Brain small vessel disease with hemorrhage (BSVDH) (infantile hemiparesis)
	Porencephaly, familial (PCEPH)
COL4A2	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps (HANAC)
	Brain small vessel disease with Axenfeld-Rieger anomaly
	Brain small vessel disease with hemorrhage (BSVDH) (infantile hemiparesis)
COL4A3	Alport syndrome, autosomal dominant (APSAD)
	Alport syndrome, autosomal recessive (APSAR)
	Hematuria, benign familial (BFH) (Thin-basement-mambrane nephropathy)
COL4A4	Alport syndrome, autosomal recessive
	Hematuria, benign familial (BFH) (=Thin-basement-membrane nephropathy)
COL4A5+del	Alport syndrome X-linked (APSX)
	Leiomatosis, diffuse, with Alport syndrome (DL-ATS)
COL5A1+del+dup	Ehlers-Danlos syndrome, type I (EDS I) (EDS1) (EDS, severe classic type)
	Ehlers-Danlos syndrome, type II (EDS II) (EDS2) (EDS, mild classic type)
COL5A2	Ehlers-Danlos syndrome, type I (EDS I) (EDS1) (EDS, severe classic type)
COL9A1	epiphyseal dysplasia, multiple, type 6 (EDM6)
	Stickler Syndrome, autosomal recessive, COL9A1-related (COL9A1ARSTL)
COL9A2	Epiphyseal dysplasia, multiple, 2 (EDM2)
	Intervertebral disc disease (IDD), susceptibility to
	Stickler syndrome, type V (STL5)
COL9A3	Epiphyseal dysplasia, multiple, 3 (EDM3)
	Epiphyseal dysplasia, multiple, with myopathy
	Hearing loss, non-syndromic
	Intervertebral disc disease, susceptibility to
	Pseudoachondroplasia (PSACH)
COL10A1	Metaphyseal chondrodysplasia, Schmid type (MCDS)
COL11A1+del	Fibrochondrogenesis
	Lumbar disc herniation, susceptibility to
	Marshall syndrome (MARSHS)
	Stickler syndrome, type II (STL2)
	Stickler syndrome, vitreous type 2

COL11A2	deafness, autosomal dominant type 13 (DFNA13)
	deafness, autosomal recessive type 53 (DFNB53)
	OSMED, heterozygous (WZS)
	Otospondylomegaepiphyseal dysplasia (OSMED)
	Stickler syndrome, type III (STL3)
	Weissenbacher-Zweymueller syndrome (WZS)
COMP+del	Epiphyseal dysplasia, Fairbank type (EDMF)
	Epiphyseal dysplasia, multiple 1 (EDM1)
	Epiphyseal dysplasia, Ribbing type (EDMR)
	Pseudoachondroplasia (PSACH)
CPT2	CPT deficiency, hepatic, type II (Carnitine palmitoyltransferase II deficiency, infantile)
	CPT II deficiency, lethal neonatal (Carnitine palmitoyltransferase 2 deficiency, lethal neonatal)
	CPT II deficiency, myopathic (Carnitine palmitoyltransferase 2 deficiency, late-onset)
	Encephalopathy, acute, infection-induced, 4, susceptibility to (IIAE4)
CPX (offiz. EBP)	Chondrodysplasia punctata, X-linked dominant, 2 (CDPX2) (Conradi-Hunermann-Happle syndrome
CREB3L2	chondrocyte differentiation, key role in
	epiphyseal cartilage, key role in formation of
	Fibromyxoid sarcoma, low grade (LGFMS)
	transcription of unfolded protein response target genes, regulation of
CREBBP+del	16p13.3 deletion syndrome
	leukemia, acute myeloid
	Rubinstein-Taybi syndrome type 1 (RSTS1)
CRELD1	Atrioventricular septal defect, partial, with heterotaxy syndrome
	Atrioventricular septal defect, susceptibility to, 2 (AVSD2)
CRTAP	Osteogenesis imperfecta, type IIB (OI2B)
	Osteogenesis imperfecta, type VII (OI7)
CSGALNACT1+del	role in enchondral ossification and aggrecan metabolism; required for normal cartilage development
CTC1	Cerebroretinal microangiopathy with calcifications and cysts (CRMCC) (Coats plus syndrome)
CTNNB1	Adenomas**,** salivary gland pleomorphic (PSA)
	Colorectal cancer (CRC)
	Hepatoblastoma
	Hepatocellular carcinoma (HCC)
	Medulloblastoma (MDB)
	Mesothelioma, malignant; susceptibility to (MESOM)
	Ovarian cancer (OC)
	Pilomatricoma (PTR)

CTSK	Pycnodysostosis (PKND)
CUL7	3-M syndrome 1 (3M1)
CX26 (offiz. GJB2)	Bart-Pumphrey syndrome (BPS)
	Deafness, autosomal dominant 3A (DFNA3A)
	Deafness, autosomal recessive 1A (DFNB1A)
	Hystrix-like ichthyosis with deafness (HID syndrome)
	Keratitis-ichthyosis-deafness syndrome (KID syndrome)
	Keratoderma, palmoplantar, with deafness (PPKDFN)
	Vohwinkel syndrome (VS)
CXCR4	Myelokathexis, isolated
	WHIM (Warts, hypogammaglobulinemia, infections, and myelokathexis) syndrome
CYP7B1	Bile acid synthesis defect, congenital, 3 (CBAS3)
	Spastic paraplegia 5A, autosomal recessive (SPG5A)
D2HGDH+del	D-2-Hydroxyglutaric aciduria 1 (D2HGA) (D2HGA1)
DBP+del (offiz. HSD17B4)	D-bifunctional protein deficiency (DBPD)
	Perrault syndrome (PRS)
DCLRE1C(=ARTEMIS)+del	Omenn syndrome (Reticuloendotheliosis, familial, with eosinophilia)
	severe combined immunodeficiency Athabaskan-type (SCIDA)
	severe combined immunodeficiency with sensitivity to ionizing radiation (RS-SCID)
DHCR7+del	Smith-Lemli-Opitz syndrome (SLOS)
DKC1	Dyskeratosis congenita, X-linked
DLD (=LAD)	Dihydrolipoamide dehydrogenase deficiency (DLDD) (Maple syrup urine disease, type III)
DLL3	Spondylocostal dysostosis, autosomal recessive, 1 (SCDO1)
DNASE2	apoptosis, major role during
	erythropoiesis, major role during
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome (ICF1)
DTDST (offiziell SLC26A2)	Achondrogenesis Ib (ACG1B)
	Atelosteogenesis II (AO2)
	De la Chapelle dysplasia (DLCD)
	Diastrophic dysplasia (DTD)
	Diastrophic dysplasia, broad bone-platyspondylic variant (DTDB)
	Epiphyseal dysplasia, multiple, 4 (EDM4)
DYM+dup	Dyggve-Melchior-Clausen disease (DMC)
	Smith-McCort dysplasia (SMC)

DYNC2H1	Asphyxiating thoracic dystrophy 3 (ATD3)
	Short rib-polydactyly syndrome, type II, digenic (SRPS2)
	Short rib-polydactyly syndrome, type III (SRPS3) (Verma-Naumoff syndrome)
EBP (=CPX)	Chondrodysplasia punctata, X-linked dominant, 2 (CDPX2) (Conradi-Hunermann-Happle syndrome
ECSIT	Complex I deficiency, mitochondrial
EDN3	Central hypoventilation syndrome, congenital (CCHS) (Ondine curse)
	Hirschsprung disease, susceptibility to, 4 (HSCR4) (aganglionic megacolon (MGC))
	Waardenburg syndrome, type 4B (WS4B) (WS4B with Hirschsprung disease)
EFNB1+del+dup	Craniofrontonasal syndrome (CFNS)
ELN+del	Cutis laxa, autosomal dominant, 1 (ADCL1)
	Supravalvar aortic stenosis (SVAS) (SVAS, Eisenberg type)
ELNdel	Williams-Beuren syndrome (WBS)
EP300+del	Colorectal cancer (CRC)
	Rubinstein-Taybi syndrome 2 (RSTS2)
ERCC3 (=XPB)	Ichthyosiform erythroderma with hair abnormality and mental and growth retardation
	Trichothiodystrophy, photosensitive (TTDP)
	Xeroderma pigmentosum, group B (XBP)
	Xeroderma pigmentosum/Cockayne syndrome (XPB/CS)
ETFA+del	Glutaric acidemia IIA (ETFA deficiency)
	Multiple acyl-CoA dehydrogenase deficiency (MADD) (Glutaric acidemia IIA)
ETFB	Glutaric acidemia IIB (ETFB deficiency)
	Multiple acyl-CoA dehydrogenase deficiency (MADD) (Glutaric acidemia IIB)
ETFDH	Glutaric acidemia IIC (GA2C) (ETFDH deficiency)
	Multiple acyl-CoA dehydrogenase deficiency (MADD) (Glutaric acidemia IIC)
EVC (=EVC1)+del	Ellis-van Creveld syndrome (EVC) (chondroectodermal dysplasia)
	Weyers acrodental dysostosis (WAD) (Curry-Hall syndrome)
EVC2+del	Ellis-van Creveld syndrome (EVC) (chondroectodermal dysplasia)
	Weyers acrodental dysostosis (WAD) (Curry-Hall syndrome)
EXT1+del (=TRPS2)	Chondrosarcoma (CHDSA)
	Exostoses, multiple, type 1 (EXT1) (Osteochondromas, multiple)
	Langer-Giedion syndrome (LGS) (Trichorhinophalangeal syndrome, type II (TRPS2))
EXT2+del	Chondrosarcoma (CHDSA)
	Exostoses, multiple, type 2 (EXT 2) (= Osteochondromas, multiple)
	Potocki-Shaffer syndrome (PSS) (11p11.2 Deletion Syndrome)
EZH2	Weaver syndrome (WES) (Weaver-Smith syndrome (WSS))

