Krankheit	Gen
3M syndrome	CUL7, OBSL1, CCDC8, FBXW8
46,XX Gonadal dysgenesis, complete, SRY-positive	SRY
46,XX sex reversal 1 (SRXX1)	SRY
46,XX sex reversal with dysgenesis of kidneys, adrenals, and lungs (SERKAL)	WNT4
46,XX sex reversal, SRY-positive	SRY
46,XX True hermaphroditism, SRY-positive	SRY
46,XY Gonadal dysgenesis, complete	SRY
46,XY gonadal dysgenesis, complete or partial, with or without adrenal failure	NR5A1
46,XY Gonadal dysgenesis, complete, SRY-related	SRY
46,XY sex reversal (SRXY)	SRY, NR5A1
46,XY sex reversal, partial or complete, NR5A1-related	NR5A1
46,XY sex reversal, SRY-related	SRY
46,XY True hermaphroditism, SRY-related	SRY
Aarskog-Scott syndrome (AAS)	FGD1+del
Abdominal aortic aneurysm	COL3A1+del
Achondrogenesis IA (ACG1A)	TRIP11
Achondrogenesis Ib (ACG1B)	SLC26A2 (=DTDST)
Achondrogenesis II (ACG2)	COL2A1+del
Achondrogenesis, Houston-Harris type	TRIP11
Achondrogenesis, Langer-Saldino type	COL2A1+del
Achondrogenesis, type 2, formerly	GDF5 (=CDMP1)
Achondroplasia (ACH)	FGFR3
Acid-labile subunit, deficiency of (ALSD)	IGFALS
Acrocallosal syndrome	KIF7
Acrocapitofemoral dysplasia (ACFD)	IHH
Acrocephalopolysyndactyly type 2 (ACPS2)	RAB23
Acrocephalosyndactyly (ACS)	FGFR1+del (=KAL2), FGFR2+del, TWIST1+del
Acrocephaly, skull asymmetry, and mild syndactyly	FGFR2+del, TWIST1+del
Acro-dermato-ungual-lacrimal-tooth (ADULT) syndrome	TP63 (=p63)
Acrodysostosis, with or without hormone resistance (ACRDYS)	PRKAR1A+del, PDE4D
Acrofacial dysostosis, type Weyers	EVC (=EVC1)+del, EVC2+del
Acromesomelic chondrodysplasia with genital anomalies (AMDGA)	BMPR1B
Acromesomelic dwarfism	GDF5 (=CDMP1)
Acromesomelic dysplasia, Grebe type (AMDG)	GDF5 (=CDMP1)
Acromesomelic dysplasia, Hunter-Thompson type (AMDH)	GDF5 (=CDMP1)
Acromesomelic dysplasia, Maroteaux type (AMDM)	NPR2
Acromicric dysplasia (ACMICD)	FBN1+del
Acropachy, hereditary	HPGD
Acyl-CoA dehydrogenase medium chain deficiency (ACADM)	ACADM (=MCAD)+del
Acyl-CoA dehydrogenase very long chain deficiency (ACADVLD)	ACADVL (=VLCAD)
Adenocarcinoma of lung, somatic	BRAF
Adenoma, periampullary, somatic	APC+del+dup
Adenomas, salivary gland pleomorphic (PSA; SGPA)	CTNNB1; LIFR
Adenomatous polyposis coli, familial (FAP)	APC+del+dup
Adrenal hyperplasia, congenital, due to cytochrome P450 oxidoreductase deficiency	POR+del
Adrenocarcinoma of kidney	VHL+del
Adrenocortical insufficiency	NR5A1
Adrenocortical nodular dysplasia, primary	PRKAR1A+del
Adrenocortical tumor, somatic	PRKAR1A+del
ADULT syndrome (= Acro-dermato-ungual-lacrimal-tooth syndrome)	TP63 (=p63)
AEC syndrome	TP63 (=p63)
AEG syndrome	SOX2+del
Aganglionic megacolon (MGC)	EDN3
AGAT deficiency	GATM
Alagille syndrome, type 1 (ALGS1)	JAG1+del+dup
Al-Aqeel Sewairi syndrome	MMP14, MMP2
Al-Awadi/Raas-Rothschild syndrome (AARRS)	WNT7A
Albright hereditary osteodystrophy-like syndrome	HDAC4+del
Allan-Herndon syndrome	SLC16A2 (=MCT8)+del
Allan-Herndon-Dudley syndrome (AHDS)	SLC16A2 (=MCT8)+del
Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity (T-CMVA)	RAG1+del
Alpha-Galactosidase A deficiency	GLA+del
Alpha-thalassemia myelodysplasia syndrome (ATMDS)	ATRX+del+dup
Alpha-thalassemia/mental retardation syndrome, nondeletion type	ATRX+del+dup
Alpha-thalassemia/mental retardation syndrome, X-linked (ATRX)	ATRX+del+dup
Alport syndrome	COL4A5, COL4A3, COL4A4
Alport syndrome and diffuse leiomatosis (ATS-DL)	COL4A5+del
Amyotrophy, neurogenic scapuloperoneal, New England type	TRPV4
Anauxetic dysplasia	RMRP, POP1
Anderson-Fabry disease	GLA+del
Anemia, X-linked, with/without neutropenia and/or platelet abnormalities (XLANP)	GATA1
Aneurysm, abdominal aortic (AAA)	COL3A1+del
Aneurysm, aortic and cerebral, with arterial tortuosity and skeletal manifestations	TGFB2+del
Aneurysms-osteoarthritis syndrome	SMAD3
Angelman syndrome (AS)	UBE3A+del