FAM110B	tumor progression, may be involved in
FBN1+del	Acromicric dysplasia (ACMICD)
	Aortic aneurysm, ascending, and dissection
	Ectopia lentis, familial (EL)
	Geleophysic dysplasia 2 (GPHYSD2)
	Marfan syndrome (MFS) (Marfan syndrome, type 1 (MFS1))
	MASS syndrome (overlap connective tissue disease (OCTD))
	Stiff skin syndrome (SSKS)
	Weill-Marchesani syndrome 2 (WMS2)
FBN2	Arthrogryposis, distal, type 9 (DA9) (Beals syndrome)
FBXW8	3-M syndrome, candidate
FGD1+del	Aarskog-Scott syndrome (AAS) (Faciogenital dysplasia (FGDY))
	Mental retardation, X-linked syndromic 16 (MRXS16)
FGF8	Hypogonadotropic hypogonadism
	Kallmann syndrome 6 (KAL6)
FGF9	Multiple synostoses syndrome 3 (SYNS3)
FGF10+del	Aplasia of lacrimal and salivary glands (ALSG)
	LADD (Lacrimoauriculodentodigital) syndrome (LADDS) (Levy-Hollister syndrome)
FGF17	embryonic development, important role in the regulation of; embryonic brain, signaling molecule in t induction and patterning of the
FGF23	Hypophosphatemic rickets, autosomal dominant (ADHR) (Vitamin D-resistant rickets, AD)
	Osteomalacia, tumor-induced
	Tumoral calcinosis, hyperphosphatemic, familial HFTC) (Morbus Teutschlaender)
FGFR1+del (=KAL2)	Hypogonadotropic hypogonadism
	Jackson-Weiss syndrome (JWS) (craniosynostosis, midfacial hypoplasia, and foot anomalies)
	Kallmann syndrome 2 (KAL2)
	Osteoglophonic dysplasia (OGD)
	Pfeiffer syndrome (PS) (Acrocephalosyndactyly, type V (ACS5))
	Trigonocephaly, nonsyndromic (TRICEPH) (Craniosynostosis, metopic)

FGFR2+del	Antley-Bixler syndrome, type 2 (ABS2) (ABS without genital anomalies or disordered steroidogenes
	Apert syndrome (APRS) (Acrocephalosyndactyly type 1 (ACS1))
	Beare-Stevenson cutis gyrata syndrome (BSTVS)
	Craniofacial-skeletal-dermatologic dysplasia
	Craniosynostosis, nonspecific
	Crouzon syndrome (CS) (Craniofacial dysostosis type I (CFD1))
	Gastric cancer, somatic
	Jackson-Weiss syndrome (JWS) (craniosynostosis, midfacial hypoplasia, and foot anomalies)
	LADD (Lacrimoauriculodentodigital) syndrome (LADDS) (Levy-Hollister syndrome)
	Pfeiffer syndrome (PS) (Acrocephalosyndactyly, type V (ACS5))
	Saethre-Chotzen syndrome (SCS) (Acrocephalodyndactyly, type III (ACS3))
	Scaphocephaly and Axenfeld-Rieger anomaly
	Scaphocephaly, maxillary retrusion, and mental retardation
FGFR3	Achondroplasia (ACH)
	Bladder cancer, somatic (BLC)
	CATSHL (camptodactyly tall stature and hearing loss) syndrome
	Cervical cancer, somatic (CERCA)
	Crouzon syndrome with acanthosis nigricans
	Hypochondroplasia
	Keratosis, seborrheic, somatic
	LADD (Lacrimoauriculodentodigital) syndrome (LADDS) (Levy-Hollister syndrome)
	Muenke syndrome
	multiple myeloma (MM)
	Nevus, keratinocytic, nonepidermolytic (KNEN)
	Spermatocytic seminoma, somatic
	Thanatophoric dysplasia, type I (TD1)
	Thanatophoric dysplasia, type II
FKBP10	Osteogenesis imperfecta, type XI (OI11)
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss (EDSKMH)
FLI1	Ewing sarcoma (ES)
	Hemangiomatosis
	Leukemia, acute lymphoblastic
	Paris-Trousseau thrombocytopenia (TCPT)

FLNB	Atelosteogenesis, type I (AOI) (Spondylohumerofemoral hypoplasia)
	Atelosteogenesis, type III (AOIII) (AO3)
	Boomerang dysplasia
	Larsen syndrome (LRS), autosomal dominant
	Spondylocarpotarsal synostosis syndrome (SCT)
FOXC2+del	Lymphedema, hereditary, type 2 (LMPH2) (Meige disease)
	Lymphedema-distichiasis syndrome (LYDS)
	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus
	Yellow nail syndrome (YNS) (Lymphedema and yellow nails (LYYN))
FOXG1+del	Rett syndrome, congenital variant (RTTCV)
FOXI1	Enlarged vestibular aqueduct (EVA)
	Pendred syndrome (PDS)
FOXL2+del+dup	Blepharophimosis, epicanthus inversus, and ptosis, type 1 (BPES1)
	Blepharophimosis, epicanthus inversus, and ptosis, type 2 (BPES2)
	Premature ovarian failure 3
G6PC	Glycogen storage disease Ia (GSD1A) (Von Gierke disease)
GALNS+del	Mucopolysaccharidosis type 4A (MPS4A) (Morquio A syndrome)
GALT+del	Galactosemia (Galactose-1-phosphate uridylyltransferase (GALT) deficiency)
GAMT	Cerebral creatine deficiency syndrome 2 (CCDS2) (Guanidinoacetate methyltransferase (GAMT) de
GATA1	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities (XLANP)
	Leukemia, megakaryoblastic, with or without Down syndrome, somatic
	Thrombocytopenia with beta-thalassemia, X-linked (XLTT)
	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia (XLTDA)
GATA2	GATA2 deficiency (MonoMAC (monocytopenia, NK- and B- lymphocytopenia, severe infections with avium complex (MAC), and risk of progression to MDS/AML))
	Emberger syndrome
GATA4+del	Atrial septal defect 2 (ASD2)
	Atrioventricular septal defect 4 (AVSD4)
	Tetrology of Fallot (TOF)
	Ventricular septal defect 1 (VSD1)
GATM (= AGAT)	Arginine:glycine amidinotransferase (AGAT) deficiency (Creatine deficiency syndrome due to AGAT deficiency)

GDF5 (=CDMP1)	Acromesomelic dysplasia, Hunter-Thompson type (AMDH)
	Brachydactyly, type A2 (BDA2) (Brachymesophalangy II)
	Brachydactyly, type C (BDC) (Brachydactyly, Haws type)
	Chondrodysplasia, Grebe type (acromesomelic dysplasia Grebe type (AMDG))
	Fibular hypoplasia and complex brachydactyly (Du Pan syndrome (DPS))
	Multiple synostoses syndrome 2 (SYNS2)
	Osteoarthritis, susceptibility 5 (OS5)
	Symphalangism, proximal (SYM1) (Cushing symphalangism)
GDF6	Klippel-Feil syndrome 1, autosomal dominant (KFS1)
	Microphthalmia, isolated 4 (MCOP4)
	Microphthalmia, isolated, with coloboma 6, digenic (MCOPCB6)
	Ocular coloboma
GH1+del+dup	Growth hormone deficiency, isolated, type IA (IGHD1A) (Pituitary dwarfism I)
	Growth hormone deficiency, isolated, type IB (IGHD1B) (Dwarfism of Sindh)
	Growth hormone deficiency, isolated, type II (IGHD2)
	Kowarski syndrome (KWKS) (Biodefective growth hormone)
GHRHR	Growth hormone deficiency, isolated, type IB (IGHD1B) (Dwarfism of Sindh)
GJB2 (=CX26)	Bart-Pumphrey syndrome (BPS)
	Deafness, autosomal dominant 3A (DFNA3A)
	Deafness, autosomal recessive 1A (DFNB1A)
	Hystrix-like ichthyosis with deafness (HID syndrome)
	Keratitis-ichthyosis-deafness syndrome (KID syndrome)
	Keratoderma, palmoplantar, with deafness (PPKDFN)
	Vohwinkel syndrome (VS)
GJC2	Leukodystrophy, hypomyelinating, 2 (HLD2) (Pelizaeus-Merzenbacher-like disease, 1 (PMLD1))
	Lymphedema, hereditary, IC (LMPH1C)
	Spastic paraplegia 44, autosomal recessive (SPG44)
GLA+del	Fabry disease (Angiokeratoma corporis diffusum)
	Fabry disease, cardiac variant
GLB1+del+dup	Gangliosidosis, generalized GM1, late infantile type
	GM1-gangliosidosis, type I, II and III
	Mucopolysaccharidosis type IVB (MPS4B) (Morquio syndrome B)

GLI3+del+dup	Congenital hypothalamic hamartoma syndrome (CHHS)
	Crossed polydactyly, type I (CP1)
	Greig cephalopolysyndactyly syndrome (GCPS) (Polysyndactyly with peculiar skull shape)
	Hypothalamic hamartomas, somatic
	Pallister-Hall syndrome (PHS) (Hypothalamic hamartoblastoma, hypopituitarism, imperforate anus, postaxial polydactyly)
	Polydactyly, postaxial, type A1 (PAPA1)
	Polydactyly, postaxial, type B (PAPB)
	Polydactyly, preaxial, type IV (Polysyndactyly, uncomplicated)
GLRA1+del	Hyperekplexia, hereditary 1 (HKPX1)
GNPTAB+del+dup	Mucolipidosis II alpha/beta (ML II) (I-cell disease (ICD))
	Mucolipidosis III alpha/beta (ML III) (Pseudo-Hurler polydystrophy)
GNRH1	Eunuchoidism, familial hypogonadotropic (Gonadotropin deficiency, familial idiopathic (FIGD))
	Hypogonadotropic hypogonadism
GNRHR	Fertile eunuch syndrome (Hypogonadism with spermatogenesis)
	Hypogonadotropic hypogonadism
GORAB (=SCYL1BP1)	Geroderma osteodysplasticum (GO) (Walt Disney dwarfism)
GPC3	Wilms tumor, somatic
GPC3+del	Simpson-Golabi-Behmel syndrome, type 1 (SGBS1)
GPC4del+dup	Simpson-Golabi-Behmel syndrome, type 1 (SGBS1)
HDAC4+del	Brachydactyly-mental retardation syndrome (BDMR) (Albright hereditary osteodystrophy-like syndrc
	Chromosome 2q37.2 deletion syndrome
HES7	Spondylocostal dysostosis 4, autosomal recessive (SCDO4)
HESX1+del	Growth hormone deficiency with pituitary anomalies
	Pituitary hormone deficiency, combined, 5
	Septooptic dysplasia
HMGA2	Hamartomas, pulmonary chondroid
	Leiomyoma, uterine, somatic (UL)
	Lipomatosis, multiple
HOXA1	Athabaskan brainstem dysgenesis syndrome (ABSD) (Navajo brainstem syndrome)
	Bosley-Salih-Alorainy syndrome (BSAS)
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia
HOXB1	tooth development, role in ?
HOXC6	Shoulder girdle complex, role in the development of the marsupial's

HOXD13+del	Brachydactyly, type D (BDD) (Stub thumb)
	Brachydactyly, type E1 (BDE1)
	Brachydactyly-syndactyly syndrome (BDSD)
	Syndactyly, type V (SDTY5) (Syndactyly with metacarpal and metatarsal fusion)
	Synpolydactyly 1 (SPD1)
	Synpolydactyly with foot anomalies
	VACTERL (vertebral defects, anal atresia, cardiac malformations, tracheoesophageal fistula with esophageal atresia, and radial or renal dysplasia, limb anomalies) association
HPGD	Cranioosteoarthropathy (COA)
	Digital clubbing, isolated congenital (Acropachy, hereditary)
	Hypertrophic osteoarthropathy, primary, autosomal recessive, 1 (PHOAR1) (Pachydermoperiostosis autosomal recessive) (Touraine-Solente-Gole syndrome)
HRAS	Bladder cancer, somatic (BLC)
	Costello syndrome (Faciocutaneoskeletal syndrome (FCSS))
	Hurthle cell thyroid carcinoma (HCTC)
	Myopathy, congenital, with excess of muscle spindles (CMEMS)
	Thyroid carcinoma, follicular, somatic (FTC)
HSD17B4+del (= DBP)	D-bifunctional protein deficiency (DBPD)
	Perrault syndrome (PRS)
HSPB11 (=IFT25)	apoptotic cell death, role in prevention of
HSPG2+del	Dyssegmental dysplasia, Silverman-Handmaker type (DDSH)
	Schwartz-Jampel syndrome, type 1 (SJS1)
IDH1	Glioma (GLM)
	Leukemia, acute myeloid (AML)
	Maffucci syndrome (Enchondromatosis, multiple, Maffucci type)
	Metaphyseal chondromatosis with urinary excretion of D-2-hydroxy-glutaric acid (MC-HGA)
	Ollier disease (Enchondromatosis, multiple, Ollier type)
	Osteoarthritis, familial early-onset (FOA), susceptibility to
IDH2	D-2-hydroxyglutaric aciduria type 2 (D2HGA2)
	Maffucci syndrome (= Enchondromatosis, multiple, Maffucci type)
	Metaphyseal chondromatosis with urinary excretion of D-2-hydroxy-glutaric acid (MC-HGA)
	Ollier disease (Enchondromatosis, multiple, Ollier type)
IFITM5	Osteogenesis imperfecta, type 5 (OI5)
IFT20	ciliary assembly, function in
IFT25 (offiz. HSPB11)	apoptotic cell death, role in prevention of
IFT27	protein is ras-related, but the function is unknown

IFT43	Cranioectodermal dysplasia 3 (CED3)
IFT80	Asphyxiating thoracic dystrophy 2 (ATD2) (Jeune syndrome 2)
IFT122	Cranioectodermal dysplasia type 1 (CED1)
IGFALS	Acid-labile subunit, deficiency of (ALSD)
IHH	Acrocapitofemoral dysplasia (ACFD)
	Brachydactyly, type A1 (BDA1)
IKBKG (=NEMO)+del	Atypical mycobacteriosis, familial (AMCBX1)
	Ectodermal dysplasia, anhidrotic, with immune deficiency (EDA-ID)
	Ectodermal dysplasia, hypohidrotic, with immune deficiency (HED-ID)
	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency
	Immunodeficiency, isolated
	Incontinentia pigmenti, type II, formerly
	Invasive pneumococcal disease, recurrent isolated, 2 (IPD2)
IMPAD1	chondrodysplasia with joint dislocations, GPAPP type (CDP-GPAPP)
INI1 (offiz. SMARCB1)+del	Rhabdoid predisposition syndrome 1 (RPS1)
	Rhabdoid tumor (RDT) (Teratoid tumor, atypical)
	Schwannomatosis (Neurofibromatosis type 3 (NF3))
IRF6+del	Orofacial cleft 6, susceptibility to (OFC6)
	Popliteal pterygium syndrome 1 (PPS)
	van der Woude syndrome 1 (VWS1) (Cleft lip and/or palate with mucous cysts of lower lip)
IVD	Isovaleric acidemia (IVA)
JAG1+del+dup	Alagille syndrome, type 1 (ALGS1)
	Deafness, congenital heart defects, and posterior embryotoxon
	Tetralogy of Fallot (TOF)
JAK2	Budd-Chiari syndrome, susceptibility to (BDCHS)
	Erythrocytosis, somatic
	Leukemia, acute myelogenous (AML) (= Leukemia, acute myeloid)
	Myelofibrosis, somatic
	Polycythemia vera (PV) (= Polycythemia rubra vera (PRV))
	Thrombocythemia 3 (THCYT3) (= Thrombocytosis)

KAL1+del	Kallmann syndrome 1 (KAL1) (Hypogonadotropic hypogonadism and anosmia (HHA))
KAL2+del (offiz. FGFR1)	Hypogonadotropic hypogonadism
	Jackson-Weiss syndrome (JWS) (craniosynostosis, midfacial hypoplasia, and foot anomalies)
	Kallmann syndrome 2 (KAL2)
	Osteoglophonic dysplasia (OGD)
	Pfeiffer syndrome (PS) (Acrocephalodyndactyly, type V (ACS5))
	Trigonocephaly, nonsyndromic (TRICEPH) (Craniosynostosis, metopic)
KAL3 (offiz. PROKR2)	Kallmann syndrome 3
KAL4 (offiz. PROK2)	Hypogonadism, hypogonadotropic
	Kallmann syndrome 4
KAL5 (off. CHD7)+del	CHARGE syndrome
	hypogonadotropic hypogonadism, idiopathic (IHH)
	idiopathic scoliosis 3, susceptibility to (IS3)
	Kallmann syndrome 5 (KAL5)
KCNJ11	Diabetes mellitus, permanent neonatal (PNDM) (Diabetes mellitus, permanent, of infancy (PDMI)
	Diabetes mellitus, permanent neonatal, with neurologic features
	Diabetes mellitus, transient neonatal, 3 (TNDM3)
	Diabetes mellitus, type 2, susceptibility to (= noninsulin-dependent diabetes mellitus (NIDDM), susc to)
	Hyperinsulinemic hypoglycemia, familial, 2 (HHF2)
KCTD1	Scalp-ear-nipple syndrome (SENS) (Finlay-Marks syndrome)
KIF7	Acrocallosal syndrome
	Hydrolethalus syndrome 2
	Joubert syndrome 12
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2 (SEMDJL2)
KISS1R	Hypogonadotropic hypogonadism
	Precocious puberty, central
KIT+del	Gastrointestinal stromal tumor, familial (GIST)
	Leukemia, acute myeloid (AML)
	Mast cell disease (= Mastocytosis)
	Testicular germ cell tumors (TGCT) (Male germ cell tumor (MGCT))
	Piebald trait (PBT) (Piebaldism)
	Urticaria pigmentosa