Angelman syndrome-like	MECP2+del, CDKL5+del+dup
Angioedema, hereditary, type I+II (HAE1+2)	SERPING1 (=C1NH)+del
Angiokeratoma corporis diffusum	GLA+del
Angioneurotic edema, hereditary (HANE)	SERPING1 (=C1NH)+del
Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps (HANAC)	COL4A1, COL4A2
Aniridia (AN)	PAX6+del
Aniridia II (AN2), formerly	PAX6+del
Aniridia, cerebellar ataxia, and mental deficiency (ACAMD)	PAX6+del
Aniridia, cerebellar ataxia, and mental retardation (ACAMR)	PAX6+del
Ankyloblepharon-ectodermal defects-cleft lip/palate (AEC)	TP63 (=p63)

Anophthalmia	VAX1
Anophthalmia, clinical, with associated anomalies	SOX2+del
Anophthalmia-esophageal-genital (AEG) syndrome	SOX2+del
Antibody deficiency and immune dysregulation, PLACG2-associated (PLAID)	PLCG2del
Antley-Bixler syndrome (ABS)	POR+del, FGFR2+del
Antoplo disease	LAMP2+del
Anus, imperforate, with hand, foot, and ear anomalies	SALL1+del
Aortic aneurysm and dissection, familial thoracic (TAAD)	MYH11+dup
Aortic aneurysm, ascending, and dissection	FBN1+del
Aortic aneurysm, familial abdominal 1 (AAA1)	COL3A1+del, TGFBR2, MYH11+dup, TGFBR1+dup, ACTA2, MYLK
Aortic dissection, familial, with or without aortic aneurysm	MYLK
Aortic stenosis, calcific	NOTCH1+del
Aortic valve disease	NOTCH1+del
Aortic valve disease 1 (AOVD1)	NOTCH1+del
Aortic valve, bicuspid	NOTCH1+del
Aortic valve, calcification of	NOTCH1+del
Apert syndrome (APRS)	FGFR2+del
Aplasia of lacrimal and salivary glands (ALSG)	FGF10+del
Aplastic anemia, susceptibility to (AA)	TERC+del, TERT+del
APOC2 deficiency	APOC2+del
Apolipoprotein C-II deficiency	APOC2+del
Arginine:glycine amidinotransferase deficiency	GATM
Arthrochalasis multiplex congenita	COL1A2+del
Arthrogryposis multiplex congenita, distal, type 1 (AMCD1)	TPM2
Arthrogryposis multiplex congenita, distal, type 2B (AMCD2B)	TNNT3, TNNI2, MYH3, TPM2
Arthrogryposis multiplex congenita, distal, type II, with craniofacial abnormalities	TNNT3, TNNI2, TPM2
Arthrogryposis, distal, type 1 (DA1)	TPM2
Arthrogryposis, distal, type 1A (DA1A)	TPM2
Arthrogryposis, distal, type 2A (DA2A)	MYH3
Arthrogryposis, distal, type 2B (DA2B)	TNNT3, TNNI2, MYH3, TPM2
Arthrogryposis, distal, type 9 (DA9)	FBN2
Arthroophthalmopathy, hereditary progressive (AOM)	COL2A1+del
Arthropathy, progressive pseudorheumatoid, of childhood (PPAC)	WISP3
Asphyxiating thoracic dystrophy	WDR34, IFT80, DYNC2H1
Asplenia, familial	RPSA (= LAMR1)
Asplenia, isolated congenital (ICAS)	RPSA (= LAMR1)
Ataxia with lactic acidosis II	PC
Ateliotic dwarfism with hypogonadism	PROP1+del
Atelosteogenesis (AO)	FLNB, SLC26A2 (=DTDST)
Athabaskan brainstem dysgenesis syndrome (ABSD)	HOXA1
Atopy	SPINK5
ATR, nondeletion type	ATRX+del+dup
Atrial myxoma, familial	PRKAR1A+del
Atrial septal defect (ASD)	GATA4+del, NKX2-5
Atriodigital dysplasia	TBX5+del+dup
Atrioventricular septal defect (AVSD)	GATA4+del, CRELD
ATR-X syndrome	ATRX+del+dup
Atypical mycobacterial infection, disseminated, X-linked type 1	IKBKG (=NEMO)+del
Atypical mycobacteriosis, familial (AMCBX1)	IKBKG (=NEMO)+del
Autism susceptibility, X-linked 3 (AUTSX3)	MECP2+del
Autonomic control, congenital failure of	EDN3
Avascular necrosis of the femoral head (ANFH)	COL2A1+del
Axenfeld-Rieger syndrome (ARS)	PITX2+del
Bannayan-Riley-Ruvalcaba syndrome (BRRS)	PTEN+del
Bannayan-Zonana syndrome (BZS)	PTEN+del
Baraitser-Burn syndrome	TCTN3
Barraquer-Simons syndrome	LMNB2
Bart-Pumphrey syndrome (BPS)	GJB2 (=CX26)
Basal cell carcinoma, somatic (BCC)	PTCH1+del, PTCH2
Basal cell nevus syndrome (BCNS)	PTCH1+del
Beals syndrome	FBN2
Beare-Stevenson cutis gyrata syndrome (BSTVS)	FGFR2+del
Beckwith-Wiedemann syndrome (BWS)	CDKN1C+dup; NSD1 (=SOTOS)+del
Benign hypermobility syndrome	COL3A1+del
Berardinelli syndrome	BSCL2+del
Berardinelli-Seip congenital lipodystrophy (BSCL)	AGPAT2+del, BSCL2+del, CAV1, PTRF