KRAS	Bladder cancer, somatic
	Breast cancer, somatic
	Cardiofaciocutaneous syndrome 2 (CFC2)
	Gastric cancer, somatic
	Leukemia, acute myeloid (AML)
	Lung cancer, somatic
	Noonan syndrome 3 (NS3)
	Pancreatic cancer, somatic
	Schimmelpfennig-Feuerstein-Mims syndrome (SFM), somatic mosaic
KRT14	Dermatopathia pigmentosa reticularis (DPR)
	Epidermolysis bullosa simplex Dowling-Meara type (EBS-DM)
	Epidermolysis bullosa simplex Koebner type (EBS-K)
	Epidermolysis bullosa simplex Weber-Cockayne type (EBS-WC)
	Epidermolysis bullosa simplex, autosomal recessive (AREBS)
	Naegeli-Franceschetti-Jadassohn syndrome (NFJS) (Naegeli syndrome)
L2HGDH+del	L-2-Hydroxyglutaric aciduria
LAD (offiz. DLD)	Dihydrolipoamide dehydrogenase deficiency (DLDD) (E3 deficiency) (Maple syrup urine disease, ty
LAMP2+del	Danon disease (DAND) (= Glycogen storage disease type 2B (GSD2B), formerly)
LAMR1 (offiz. RPSA)	Asplenia, isolated congenital
LEPRE1+del	Osteogenesis imperfecta, type 8 (OI8)
LFNG	Spondylocostal dysostosis, type 3 (SCDO3)
LHX3+del	Pituitary hormone deficiency, combined, 3 (CPHD3)
LHX4+del	Leukemia, acute lymphoblastic (ALL)
	Pituitary hormone deficiency, combined, 4 (CPHD4)
LIFR	Adenomas, salivary gland pleomorphic (PSA; SGPA)
	Stueve-Wiedemann syndrome (SWS) (Schwartz-Jampel syndrome, type 2 (SJS2))

LMNA+del	Cardiomyopathy, dilated, 1A (CMD1A)
	Charcot-Marie-Tooth disease, axonal, type 2B1 (CMT2B1)
	Emery-Dreifuss muscular dystrophy 2, autosomal dominant (EDMD2)
	Emery-Dreifuss muscular dystrophy, atypical, autosomal recessive
	Heart-hand syndrome, Slovenian type
	Hutchinson-Gilford progeria syndrome (HGPS) (= Progeria)
	Lipodystrophy, familial partial, type 2 (FPLD2) (Dunnigan type)
	Malouf syndrome (Cardiomyopathy, dilated, with hypergonadotropic hypogonadism)
	Mandibuloacral dysplasia with type A lipodystrophy (MADA) (Craniomandibular dermatodysostosis)
	Mandibuloacral dysplasia with type A lipodystrophy, atypical
	Muscular dystrophy, congenital, LMNA-related (MDCL)
	Muscular dystrophy, limb-girdle, type 1B (LGMD1B)
	Progeria syndrome, childhood onset
	Restrictive dermopathy, lethal (Tight skin contracture syndrome, lethal)
LMNB2	Lipodystrophy, partial, acquired, susceptibility to (APLD) (Barraquer-Simons syndrome)
LMX1B+del	Nail-patella syndrome (NPS) (Onychoosteodysplasia)
	Nail-patella syndrome with primary open angle glaucoma (POAG)
LNP (offiz. NUSAP1)	spindle microtubule organization, role in
LPIN2	Majeed syndrome (MAJEEDS) (chronic recurrent multifocal osteomyelitis (CRMO), congenital dyserythropoietic anemia (CDA), and neutrophilic dermatosis (Sweet syndrome))
LPL+del+dup	Hyperlipidemia, familial combined (FCHL)
	Hyperlipoproteinemia, type I (Lipoprotein lipase (LPL) deficiency)
MAFB	Multicentric carpotarsal osteolysis syndrome (MCTO)
MAP2K1 (=MEK1)	Cardiofaciocutaneous syndrome 3 (CFC3)
MAP2K2 (=MEK2)	Cardiofaciocutaneous syndrome 4 (CFC4)
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III d
	Methionine adenosyltransferase (MAT) deficiency, autosomal recessive
MATN3	Epiphyseal dysplasia, multiple, 5 (EDM5)
	Osteoarthritis, susceptibility to, 2 (OS2)
	Spondyloepimetaphyseal dysplasia (SEMD)
MBOAT1+del	Brachydactyly-syndactyly syndrome, novel
MC4R	Obesity, autosomal dominant (OBESITY)
MCAD (offiz. ACADM)+del	Acyl-CoA dehydrogenase medium chain (ACADM) deficiency = medium chain Acyl-CoA dehydroge deficiency (MCAD)

MECP2+del+dup	Angelman syndrome-like
	Autism susceptibility, X-linked 3 (AUTSX3)
	Encephalopathy, neonatal severe
	Mental retardation, X-linked syndromic, Lubs type (MRXSL) (MECP2 duplication syndrome)
	Mental retardation, X-linked, syndromic 13 (MRXS13)
	Rett syndrome (RTT; RTS)
	Rett syndrome, preserved speech variant (RTT-PSV) (Rett syndome, atypical)
MED12	Lujan-Fryns syndrome (Mental retardation, X-linked, with marfanoid habitus)
	Ohdo syndrome, X-linked (OHDOX) (Blepharophimosis-mental retardation syndrome, Maat-Kievit-B type)
	Opitz-Kaveggia syndrome (OKS) (FG syndrome 1 (FGS1))
MEF2C+del	Bone development and chondrocyte hypertrophy, control of
	Chondrocyte hypertrophy and bone development, control of
	Mental retardation, autosomal dominant 20 (MRD20)
	Chromosome 5q14.3 deletion syndrome
MEK1 (offiz. MAP2K1)	Cardiofaciocutaneous syndrome 3 (CFC3)
MEK2 (offiz. MAP2K2)	Cardiofaciocutaneous syndrome 4 (CFC4)
MESP2	Spondylocostal dysostosis type 2 (SCDO2)
MID1+del+dup	Opitz GBBB syndrome, X-linked (Opitz syndrome (OS))
MLH1+del	Cafe-au-lait spots with glioma or leukemia
	Colorectal cancer, hereditary nonpolyposis, type 2 (HNPCC2)
	Endometrial cancer (ENDMC), susceptibility to
	Lobular carcinoma in situ (LCIS)
	Mismatch repair cancer syndrome (MMRCS) (Turcot syndrome) (Brain tumor-polyposis syndrome 1
	Muir-Torre syndrome (MRTES)
MLH3	Colon cancer, hereditary nonpolyposis, type 7 (HNPCC7)
	Colorectal cancer, somatic (CRC)
	Endometrial cancer
MLP (=C-MLP)	Myelofibrosis with myeloid metaplasia, somatic (MMM)
	Thrombocythemia 2 (THCYT2)
	Thrombocytopenia, congenital amegakaryocytic (CAMT)
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type

MMP2	Multicentric osteolysis, nodulosis, and arthropathy (MONA) (Nodulosis-arthropathy-osteolysis syndr
MMP9	Lumbar disc herniation (LDH), susceptibility to
	Metaphyseal anadysplasia type 2 (MANDP2)
MMP13	Metaphyseal anadysplasia type 1 (MANDP1)
	Spondyloepimetaphyseal dysplasia, Missouri type (SEMD-MO)
MMP14	Torg-Winchester syndrome (Nodulosis-arthropathy-osteolysis (NAO) syndrome)
	Winchester syndrome (WNCHRS)
MOPD2 (offiz. PCNT)+del	Microcephalic osteodysplastic primordial dwarfism, type II (MOPD2) (Osteodysplastic primordial dw type II)
	Seckel syndrome
MSH2+del	Cafe-au-lait spots, multiple, with leukemia (DD zu MMRCS)
	Colorectal cancer, hereditary nonpolyposis, type 1 (HNPCC1) (Lynch syndome 1)
	Colorectal cancer, hereditary nonpolyposis, type 8 (HNPCC8)
	Endometrial cancer (ENDMC), susceptibility to
	Mismatch repair cancer syndrome (MMRCS) (Turcot syndrome (Brain tumor-polyposis syndrome 1)
	Muir-Torre syndrome (MRTES)
	Neurofibromatosis, type I, with leukemia (DD zu MMRCS)
MSH6+del	Colorectal cancer, hereditary nonpolyposis, type 5 (HNPCC5)
	Endometrial cancer, familial (ENDMC)
	Mismatch repair cancer syndrome (MMRCS) (Turcot syndrome (Brain tumor-polyposis syndrome 1)
	Ovarian cancer, endometrioid type
MSX2+del	Craniosynostosis, type 2 (CRS2) (Craniosynostosis Boston-type (CSB))
	Parietal foramina 1 (PFM1) (Cranium bifidum occultum)
	Parietal foramina with cleidocranial dysplasia (PFMCCD) (Cleidocranial dysplasia with parietal forar
MYH3	Arthrogryposis, distal, type 2A (DA2A) (Freeman-Sheldon syndrome (FSS))
	Arthrogryposis, distal, type 2B (DA2B) (Sheldon-Hall syndrome (SHS)))Arthrogryposis multiplex co distal, type 2B (AMCD2B))
MYH11+dup	Aortic aneurysm, familial thoracic 4 (AAT4)
MYLK	Aortic aneurysm, familial thoracic 7 (AAT7)
MYO6	Deafness, autosomal dominant 22 (DFNA22)
	Deafness, autosomal dominant 22 (DFNA22), with hypertrophic cardiomyopathy
	Deafness, autosomal recessive 37 (DFNB37)

NALP3 (offiz. NLRP3)	CINCA (chronic infantile neurologic cutaneous and articular) syndrome
	Cold-induced autoinflammatory syndrome, familial (FCAS) (Cold urticaria, familial (FCU))
	Muckle-Wells syndrome (MWS) (Urticaria-deafness-amyloidosis (UDA) syndrome)
NALP12 (offiz. NLRP12)	Cold autoinflammatory syndrome type 2, familial (FCAS2)
NELF	Hypogonadotropic hypogonadism
	Kallmann syndrome
NEMO (offiz. IKBKG)+del	Atypical mycobacteriosis, familial (AMCBX1)
	Ectodermal dysplasia, anhidrotic, with immune deficiency (EDA-ID)
	Ectodermal dysplasia, hypohidrotic, with immune deficiency (HED-ID)
	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency
	Immunodeficiency, isolated
	Incontinentia pigmenti, type II, formerly (IP2, formerly)
	Invasive pneumococcal disease, recurrent isolated, 2 (IPD2)
NF1+del	Leukemia, juvenile myelomonocytic (JMML)
	Melanoma, desmoplastic neurotrophic (DNM)
	Neurofibromatosis, familial spinal (FSNF)
	Neurofibromatosis, type 1 (Von recklinghausen disease)
	Neurofibromatosis-Noonan syndrome (NFNS)
	Watson syndrome (= Pulmonic stenosis with cafe-au-lait spots)
NFIX	Marshall-Smith syndrome (MSS)
	Sotos syndrome 2
NHP2 (=NOLA2)	Dyskeratosis congenita, autosomal recessive 2 (DKCB2)
NIPBL+del	Cornelia de Lange syndrome type 1 (CDLS1)
NKX2-5	Atrial septal defect 7, with or without AV conduction defects (ASD7)
	Conotruncal heart malformations, variable (CTHM)
	Hypoplastic left heart syndrome 2 (HLHS2)
	Hypothyroidism, congenital nongoitrous, 5 (CHNG5)
	Tetrology of Fallot (TOF)
	Truncus arteriosus communis (Persistent truncus arteriosus (PTA))
	Ventricular septal defect 3 (VSD3)
NLRP3 (=NALP3)	CINCA (chronic infantile neurologic cutaneous and articular) syndrome (CINCA)
	Cold-induced autoinflammatory syndrome, familial (FCAS) (Cold urticaria, familial (FCU))
	Muckle-Wells syndrome (MWS) (Urticaria-deafness-amyloidosis (UDA) syndrome)
NLRP12 (=NALP12)	Cold autoinflammatory syndrome type 2, familial (FCAS2)

NOG	Brachydactyly, type B2 (BDB2)
	Multiple synostosis syndrome 1 (SYNS1) (Symphalangism-brachydactyly syndrome)
	Stapes ankylosis with broad thumb and toes (SABTS) (Teunissen-Cremers syndrome)
	Symphalangism, proximal (SYM1) (Cushing symphalangism)
	Synostosis of talus and calcaneus with short stature
	Tarsal-carpal coalition syndrome (TCC)
NOLA2 (offiz. NHP2)	Dyskeratosis congenita, autosomal recessive 2 (DKCB2)
NOLA3 (offiz. NOP10)	Dyskeratosis congenita, autosomal recessive 1 (DKCB1)
NOP10 (=NOLA3)	Dyskeratosis congenita, autosomal recessive 1 (DKCB1)
NOTCH1+del	Aortic valve disease 1 (AOVD1)
	Leukemia, T-cell acute lymphoblastic
NPR2	Acromesomelic dysplasia, Maroteaux type (AMDM)
	St. Helena dysplasia
NPR3	Hypertension, salt-resistant (?)
NR5A1	46,XY sex reversal 3 (SRXY3)
	Adrenocortical insufficiency
	Hypogonadotropic hypogonadism with or without anosmia (HH)
	Premature ovarian failure 7 (POF7)
	Spermatogenic failure 8 (SPGF8)
NRXN1+del	Pitt-Hopkins-like syndrome 2 (PTHSL2)
	Chromosome 2p16.3 deletion syndrome
	Schizophrenia, susceptibility to, 17
NSD1 (=SOTOS)+del	Beckwith-Wiedemann syndrome (BWS)
	Leukemia, acute myeloid (AML)
	Sotos syndrome (SOTOSS) (Cerebral gigantism)
NUSAP1 (=LNP)	spindle microtubule organization, role in
OBSL1	3M syndrome 2 (3M2)
OPG (offiz. TNFRSF11B)	Paget disease, juvenile (JPD) (Hyperphosphatasia, hereditary)
OTX2+del	Microphthalmia, syndromic 5 (MCOPS5)
	Pituitary hormone deficiency, combined, 6 (CPHD6)
	Retinal dystrophy, early-onset, and pituitary dysfunction

p63 (offiz. TP63)	ADULT (Acro-dermato-ungual-lacrimal-tooth) syndrome
	Cervical cancer
	Colon cancer
	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 (EEC3)
	Hay-Wells syndrome (ankyloblepharon-ectodermal defects-cleft lip/palate (AEC) syndrome)
	Head cancer
	Limb-mammary syndrome (LMS)
	Lung cancer
	Neck cancer
	Orofacial cleft 8 (OFC8)
	Ovarian cancer
	Rapp-Hodgkin syndrome (RHS) (= Ectodermal dysplasia Rapp-Hodgkin type (EDRH))
	Split-hand/foot malformation 4 (SHFM4)
PACSIN3	vesicle formation and transport, role in
PAH+del	Hyperphenylalaninemia, mild (HPA, mild)
	Hyperphenylalaninemia, non-PKU mild (HPA, non-PKU mild)
	Phenylketonuria (PKU) (PAH deficiency)
PAPSS2	Brachyolmia, autosomal recessive
	Spondylodysplasia and premature pubarche
	Spondyloepimetaphyseal dysplasia, Pakistani type (SEMD, PA)
PAX2+del	Optic nerve coloboma with renal disease (Renal-coloboma syndrome)
	Renal hypoplasia, isolated
PAX3+del	Craniofacial-deafness-hand syndrome (CDHS)
	Rhabdomyosarcoma 2 (RMS2) (Rhabdomyosarcoma, alveolar (RMSA))
	Waardenburg syndrome, type 1 (WS1)
	Waardenburg syndrome, type 3 (WS3)

PAX6+del	Aniridia (AN) (Aniridia II (AN2), formerly)
	Cataract with late-onset corneal dystrophy
	Coloboma of optic nerve (COLON)
	Coloboma, ocular (COLO)
	Ectopia pupillae
	Foveal hypoplasia and presenile cataract syndrome (O'Donnell-Papas syndrome)
	Foveal hypoplasia with anterior segment anomalies
	Foveal hypoplasia, isolated
	Gillespie syndrome (GS) (Aniridia, cerebellar ataxia, and mental deficiency (ACAMD))
	Keratitis, hereditary (KERH)
	Morning glory disc anomaly
	Optic nerve aplasia, bilateral (BONA)
	Optic nerve head pits, bilateral, congenital
	Optic nerve hypoplasia, bilateral (BONH)
	Peters anomaly (PAN)
	WAGR (Wilms tumor, aniridia, genitourinary anomalies , and mental retardation) syndrome (chromo 11p13 deletion syndrome)
	WAGRO (Wilms tumor, aniridia, genitourinary anomalies, mental retardation, and obesity) syndrome (chromosome 11p13-p12 deletion syndrome)
PC	Pyruvate carboxylase (PC) deficiency
PCNT (=MOPD2)+del	Microcephalic osteodysplastic primordial dwarfism, type II (MOPD2)
	Seckel syndrome
PDE4D	Acrodysostosis type 2, with or without hormone resistance (ACRDYS2)
PEX7+del	Refsum disease, adult (RDA) (Motor and sensory neuropathy, hereditary, 4 (HMSN4))
	Rhizomelic chondrodysplasia punctata, type 1 (RCDP1) (Chondrodysplasia punctata, rhizomelic for
PGN (offiz. SPG7)+del	Spastic paraplegia 7, autosomal recessive (SPG7)
PHEX+del+dup	Hypophosphatemic rickets, X-linked dominant (XLHR) (Vitamin D-resistant rickets, X-linked)
PIN1	Hypogonadotropic Hypogonadism without anosmia
PITX2+del	Axenfeld-Rieger syndrome, type 1 (RIEG1) (Rieger syndrome type 1)
	Iridogoniodysgenesis, type 2 (IRID2)
	Peters anomaly (PAN)
	Ring dermoid of cornea (RDC)
PLCG2 (nur del)	Familial cold autoinflammatory syndrome 3 (FCAS3)
PLOD1+del+dup	Ehlers-Danlos syndrome, type VI (EDS6) (EDS, kyphoscoliotic type)
PLOD2	Bruck syndrome 2 (BRKS2) (Osteogenesis imperfecta with congenital joint contractures)