Beta-Galactosidase-1 (GLB1) deficiency	GLB1+del+dup
Bicuspid aortic valve (BAV)	NOTCH1+del
Bile acid synthesis defect, congenital, 3 (CBAS3)	CYP7B1
Biodefective growth hormone	GH1+del+dup
Blackfan-Diamond Syndrome; BDS	Phase 1: Sequenzierung RPS19, RPL5, RPL11, RPS26, RPL35a Phase 2: Deletionsanalyse der RP-Gene mittels Array-CGH Phase 3: Sequenzierung RPS10, RPS24, RPS7, RPL15, RPL26, RPL15, GATA1
Bladder cancer, somatic (BLC)	KRAS, FGFR3, HRAS
Blepharophimosis, epicanthus inversus, and ptosis	FOXL2+del+dup, TWIST1+del
Blepharophimosis-mental retardation syndrome, Maat-Kievit-Brunner type	MED12
Bohring-Opitz syndrome	CD96
Bone marrow failure, familial (BMFF)	SRP72
Bone marrow failure, telomere-related, 1	TERT
Bone modeling defect of hands and feet	PTH1R (= PTHR)
Boomerang dysplasia	FLNB
Bosley-Salih-Alorainy syndrome (BSAS)	HOXA1
Brachydactyly, Haws type	GDF5 (=CDMP1)
Brachydactyly, type A1 (BDA1)	IHH
Brachydactyly, type A2 (BDA2)	GDF5 (=CDMP1), BMP2+del+dup, BMPR1B
Brachydactyly, type B2 (BDB2)	NOG
Brachydactyly, type C (BDC)	GDF5 (=CDMP1)
Brachydactyly, type D (BDD)	HOXD13+del
Brachydactyly, type E (BDE)	HOXD13+del

Brachydactyly, type E1 (BDE1)	HOXD13+del
Brachydactyly-mental retardation syndrome (BDMR)	HDAC4+del
Brachydactyly-syndactyly syndrome (BDSD)	HOXD13+del, MBOAT1+del
Brachymelic primordial dwarfism	RNU4ATAC
Brachymesophalangy II	GDF5 (=CDMP1), BMP2+del+dup, BMPR1B
Brachyolmia, autosomal dominant	TRPV4
Brachyolmia, autosomal recessive	PAPSS2
Brachyrachia	TRPV4
Brain small vessel disease with Axenfeld-Rieger anomaly	COL4A1, COL4A2
Brain small vessel disease with hemorrhage (BSVDH)	COL4A1, COL4A2
Brain tumor, posterior fossa, of infancy, familial)	SMARCB1 (=INI1)+del
Brain tumor-polyposis syndrome 1 (BTPS1	MLH1+del, MSH2+del, MSH6+del, PMS2+del
Brain tumor-polyposis syndrome 2	APC+del+dup
Breast cancer, somatic (BC)	KRAS, AKT1
Bruck syndrome 2 (BRKS2)	PLOD2
Brunzell syndrome	AGPAT2+del, BSCL2+del
Budd-Chiari syndrome, susceptibility to (BDCHS)	JAK2
Bulldog syndrome	GPC3+del, GPC4del+dup
C syndrome (CSYN)	CD96
C1 esterase inhibitor, deficiency of	SERPING1 (=C1NH)+del
C8 beta deficiency	C8B
C8 deficiency, type II	C8B
C8B deficiency	C8B
Cafe-au-lait spots with glioma or leukemia	MLH1+del
Cafe-au-lait spots with pulmonic stenosis	NF1+del
Cafe-au-lait spots, multiple, with leukemia (DD zu MMRCS)	MSH2+del
Caffey disease (CAFFD)	COL1A1+del
Calcinosis, tumoral, with hyperphasphatemia	FGF23
Camptodactyly tall stature and hearing loss (CATSHL) syndrome	FGFR3
Cardiofaciocutaneous syndrome (CFCS)	BRAF, KRAS, MAP2K1 (=MEK1), MAP2K2 (=MEK2)
Cardiogenital syndrome	LMNA+del
Cardiomyopathy, congestive, with hypergonadotropic hypogonadism	LMNA+del
Cardiomyopathy, dilated	LMNA+del, TNNI3, TPM1
Cardiomyopathy, dilated, with hypergonadotropic hypogonadism	LMNA+del
Cardiomyopathy, dilated, with premature ovarian failure	LMNA+del
Cardiomyopathy, dilated, with primary testicular failure	LMNA+del
Cardiomyopathy, familial hypertrophic	TPM1, TNNI3
Carney complex, type 1 (CNC1)	PRKAR1A+del
Carney myxoma-endocrine complex	PRKAR1A+del
Carney syndrome (CAR)	PRKAR1A+del
Carnitine palmitoyltransferase 2 deficiency, infantile (CPT2DI)	CPT2
Carnitine palmitoyltransferase 2 deficiency, late-onset (CPT2D)	CPT2
Carnitine palmitoyltransferase 2 deficiency, lethal neonatal (CPT2D-LN)	CPT2
Carpenter syndrome (CARPS)	RAB23
Carpenter-Waziri syndrome (CWS)	ATRX+del+dup
Cartilage-hair hypoplasia (CHH)	RMRP
Cartilage-hair hypoplasia variant, skeletal manifestations only (CHHV)	RMRP
Cartilage-hair hypoplasia-like skeletal dysplasia without hypotrichosis or	RMRP
Cataract with late-onset corneal dystrophy	PAX6+del
Catlin marks	MSX2+del
CATSHL syndrome (= camptodactyly tall stature and hearing loss syndrome)	FGFR3
CBL syndrome	CBL