PMS2+del	Colorectal cancer, hereditary nonpolyposis, type 4 (HNPCC4) (Lynch syndrome 4)
	Mismatch repair cancer syndrome (MMRCS) (Turcot syndrome) (Brain tumor-polyposis syndrome 1
POMC	Obesity, early-onset, susceptibility to
	Proopiomelanocortin deficiency (Obesity, adrenal insufficiency, and red hair)
POP1	Anauxetic dysplasia (Spondylometaepiphyseal dysplasia, Menger type)
POR+del	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis (ABS1)
	Cytochrome P450 oxidoreductase deficiency (POR deficiency)
	Disordered steroidogenesis due to cytochrome P450 oxidoreductase deficiency (DISPORD)
PORCN+del	Focal dermal hypoplasia (FODH) (Goltz syndrome; Goltz-Gorlin syndrome)
POU1F1+del	Pituitary hormone deficiency, combined, 1 (CPHD1)
PPIB	Osteogenesis imperfecta, type IX (OI9)
PRKAR1A+del	Acrodysostosis 1, with or without hormone resistance (ACRDYS1)
	Adrenocortical tumor, somatic
	Carney complex, type 1 (CNC1)
	Myxoma, intracardiac (INTMYX)
	Pigmented nodular adrenocortical disease, primary, 1 (PPNAD1) (Cushing syndrome, adrenal, due PPNAD1)
	Thyroid carcinoma, papillary, somatic (PTC)
PRKG1 (=CGKI)	nitric oxide/cGMP signaling pathway, key mediators
	signal transduction processes in diverse cell types, important components
PROK2 (=KAL4)	Hypogonadism, hypogonadotropic
	Kallmann syndrome 4
PROKR2 (=KAL3)	Kallmann syndrome 3
PROP1+del	Pituitary hormone deficiency, combined, 2 (CPHD2) (Panhypopituitarism)
PRSS1	Pancreatitis, hereditary (PCTT)
	Trypsinogen deficiency
PTCH1+del	Basal cell carcinoma, somatic (BCC)
	Basal cell nevus syndrome (BCNS) (Gorlin syndrome; Gorlin-Goltz syndrome)
	Holoprosencephaly-7 (HPE7)
	Medulloblastoma, somatic (MDB)
PTCH2	Basal cell carcinoma, somatic (BCC)
	Macrostomia, isolated (Lateral cleft, isolated)
	Medulloblastoma, somatic (MDB)

PTEN+del	Bannayan-Riley-Ruvalcaba syndrome (BRRS)
	Chromosome 10q23 deletion syndrome
	Cowden disease (CD) (Cowden syndrome (CS)) (Multiple hamartoma syndrome (MHAM))
	Endometrial carcinoma, somatic (ENDMC)
	Glioma susceptibility 2 (GLM2)
	Juvenile polyposis syndrome, infantile
	Lhermitte-Duclos syndrome (LDD) (Cerebelloparenchymal disorder VI (CPD6))
	Macrocephaly/autism syndrome (MCEPHAS)
	Melanoma, malignant, somatic
	Meningioma
	Meningioma
	Oligodendroglioma
	Prostate cancer, somatic (PC)
	Proteus syndrome
	PTEN hamartoma tumor syndrome (PHTS) (includes Cowden syndrome (CS), Bannayan-Riley-Ruv syndrome (BRRS), Proteus syndrome (PS), and Proteus-like syndrome)
	Squamous cell carcinoma, head and neck, somatic (HNSCC)
	Thyroid carcinoma, follicular, somatic (FTC)
	VATERL (vertebral anomalies, anal atresia, congenital cardiac disease, tracheoesophageal fistula, r anomalies, radial dysplasia, and other limb defects) with macrocephaly and ventriculomegaly assoc
PTH1R (= PTHR)	Chondrodysplasia, Blomstrand type (BOCD)
	Eiken syndrome (Bone modeling defect of hands and feet)
	Enchondromatosis, multiple, Ollier type (ENCHOM) (Osteochondromatosis)
	Failure of tooth eruption, primary (PFE)
	Metaphyseal chondrodysplasia, Murk Jansen type
PTPN1	Insulin resistance, susceptibility to
PTPN11+del+dup	Leukemia, juvenile myelomonocytic (JMML)
	Pterygium colli syndrome
	LEOPARD (multiple lentigines, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonic stenosis, abnormal genitalia, retardation of growth, and sensorineural deafness) syndrome
	Metachondromatosis (METCDS)
	Noonan syndrome 1 (NS1)
PTRF	Lipodystrophy, congenital generalized, type 4 (CGL4) (Berardinelli-Seip congenital lipodystrophy, ty with muscular dystrophy)
PYCR1+del	Cutis laxa autosomal recessive type 2B (ARCL2B) (Cutis laxa with progeroid features)
	Cutis laxa autosomal recessive type IIIB (ARCL3B) (De Barsy Syndrome B)

RAB23	Carpenter syndrome (CARPS) (Acrocephalopolysyndactyly type 2 (ACPS2))
RAF1+del+dup	LEOPARD syndrome 2
	Noonan syndrome 5 (NS5)
RAG1+del	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infectior autoimmunity (T-CMVA)
	Combined cellular and humoral immune defects with granulomas (CCHIDG)
	Omenn syndrome (OS) (Severe combined immunodeficiency with hypereosinophilia)
	Severe combined immunodeficiency, B cell-negative (B- SCID)
RAG2+del	Combined cellular and humoral immune defects with granulomas (CCHIDG)
	Omenn syndrome (OS) (Severe combined immunodeficiency with hypereosinophilia)
	Severe combined immunodeficiency, B cell-negative (= B- SCID)
RIT1	Noonan syndrome 8 (NS8)
RMRP	Anauxetic dysplasia (Spondylometaepiphyseal dysplasia, Menger type)
	Cartilage-hair hypoplasia (CHH) (Metaphyseal chondrodysplasia, McKusick type)
	Metaphyseal dysplasia without hypotrichosis (Cartilage-hair hypoplasia-like skeletal dysplasia witho hypotrichosis or immunodeficiency)
RNU4ATAC	Microcephalic osteodysplastic primordial dwarfism, type I (MOPD1) (Taybi-Linder syndrome (TALS)
RPL5	Aase syndrome
	Aase-Smith syndrome II
	Diamond-Blackfan anemia 6
RPL11	Diamond-Blackfan anemia 7
RPL15	Diamond-Blackfan anemia 12
RPL26	Diamond-Blackfan anemia 11
RPL35A	Diamond-Blackfan anemia 5
RPS6KA3 (=RSK2) +del+dup	Coffin-Lowry syndrome (CLS)
	Mental retardation, X-linked 19 (MRX19)
RPS7	Diamond-Blackfan anemia 8
RPS10	Diamond-Blackfan anemia 9
RPS15	Diamond-Blackfan anemia, candidate
RPS17	Diamond-Blackfan anemia 4