Cchondrodystrophy with sensineural deafness	COL2A1+del, COL11A2
Central hypoventilation syndrome, congenital (CCHS)	EDN3
Cephaloskeletal dysplasia	RNU4ATAC
Cerebelloparenchymal disorder VI (CPD6)	PTEN+del
Cerebral creatine deficiency syndrome (CCDS)	SLC6A8+del, GAMT, GATM
Cerebral gigantism	NSD1 (=SOTOS)+del, NFIX
Cerebroretinal microangiopathy with calcifications and cysts (CRMCC)	CTC1
Cervical cancer	TP63 (=p63)
Cervical cancer, somatic (CERCA)	FGFR3
CFC syndrome	BRAF, KRAS, MAP2K1 (=MEK1), MAP2K2 (=MEK2)
Charcot-Marie-Tooth disease	LMNA+del, TRPV4
Charcot-Marie-Tooth neuropathy	LMNA+del, TRPV4
CHARGE syndrome	CHD7 (=KAL5)+del
Chondrodysplasia punctata, rhizomelic form (CDPR)	PEX7+del
Chondrodysplasia punctata, X-linked dominant, 2 (CDPX2)	EBP (=CPX)
Chondrodysplasia with joint dislocations, GPAPP type (CDP-GPAPP)	IMPAD1
Chondrodysplasia with multiple dislocations, CDMD	CHST3 (=C6ST1)
Chondrodysplasia, Blomstrand type (BOCD)	PTH1R (= PTHR)
Chondrodysplasia, Grebe type	GDF5 (=CDMP1)
Chondrodystrophia calcificans punctata	PEX7+del
Chondroectodermal dysplasia	EVC (=EVC1)+del, EVC2+del
Chondrogenesis imperfecta	COL2A1+del
Chondrosarcoma (CHDSA)	EXT1+del (=TRPS2), EXT2+del
Chotzen syndrome	FGFR2+del, TWIST1+del
Chromosome 10q23 deletion syndrome	BMPR1A+del, PTEN+del
Chromosome 11p11.2 deletion Syndrome	EXT2+del, ALX4
Chromosome 11p13 deletion syndrome	PAX6del, WT1del
Chromosome 11p13-p12 deletion syndrome	PAX6del, WT1del
Chromosome 16p13.3 deletion syndrome	CREBBP+del
Chromosome 2p16.3 deletion syndrome	NRXN1del
Chromosome 2q37.2 deletion syndrome	HDAC4+del
Chromosome 5q14.3 deletion syndrome	MEF2C del
Chromosome 8q24.1 deletion syndrome	TRPS1del
Chromosome Xq22.3 centromeric deletion syndrome	COL4A5del
Chronic infantile neurologic cutaneous and articular syndrome (CINCA)	NLRP3 (=NALP3)
Chronic neurologic cutaneous and articular syndrome	NLRP3 (=NALP3)
Chronic recurrent multifocal osteomyelitis (CRMO), congenital dyserythropoietic	LPIN2

Chudley-Lowry syndrome	ATRX+del+dup
Chylomicronemia, familial	LPL+del+dup
C-II Anapolipoproteinemia	APOC2+del
CINCA syndrome	NLRP3 (=NALP3)
Cleft lip and/or palate with mucous cysts of lower lip	IRF6+del
Cleft lip with or without cleft palate, nonsyndromic (CL/P)	BMP4, IRF6+del, TP63 (=p63)
Cleft lip/palate, paramedian mucous cysts of the lower lip, popliteal pterygium, digital and geneital anomalies faciogenitopopliteal syndrome	IRF6+del
Cleidocranial dysostosis (CLCD)	RUNX2+del
Cleidocranial dysplasia (CCD)	RUNX2+del
Cleidocranial dysplasia with parietal foramina	MSX2+del
C-like syndrome (CLSYN)	CD96
Clubbing of digits	HPGD
Coats plus syndrome	CTC1
Coffin-Lowry syndrome (CLS)	RPS6KA3 (=RSK2) +del+dup
Cold autoinflammatory syndrome, familial (FCAS)	NLRP3 (=NALP3), NLRP12 (=NALP12)
Cold urticaria, familial (FCU)	NLRP3 (=NALP3)
Cold-induced autoinflammatory syndrome, familial (FCAS)	NLRP3 (=NALP3)
Coloboma of iris, choroid, and retina (COI) = Coloboma, uveoretinal)	PAX6+del
Coloboma of optic nerve (COLON)	PAX6+del
Coloboma of optic nerve with renal disease	PAX2+del
Coloboma, ocular (COLO)	PAX6+del
Coloboma, uveoretinal	PAX6+del
Colorectal cancer (CRC)	CTNNB1, TP63 (=p63), EP300+del, SMAD4
Colorectal cancer, hereditary nonpolyposis (HNPCC)	MSH2+del, MLH1+del, PMS2+del, MSH6+del, TGFBR2+del, MLH3
Colorectal cancer, somatic (CRC)	MLH3, AKT1, APC, BRAF
Combined cellular and humoral immune defects with granulomas (CCHIDG)	RAG1+del, RAG2+del
Combined immunodeficiency with susceptibility to mycobacterial, viral, and fungal infections	GATA2
Commissural cleft, isolated	PTCH2
Complement component 4 (C4), partial deficiency of	SERPING1 (=C1NH)+del
Complement component 8 deficiency, type II (C8D2)	C8B
Complement component 8B deficiency	C8B
Complex I deficiency, mitochondrial	ECSIT
Congenital 'healed' cleft lip (CHCL)	BMP4
Congenital hypothalamic hamartoma syndrome (CHHS)	GLI3+del+dup
Congenital mydriasis	ACTA2
Conotruncal anomaly face syndrome (CAFS)	NKX2-5
Conotruncal heart malformations, variable (CTHM)	NKX2-5
Conradi-Hunermann-Happle syndrome	EBP (=CPX)
Contractural arachnodactyly, congenital (CCA)	FBN2
Copper transport disease	ATP7A+del+dup
Cornelia de Lange syndrome (CDLS)	NIPBL+del, SMC1A (=SMC1L1)+del+dup , SMC3
Coronary artery disease	TERT
Coronary artery disease, susceptiblity to (CAD)	TERT+del
Corpus callosum, agenesis of (ACC), with abnormal genitalia	ARX+del+dup
Cortical dysplasia of Taylor (CDT)	TSC1+del
Cortical dysplasia of Taylor with balloon cells (CDTBC)	TSC1+del
Cortical dysplasia of Taylor without balloon cells	TSC1+del
Cortical dysplasia of Taylor, dysplasia only (CDTD)	TSC1+del
Cortical hyperostosis with hyperphosphatemia	FGF23
Costello syndrome	HRAS
Cousin syndrome (COUSS)	TBX15
Cowden disease (CD)	PTEN+del
Cowden syndrome (CS)	PTEN+del
CPT deficiency, hepatic, type II	CPT2
CPT II deficiency, lethal neonatal	CPT2
CPT II deficiency, myopathic	CPT2
Craniodiaphyseal dysplasia, autosomal dominant (CDD)	SOST
Cranioectodermal dysplasia (CED)	IFT122, WDR35+del, IFT43
Craniofacial dysmorphism, hypoplasia of scapula and pelvis, and short stature	TBX15
Craniofacial dysostosis type I (CFD1)	FGFR2+del
Craniofacial-deafness-hand syndrome (CDHS)	PAX3+del
Craniofacial-skeletal-dermatologic dysplasia	FGFR2+del
Craniofrontonasal dysostosis	EFNB1+del+dup
Craniofrontonasal dysplasia (CFND)	EFNB1+del+dup
Craniofrontonasal syndrome (CFNS)	EFNB1+del+dup
Craniomandibular dermatodysostosis	LMNA+del
Craniooosteoarthropathy (COA)	HPGD
Craniostenosis	FGFR2+del, TWIST1+del, MSX2+del
Craniosynostosis (CRS)	FGFR2+del, TWIST1+del, MSX2+del
Craniosynostosis with arachnodyctyly and abdominal hernias	SKI
Craniosynostosis, metopic	FGFR1+del (=KAL2)
Craniosynostosis, midfacial hypoplasia, and foot anomalies	FGFR1+del (=KAL2), FGFR2+del
Cranium bifidum occultum	MSX2+del
Cranium bifidum, hereditary	MSX2+del
Creatine deficiency syndrome due to AGAT deficiency	GATM
Creatine deficiency syndrome due to GAMT deficiency	GAMT
Creatine deficiency syndrome, X-linked	SLC6A8+del
Creatine transporter defect	SLC6A8+del
Crossed polydactyly, type I (CP1)	GLI3+del+dup
Crouzon syndrome (CS)	FGFR2+del
Crouzon syndrome with acanthosis nigricans	FGFR3
Cryopyrin-associated periodic syndrome (CAPS)	NLRP3 (=NALP3)
Currarino idiopathic osteoarthropathy (CIO)	HPGD
Curry-Hall syndrome	EVC (=EVC1)+del, EVC2+del
Cushing symphalangism	GDF5 (=CDMP1), NOG
Cushing syndrome, adrenal, due to PPNAD1	PRKAR1A+del
Cutis laxa with progeroid features	PYCR1+del

Cutis laxa, autosomal dominant, 1 (ADCL1)	ELN+del
Cutis laxa, autosomal recessive (ARCL)	ATP6V0A2+del, PYCR1+del
Cutis laxa, X-linked, formerly	ATP7A+del+dup
Cytochrome P450 oxidoreductase deficiency	POR+del
Czech dysplasia	COL2A1+del
D-2-Hydroxyglutaric aciduria (D2HGA)	D2HGDH+del, IDH2
Danon disease (DAND)	LAMP2+del
Dappled metaphysis syndrome	COL2A1+del
D-bifunctional protein deficiency (DBPD)	HSD17B4+del (= DBP)
De Barsy Syndrome B	PYCR1+del
De la Chapelle dysplasia (DLCD)	SLC26A2 (=DTDST)
Deafness, autosomal dominant (DFNA)	GJB2 (=CX26), WFS1+del, COL11A2, MYO6
Deafness, autosomal dominant 22 (DFNA22), with hypertrophic cardiomyopathy	MYO6
Deafness, autosomal recessive	GJB2 (=CX26), MYO6, COL11A2
Deafness, congenital heart defects, and posterior embryotoxon	JAG1+del+dup
Deafness, sensorineural, with imperforate anus and thumb anomalies	SALL1+del
Defect11 syndrome	EXT2+del, ALX4
Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency (DCML)	GATA2
Dental anomalies, isolated	RUNX2+del
Dental noneruption	PTH1R (= PTHR)
Denys-Drash syndrome (DDS)	WT1+del
Dermatopathia pigmentosa reticularis (DPR)	KRT14
Desbuquois dysplasia (DBQD)	CANT1+del, XYLT1