RPS19	Aase syndrome
	Aase-Smith syndrome II
	Anemia, congenital erythroid hypoplastic
	Anemia, congenital hypoplastic, of Blackfan and Diamond
	Aregenerative anemia, chronic congenital
	Blackfan-Diamond Syndrome; BDS
	Diamond-Blackfan anemia 1 (DBA1) (DBA)
	Erythrogenesis imperfecta
	Red cell aplasia, pure, hereditary
RPS24	Diamond-blackfan anemia 3
RPS26	Diamond-Blackfan anemia 10
RPSA (= LAMR1)	Asplenia, isolated congenital
RSK2 (offiz. RPS6KA3) +del+dup	Coffin-Lowry syndrome (CLS)
	Mental retardation, X-linked 19 (MRX19)
RTEL1	Dyskeratosis congenita, autosomal dominant 4 (DKCA4)
RTEL1	Dyskeratosis congenita, autosomal recessive 5 (DKCB5)
RUNX1	Leukemia, acute myeloid
RUNX1	Platelet disorder, familial, with associated myeloid malignancy (FPDMM)
RUNX2+del	Cleidocranial dysplasia (CCD) (= Cleidocranial dysostosis (CLCD))
	Dental anomalies, isolated
SALL1+del	Townes-Brocks branchiootorenal-like syndrome
	Townes-Brocks syndrome (TBS) (Renal-ear-anal-radial syndrome)
SBDS+del	Shwachman-Bodian-Diamond syndrome (SBDS) (Pancreatic insufficiency and bone marrow dysfun
SCYL1BP1 (offiz. GORAB)	Geroderma osteodysplasticum (GO) (Walt Disney dwarfism)
SDCBP	Role in linking syndecan-mediated signaling to the cytoskeleton, cytoskeletal-membrane organizatic adhesion, protein trafficking, and the activation of transcription factors; perhaps a role in melanoma differentiation and neurofibromatosis II
SECISBP2+del	Thyroid hormone metabolism, abnormal
SEDLIN (offiz. TRAPPC2)+del	Spondyloepiphyseal dysplasia tarda, X-linked (SEDT)
SEMA3A+del	Hypogonadotropic hypogonadism 16 with or without anosmia (HH16)
SERPINF1	Osteogenesis imperfecta, type VI (OI6)
SERPING1 (=C1NH)+del	Angioedema, hereditary, type I (HAE1) (C1 esterase inhibitor, deficiency of)
	Angioedema, hereditary, type II (HAE2)
	Complement component 4 (C4), partial deficiency of
SERPINH1	Osteogenesis imperfecta, type X (OI10)
	Preterm premature rupture of the membranes, susceptibility to (PPROM)

SETBP1	Leukemia, acute T-cell lymphoblastic
	Schinzel-Giedion midface retraction syndrome (SGS)
SHOX+del	Langer mesomelic dysplasia (LMD) (Dyschondosteosis, homozygous)
	Leri-Weill dyschondrosteosis (LWD) (Dyschondrosteosis (DCO))
	Short stature, idiopathic, X-linked (ISS)
SIX6	Microphthalmia, isolated, with cataract 2 (MCOPCT2)
SKI	Shprintzen-Goldberg craniosynostosis syndrome (SGS) (Craniosynostosis with arachnodyctyly and abdominal hernias)
SLC5A2+del	Renal glucosuria (GLYS1)
SLC6A8+del	Cerebral creatine deficiency syndrome 1 (CCDS1)
SLC16A2 (=MCT8)+del	Allan-Herndon-Dudley syndrome (AHDS) (Monocarboxylate transporter 8 (MCT8) deficiency) (T3 resistance) (Mental retardation, X-linked, with hypotonia)
SLC26A2 (=DTDST)	Achondrogenesis Ib (ACG1B)
	Atelosteogenesis II (AO2)
	De la Chapelle dysplasia (DLCD)
	Diastrophic dysplasia (DTD)
	Diastrophic dysplasia, broad bone-platyspondylic variant (DTDB)
	Epiphyseal dysplasia, multiple, 4 (EDM4)
SLC34A1	Fanconi renotubular syndrome 2 (FRTS2)
	Nephrolithiasis/osteoporosis, hypophosphatemic, 1 (NPHLOP1)
SLC34A3+del	Hypophosphatemic rickets with hypercalciuria (HHRH)
SLC46A1	Folate malabsorption, hereditary (HFM)
SMAD3	Loeys-Dietz syndrome, type 3 (LDS3) (Aneurysms-osteoarthritis syndrome)
SMAD4	Colorectal cancer (CRC)
	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (JPHT)
	Myhre syndrome (MYHRS) (Growth-mental deficiency syndrome of Myhre)
	Pancreatic cancer (PNCA)
	Polyposis, juvenile intestinal (PJI)
SMARCA4	Rhabdoid tumor predisposition syndrome 2 (RTPS2)
SMARCB1 (=INI1)+del	Rhabdoid predisposition syndrome 1 (RPS1)
	Rhabdoid tumor (RDT) (Teratoid tumor, atypical)
	Schwannomatosis (Neurofibromatosis type 3 (NF3))
SMC1A (=SMC1L1)+del+dup	Cornelia de Lange syndrome type 2 (CDLS2)
SMC3	Cornelia de Lange syndrome type 3 (CDLS3)
SNAI2+del	Piebald trait (PBT) (= Piebaldism)
	Waardenburg syndrome, type 2D (WS2D)

SOS1	Fibromatosis, gingival, 1 (GINGF1)
	Noonan syndrome 4 (NS4)
SOST	Craniodiaphyseal dysplasia, autosomal dominant (CDD)
	Sclerosteosis 1 (SOST1) (Hyperostosis, cortical, with syndactyly)
	Van Buchem disease (VBCH) (Hyperostosis corticalis generalis)
SOTOS (offiz. NSD1)+del	Beckwith-Wiedemann syndrome (BWS)
	Leukemia, acute myeloid (AML)
	Sotos syndrome (SOTOSS)
	Weaver syndrome (WES)
SOX2+del	Microphthalmia, syndromic 3 (MCOPS3)
	Optic nerve hypoplasia and abnormalities of the central nervous system
SP7	Osteogenesis imperfecta, type XII (OI12)
SPAST	Spastic paraplegia 4, autosomal dominant
SPG7 (=PGN)+del	Spastic paraplegia 7, autosomal recessive (SPG7)
SPINK5	Atopy
	Netherton syndrome (NETH) (= NS = Netherton disease)
SRCAP	Floating-Harbor syndrome (FLHS)
SRP72	Bone marrow failure, familial (BMFF)
SRY	46,XX sex reversal 1 (SRXX1) (46,XX Gonadal dysgenesis, complete, SRY-positive)
	46,XY sex reversal 1 (SRXY1) (46,XY Gonadal dysgenesis, complete, SRY-related)
	46,XY True hermaphroditism, SRY-related
	46;XX True hermaphroditism, SRY-positive (Ovotesticular disorder of sex development)
	Swyer syndrome (46, XY Gonadal dysgenesis, complete)
STK11	Lung cancer, non-small cell (NSCLC)
	Melanoma, malignant sporadic
	Pancreatic cancer, sporadic
	Peutz-Jeghers syndrome (PJS) (Polyposis, hamartomatous intestinal)
	Testicular tumor, sporadic
SUFU+del	Medulloblastoma, desmoplastic (MDB, desmoplastic)
TAC3	Hypogonadotropic hypogonadism
TACR3	Hypogonadotropic hypogonadism

TBX5+del+dup	Holt-Oram syndrome (HOS) (Heart-hand syndrome) (Atriodigital dysplasia)
TBX6+dup	Spondylocostal dysostosis, autosomal dominant (SCDO)
TBX15	Cousin syndrome (COUSS) (Craniofacial dysmorphism, hypoplasia of scapula and pelvis, and short (Pelviscapular dysplasia)
TBX18	anterior and posterior somite compartments, maintains the separation of
	otic fibrocyte differentiation, role in
	posterior pole of the heart, role in formation of posterior pole of the
	ureter, role in formation of
	vertebral column, role in formation of
TCAB1 (offiz. WRAP53)	Dyskeratosis congenita, autosomal recessive 3
TCOF1+del	Hemifacial microsomia (HFM) (Goldenhar syndrome) (Oculoauriculovertebral dysplasia (OAVD))
	Treacher Collins syndrome 1 (TCS1)
TCTN3	Joubert syndrome 18 (JBTS18)
	Orofaciodigital syndrome IV (OFD4) (Mohr-Majewski syndrome)
TEM8 (offiz. ANTXR1)	Hemangioma, capillary infantile, susceptibility to
TERC	Aplastic anemia, susceptibility to (AA)
	Dyskeratosis congenita, autosomal dominant 1
	Pulmonary fibrosis, idiopathic, susceptibility to (IPF)
TERT	Bone marrow failure, telomere-related, 1
	Coronary artery disease
	Dyskeratosis congenita, autosomal dominant 2
	Dyskeratosis congenita, autosomal recessive 4
	Leukemia, acute myeloid
	Melanoma, cutaneous malignant, 9
	Pulmonary fibrosis, telomere-related, 1
TGFB2+del	Loeys-Dietz syndrome, type 4 (LDS4)
TGFBR1+dup	Loeys-Dietz syndrome, type 1A (LDS1A) (Furlong syndrome)
	Loeys-Dietz syndrome, type 2A (LDS2A) (Aortic aneurysm, familial thoracic 5 (AAT5))
	Multiple self-healing squamous epithelioma, susceptiblity to (MSSE)
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6 (HNPCC6)
	Esophageal cancer, somatic
	Loeys-Dietz syndrome, type 1B (LDS1B)
	Loeys-Dietz syndrome, type 2B (LDS2B) (Aortic aneurysm, familial thoracic 3 (AAT3))
THRB	Thyroid hormone resistance, generalized, autosomal dominant (GRTH = GTHR)
	Thyroid hormone resistance, generalized, autosomal recessive (GRTH) (Refetoff syndrome)
	Thyroid hormone resistance, selective pituitary (PRTH)