Desmoid disease, hereditary (HDD)	APC+del+dup
Desmoplastic small round cell tumor (DSRCT)	WT1+del
Developmental delay, epilepsy, and neonatal diabetes (DEND)	KCNJ11
Diabetes insipidus and mellitus with optic atrophy and deafness (DIDMOAD)	WFS1+del
Diabetes mellitus, noninsulin-dependent (NIDDM)	ABCC8+del
Diabetes mellitus, noninsulin-dependent (NIDDM), association with	WFS1
Diabetes mellitus, permanent neonatal (PNDM)	ABCC8+del, KCNJ11
Diabetes mellitus, permanent neonatal, with neurologic features	KCNJ11
Diabetes mellitus, permanent, of infancy (PDMI)	ABCC8+del, KCNJ11
Diabetes mellitus, transient neonatal	ABCC8+del, KCNJ11
Diamond-Blackfan Anemia (DBA)	Phase 1: Sequenzierung RPS19, RPL5, RPL11, RPS26, RPL35a Phase 2: Deletionsanalyse der RP-Gene mittels Array-CGH Phase 3: Sequenzierung RPS10, RPS24, RPS7, RPL15, RPL26, RPL15, GATA1
Diastrophic dysplasia (DTD)	SLC26A2 (=DTDST)
Diastrophic dysplasia, broad bone-platyspondylic variant (DTDB)	SLC26A2 (=DTDST)
Digital clubbing, isolated congenital	HPGD
Dihydrolipoamide dehydrogenase deficiency (DLDD)	DLD (=LAD)
Disorder of sex development, 46,XY	NR5A1
Disordered steroidogenesis due to cytochrome P450 oxidoreductase deficiency (DISPORD)	POR+del
Disordered steroidogenesis due to POR deficiency	POR+del
DLD deficiency	DLD (=LAD)
Double-outlet right ventricle (DORV)	NKX2-5
Drash syndrome	WT1+del
Du Pan syndrome (DPS)	GDF5 (=CDMP1)
Dwarfism of Sindh	GHRHR, GH1+del+dup
Dyggve-Melchior-Clausen disease (DMC)	DYM+dup
Dyschondroplasia	IDH1, IDH2, PTH1R (= PTHR)
Dyschondrosteosis (DCO)	SHOX+del
Dyschondrosteosis, homozygous	SHOX+del
Dyskeratosis congenita	DKC1, TRC, TERT, TINF2, RTEL1, NOP10, NHP2, CTC1, C16orf57
Dysplasia gigantism syndrome, X-linked (DGSX)	GPC3+del, GPC4del+dup
Dysplastic gangliocytoma of the cerebellum	PTEN+del
Dyssegmental dysplasia, Silverman-Handmaker type (DDSH)	HSPG2+del
E3 deficiency	DLD (=LAD)
Eccrine tumors with ectodermal dysplasia	WNT10A
Ectodermal dysplasia Rapp-Hodgkin type (EDRH)	TP63 (=p63)
Ectodermal dysplasia, anhidrotic, with cleft lip/palate	TP63 (=p63)
Ectodermal dysplasia, hypohidrotic/anhidrotic	WNT10A
Ectodermal dysplasia, hypohidrotic/anhidrotic, with immune deficiency	IKBKG (=NEMO)+del
Ectopia lentis, familial (EL)	FBN1+del
Ectopia lentis, isolated, autosomal dominant	FBN1+del
Ectopia pupillae	PAX6+del
Ectrodactyly, autosomal recessive	WNT10B
Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 (EEC3)	TP63 (=p63)
EEC syndrome 3	TP63 (=p63)
Ehlers-Danlos syndrome gravis	COL1A1+del
Ehlers-Danlos syndrome type I (EDS1)	COL1A1+del
Ehlers-Danlos syndrome type VII, autosomal dominant	COL1A1+del
Ehlers-Danlos syndrome type VIIA (EDS7A)	COL1A1+del
Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss (EDSKMH)	FKBP14
Ehlers-Danlos syndrome, arterial type	COL3A1+del
Ehlers-Danlos syndrome, arthrochalasia type	COL1A1+del, COL1A2+del
Ehlers-Danlos syndrome, cardiac valvular form (EDSCV)	COL1A2+del
Ehlers-Danlos syndrome, ecchymotic type	COL3A1+del
Ehlers-Danlos syndrome, gravis type	COL5A1+del+dup, COL5A2
Ehlers-Danlos syndrome, hypermobility type	COL3A1+del
Ehlers-Danlos syndrome, kyphoscoliotic type	PLOD1+del+dup
Ehlers-Danlos syndrome, mild classic type	COL5A1+del+dup
Ehlers-Danlos syndrome, mitis type	COL5A1+del+dup
Ehlers-Danlos syndrome, occipital horn type, formerly	ATP7A+del+dup
Ehlers-Danlos syndrome, ocular-scoliotic type	PLOD1+del+dup
Ehlers-Danlos syndrome, progeroid form (EDSP)	B4GALT7, B3GALT6
Ehlers-Danlos syndrome, Sack-Barabas type	COL3A1+del

Ehlers-Danlos syndrome, severe classic type	COL5A1+del+dup, COL5A2
Ehlers-Danlos syndrome, type I (EDS I) (EDS1)	COL5A1+del+dup, COL5A2
Ehlers-Danlos syndrome, type II (EDS II) (EDS2)	COL5A1+del+dup
Ehlers-Danlos syndrome, type III (EDSIII; EDS3)	COL3A1+del
Ehlers-Danlos syndrome, type IV, autosomal dominant (EDS IV; EDS4)	COL3A1+del
Ehlers-Danlos syndrome, type IX (EDS IX) (EDS9), formerly	ATP7A+del+dup
Ehlers-Danlos syndrome, type VI (EDS6) (EDS VI)	PLOD1+del+dup
Ehlers-Danlos syndrome, type VIA, formerly (EDS6A, formerly)	PLOD1+del+dup
Ehlers-Danlos syndrome, type VIIB (EDS7B)	COL1A2+del
Ehlers-Danlos syndrome, vascular type	COL3A1+del
Eiken skeletal dysplasia (EISD)	PTH1R (= PTHR)
Eiken syndrome	PTH1R (= PTHR)
Ellis-van Creveld syndrome (EVC)	EVC (=EVC1)+del, EVC2+del
Emberger syndrome	GATA2
Emery-Dreifuss muscular dystrophy (EDMD) (EMD)	LMNA+del
Encephalopathy, acute, infection-induced, 4, susceptibility to (IIAE4)	CPT2
Encephalopathy, neonatal severe	MECP2+del
Enchondromatosis, multiple, Ollier type (ENCHOM)	IDH1, IDH2, PTH1R (= PTHR)
Endometrial cancer (ENDMC), susceptibility to	MLH1+del, MSH2+del
Endometrial cancer, familial (ENDMC)	MLH3, MSH6+del
Endometrial cancer, somatic (ENDMC)	PTEN+del
Enlarged vestibular aqueduct (EVA)	FOXI1
Epidermal nevus syndrome, formerly	KRAS
Epidermolysis bullosa simplex Dowling-Meara type (EBS-DM)	KRT14
Epidermolysis bullosa simplex Koebner type (EBS-K)	KRT14
Epidermolysis bullosa simplex Weber-Cockayne type (EBS-WC)	KRT14
Epidermolysis bullosa simplex, autosomal recessive (AREBS)	KRT14
Epidermolysis bullosa simplex, generalized (EBS generalized)	KRT14
Epidermolysis bullosa simplex, localized	KRT14
Epileptic encephalopathy, early infantile (EIEE)	ARX+del+dup, CDKL5+del+dup
Epiphyseal dysplasia, Fairbank type (EDMF)	COMP+del
Epiphyseal dysplasia, multiple, autosomal dominant (EDM)	COMP+del, MATN3, COL9A1, COL9A2, COL9A3
Epiphyseal dysplasia, multiple, autosomal recessive (EDM)	SLC26A2 (=DTDST)
Epiphyseal dysplasia, multiple, with myopia and deafness (EDMMD)	COL2A1+del
Epiphyseal dysplasia, Ribbing type (EDMR)	COMP+del
Erythrocytosis, autosomal recessive benign	VHL+del
Erythrocytosis, familial, 2 (ECYT2)	VHL+del
Erythrocytosis, somatic	JAK2
Esophageal cancer, somatic	TGFBR2
ESS1, formerly	TGFBR1+dup
ETFA deficiency	ETFA+del
ETFB deficiency	ETFB
ETFDH deficiency	ETFDH
Ethylmalonic-adipicaciduria (EMA)	ETFA+del, ETFB
Eunuchoidism, familial hypogonadotropic	GNRH1
Ewing sarcoma (ES)	FLI1
Exomphalos-Makroglossia-Gigantism syndrome (EMGS)	CDKN1C+dup; NSD1 (=SOTOS)+del
Exostoses, multiple, type (EXT)	EXT1+del (=TRPS2), EXT2+del
Exudative retinopathy with bone marrow failure (ERBMF)	TINF2
Fabry disease	GLA+del
Fabry disease, cardiac variant	GLA+del
Facioauriculovertebral sequence (FAVS)	TCOF1+del
Faciocutaneoskeletal syndrome (FCSS)	HRAS
Faciogenital dysplasia (FGDY)	FGD1+del
Failure of tooth eruption, primary (PFE)	PTH1R (= PTHR)
Familial atypical cold urticaria (FACU)	PLCG2 (nur del)
Familial cold autoinflammatory syndrome 3 (FCAS3)	PLCG2 (nur del)
Familial combined hyperlipidemia	LPL+del+dup
Familial idiopathic osteoarthropathy of childhood	HPGD
Fanconi renotubular syndrome 2 (FRTS2)	SLC34A1
Female Pseudo-Turner syndrome	PTPN11+dup
Ferguson-Smith type epithelioma	TGFBR1+dup
Fertile eunuch syndrome	GNRHR
Fetal hypokinesia sequence due to restrictive dermopathy	LMNA+del
FG syndrome (FGS)	MED12
FG syndrome 1 (FGS1)	MED12
Fibrochondrogenesis	COL11A1+del
Fibrodysplasia ossificans progressiva (FOP)	ACVR1
Fibromatosis, gingival, 1 (GINGF1) (GINGF) (GGF1)	SOS1
Fibromatosis, gingival, hereditary (HGF)	SOS1
Fibromyxoid sarcoma, low grade (LGFMS)	CREB3L2
Fibular aplasia or hypoplasia femoral bowing and poly- syn- and oligodactyly	WNT7A
Fibular hypoplasia and complex brachydactyly	GDF5 (=CDMP1)
Finlay-Marks syndrome	KCTD1
Floating-Harbor syndrome (FLHS)	SRCAP
Focal cortical dysplasia of Taylor (FCDT)	TSC1+del
Focal cortical dysplasia, type II	TSC1+del
Focal dermal hypoplasia (FODH)	PORCN+del
Folate malabsorption, hereditary (HFM)	SLC46A1
Foramina parietalia permagna (FPP)	ALX4, MSX2+del