TINF2	Dyskeratosis congenita, autosomal dominant, 3 (DKCA3)
TINF2	Revesz syndrome (exudative retinopathy with bone marrow failure (ERBMF))
TMEM38B+del	Osteogenesis imperfecta, autosomal recessive
TNFRSF11B (=OPG)	Paget disease, juvenile (JPD) (Hyperphosphatasia, chronic congenital idiopathic)
TNNI2	Arthrogryposis multiplex congenita, distal, type 2B (AMCD2B)
TNNI3	Cardiomyopathy, dilated, 1FF
	Cardiomyopathy, dilated, 2A
	Cardiomyopathy, familial hypertrophic, 7
	Cardiomyopathy, familial restrictive
TNNT3	Arthyrgryposis, distal, type 2B (DA2B) (= Arthrogryposis multiplex congenita, distal, type 2B (AMCD (Sheldon-Hall syndrome (SHS))
TNSALP (offiz. ALPL)+del	Hypophosphatasia (HOPS), adult, childhood, infantile or perinatal lethal
	Odontohypophosphatasia
TP63 (=p63)	ADULT (Acro-dermato-ungual-lacrimal-tooth) syndrome
	Cervical cancer
	Colon cancer
	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 (EEC3)
	Hay-Wells syndrome (ankyloblepharon-ectodermal defects-cleft lip/palate (AEC) syndrome)
	Head cancer
	Limb-mammary syndrome (LMS)
	Lung cancer
	Neck cancer
	Orofacial cleft 8 (OFC8)
	Ovarian cancer
	Rapp-Hodgkin syndrome (RHS) (Ectodermal dysplasia, anhidrotic, with cleft lip/palate)
	Split-hand/foot malformation 4 (SHFM4)
TPM1	Cardiomyopathy, dilated, 1Y (CMD1Y)
	Cardiomyopathy, familial hypertrophic, 3 (CMH3)
TPM2	Arthrogryposis, distal, type 1A (DA1A) (Arthrogryposis multiplex congenita, distal, type 1 (AMCD1))
	Arthrogryposis, distal, type 2B (DA2B) (Sheldon-Hall syndrome (SHS))
	Nemaline myopathy 4 (NEM4)
TRAPPC2 (= SEDLIN)+del	Spondyloepiphyseal dysplasia tarda, X-linked (SEDT)
TRIP11	Achondrogenesis, type IA (ACG1A) (Achondrogenesis, Houston-Harris type)
	Leukemia, acute myelogenous

TRPS1+del	Trichorhinophalangeal syndrome, type I (TRPS1)
	Trichorhinophalangeal syndrome, type III (TRPS3) (Sugio-Kajii syndrome)
TRPS1del	Trichorhinophalangeal syndrome, type II (TRPS2) (Langer-Giedion syndrome (LGS)) (Chromosome deletion syndrome)
TRPS2+del (offiz. EXT1)	Chondrosarcoma (CHDSA)
	Exostoses, multiple, type 1 (EXT1) (=Osteochondromas, multiple)
	Langer-Giedion syndrome (LGS) (Trichorhinophalangeal syndrome, type II (TRPS2))
TRPV4	Brachyolmia type 3 (BRAC3)
	Hereditary motor and sensory neuropathy, type IIc (HMSN2C)
	Metatropic dysplasia (MTD)
	Parastremmatic dwarfism (PSTD)
	Scapuloperoneal spinal muscular atrophy (SPSMA)
	Spondyloepiphyseal dysplasia (SED), Maroteaux type (SEDM) (Pseudo-Morquio syndrome, type 2)
	Sodium serum level QTL 1 (SSQTL1) (Hyponatremia)
	Spinal muscular atrophy, distal, congenital nonprogressive (DSMAC)
	Spondylometaphyseal dysplasia (SMD), Kozlowski type (SMDK)
TSC1+del	Focal cortical dysplasia of Taylor (FCDT)
	Focal cortical dysplasia of Taylor, type IIA
	Lymphangioleiomyomatosis (LAM) (Lymphangiomatosis)
	Tuberous sclerosis-1 (TSC1)
TSC2+del	Lymphangioleiomyomatosis, somatic
	Tuberous sclerosis-2 (TSC2)
TSPY1 (=TSPY)	Gonadoblastoma (GBY)
TWIST1+del	Craniosynostosis, type 1 (CRS1)
	Saethre-Chotzen syndrome (SCS) (Acrocephalosyndactyly, type III (ACS3))
	Saethre-Chotzen syndrome with eyelid anomalies (Blepharophimosis, epicanthus inversus, and pto (BPE3), formerly)
	Scaphocephaly (Oxycephaly)
UBE3A+del	Angelman syndrome (AS) (happy puppet syndrome, formerly)
USB1 (=C16orf57)	Poikiloderma with neutropenia (PN) (Poikiloderma with neutropenia, Clericuzio-type)
VAX1	Anophthalmia
	Microphthalmia
VEGFA	Microvascular complications of diabetes, susceptibility to, 1 (MVCD1)

VHL+del	Erythrocytosis, familial, 2 (ECYT2) (Polycythemia, Chuvash type)
	Hemangioblastoma, cerebellar, somatic
	Pheochromocytoma
	Renal cell carcinoma, somatic (RCC) (Hypernephroma) (Adrenocarcinoma of kidney)
	von Hippel-Lindau syndrome (VHLS) (von Hippel-Lindau disease (VHLD))
VLCAD (offiz. ACADVL)	Acyl-CoA dehydrogenase very long chain deficiency (ACADVLD)
WDR34	Asphyxiating thoracic dystrophy (Jeune syndrome)
	Short rib-polydactyly syndrome, type III (SRPS3) (Verma-Naumoff syndrome)
WDR35+del	Cranioectodermal dysplasia 2 (CED2) (Sensenbrenner syndrome)
	Short rib-polydactyly syndrome, type V (SRPS5)
WDR60	Short rib-polydactyly syndrome, type VI (SRPS6)
WFS1+del	Diabetes mellitus, noninsulin-dependent (NIDDM), association with
	Deafness, autosomal dominant 6/14/38 (DFNA6/14/38)
	Wolfram syndrome 1 (WFS1) (Diabetes insipidus and mellitus with optic atrophy and deafness (DID
	Wolfram-like syndrome, autosomal dominant (WFSL) (Hearing loss, progressive, with optic atrophy impaired glucose regulation)
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood (PPAC) (Progressive pseudorheumatoid dysplasia (PPD))
WNT1	Osteogenesis imperfecta
WNT4	Mullerian aplasia and hyperandrogenism (MULLAPL) (Mullerian duct failure and hyperandrogenism
	SERKAL syndrome (46,XX sex reversal with dysgenesis of kidneys, adrenals, and lungs (SERKAL)
WNT7A	Fuhrmann syndrome (FUHRS) (Fibular aplasia or hypoplasia femoral bowing and poly- syn- and oligodactyly)
	Ulna and fibula, absence of, with severe limb deficiency (Limb/pelvis-hypoplasia/aplasia syndrome (LPHAS)) (Al-Awadi/Raas-Rothschild syndrome (AARRS)) (Schinzel phocomelia syndrome)
WNT10A	Ectodermal dysplasia, hypohidrotic/anhidrotic
	Odontoonychodermal dysplasia (OODD)
	Schopf-Schulz-Passarge syndrome (SSPS) (Keratosis palmoplantaris with cystic eyelids, hypodonti hypotrichosis)
	Succedaneous teeth, agenesis of
	Tooth agenesis, selective, 4 (STHAG4) (Lateral incisors, pegged or missing)
WNT10B	Split-hand/foot malformation 6 (SHFM6) (Ectrodactyly, autosomal recessive)
WRAP53 (= TCAB1)	Dyskeratosis congenita, autosomal recessive 3

WT1+del	Denys-Drash syndrome (DDS) (Nephropathy, Wilms tumor, and genital anomalities)
	Desmoplastic small round cell tumor (DSRCT)
	Frasier syndrome (FS)
	Meacham syndrome (MEACHS)
	Mesothelioma, somatic (MESOM)
	Nephrotic syndrome, type 4 (NPHS4)
	WAGR (Wilms tumor, aniridia, genitourinary anomalies, and mental retardation) syndrome (Chromo 11p13 deletion syndrome)
	Wilms tumor, type 1 (WT1) (Nephroblastoma)
XPB (offiz. ERCC3)	Ichthyosiform erythroderma with hair abnormality and mental and growth retardation (Tay syndrome
	Trichothiodystrophy, photosensitive (TTDP)
	Xeroderma pigmentosum, group B (XPB)
	Xeroderma pigmentosum/Cockayne syndrome (XPB/CS)
XYLT1	Desbuquois dysplasia Type 2
	Pseudoxanthoma elasticum (PXE) (Gronblad-Strandberg syndrome), modifier of severity of
	Short stature syndrome, autosomal recessive, with intellectual disability
ZEB2+del	Mowat-Wilson syndrome (MWIS)