Foveal hypoplasia and presenile cataract syndrome	PAX6+del
Foveal hypoplasia with anterior segment anomalies or isolated	PAX6+del
Frasier syndrome (FS)	WT1+del
Freeman-Sheldon syndrome (FSS)	MYH3
Freeman-Sheldon syndrome variant (FSSV)	TNNT3, TNNI2, TPM2
Frontonasal dysplasia type 2 (FND2)	ALX4
Fructose intolerance, hereditary (HFI)	ALDOB+del
Fuhrmann syndrome (FUHRS)	WNT7A
Furlong syndrome	TGFBR1+dup

Galactose-1-phosphate uridylyltransferase deficiency	GALT+del
Galactosemia	GALT+del
Galactosyltransferase I deficiency, XGPT deficiency	B4GALT7
GALT deficiency	GALT+del
Gangliosidosis, generalized GM1	GLB1+del+dup
Gardner syndrome (GS)	APC+del+dup
Gastric cancer, somatic	APC+del+dup, FGFR2+del, KRAS
Gastrointestinal stromal tumor, familial (GIST)	KIT
GATA2 deficiency (MonoMAC (monocytopenia, NK- and B- lymphocytopenia, severe infections with M. avium complex (MAC), and risk of progression to MDS/AML))	GATA2
GATM deficiency	GATM
Geleophysic dysplasia (GLPD)	ADAMTSL2, FBN1+del
Genital anomaly with cardiomyopathy	LMNA+del
Geroderma osteodysplastica	GORAB (=SCYL1BP1)
Geroderma osteodysplasticum (GO)	GORAB (=SCYL1BP1)
Giant cell chondrodysplasia	FLNB
Gigantism, partial, of hands and feet, nevi, hemihypertrophy, and macrocephaly	AKT1, PTEN+del
Gillespie syndrome (GS)	PAX6+del
GLA deficiency	GLA+del
Glioma (GLM)	IDH1
Glioma susceptibility 2 (GLM2)	PTEN+del
Glutaric acidemia II (GA2)	ETFDH, ETFA+del, ETFB
Glutaric acidemia IIA	ETFA+del
Glutaric acidemia IIB	ETFB
Glutaric acidemia IIC (GA2C)	ETFDH
Glutaric aciduria II (GA2)	ETFDH, ETFA+del, ETFB
Glutaric aciduria III (GA3)	C7orf10
Glutaryl-CoA oxidase deficiency	C7orf10
Glycogen storage disease Ia (GSD1A)	G6PC
Glycogen storage disease type 2B (GSD2B), formerly	LAMP2+del
GM1-gangliosidosis	GLB1+del+dup
Golabi-Rosen syndrome	GPC3+del, GPC4del+dup
Goldblatt syndrome	N.N.
Goldenhar syndrome	TCOF1+del
Goltz syndrome	PORCN+del
Goltz-Gorlin syndrome	PORCN+del
Gonadoblastoma (GBY)	TSPY1 (=TSPY)
Gonadotropin deficiency, familial idiopathic (FIGD)	GNRH1
Gorlin syndrome	PTCH1+del
Gorlin-Goltz syndrome	PTCH1+del
Greig cephalopolysyndactyly syndrome (GCPS)	GLI3+del+dup
Gronblad-Strandberg syndrome, modifier of severity of	XYLT1
Growth hormone deficiency with pituitary anomalies	HESX1+del
Growth hormone deficiency, isolated	GHRHR, GH1+del+dup
Growth-mental deficiency syndrome of Myhre	SMAD4
Guanidinoacetate methyltransferase (GAMT) deficiency	GAMT
Hamartomas, pulmonary chondroid	HMGA2
HANAC syndrome (Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps)	COL4A1, COL4A2
Hanhart dwarfism	PROP1+del
Happy puppet syndrome, formerly	UBE3A+del
Hauptmann-Thannhauser muscular dystrophy	LMNA+del
Hay-Wells syndrome	TP63 (=p63)
Head cancer	TP63 (=p63)
Hearing loss, non-syndromic	COL9A3
Hearing loss, progressive, with optic atrophy and/or impaired glucose regulation	WFS1+del
Heart-hand syndrome	TBX5+del+dup
Heart-hand syndrome, Slovenian type	LMNA+del
Hemangioblastoma, cerebellar, somatic	VHL+del
Hemangioma, capillary infantile, susceptibility to (HCI)	ANTXR1 (=TEM8)
Hemangiomatosis	FLI1
Hematuria, benign familial (BFH)	COL4A3, COL4A4
Hemifacial microsomia (HFM)	TCOF1+del
Hemiparesis, and leukoencephalopathy, autosomal dominant	COL4A1, COL4A2
Hemiplegia, infantile, with porencephaly, type 1 (T1P)	COL4A1
Hemoglobin H disease, acquired	ATRX+del+dup
Hepatoblastoma	CTNNB1
Hepatoblastoma, somatic	APC+del+dup
Hepatocellular carcinoma (HCC)	CTNNB1
Hereditary absence of the proximal interphalangeal (PIP) joints	GDF5 (=CDMP1), NOG
Hereditary motor and sensory neuropathy, type IIc (HMSN2C)	TRPV4
Heredopathia atactica polyneuritiformis	PEX7+del
HFE hemochromatosis, modifier of	BMP2+del+dup
Hippel-Lindau disease (VHLD)	VHL+del
Hirschsprung disease, susceptibility to, 4 (HSCR4)	EDN3
Holmes-Gang syndrome	ATRX+del+dup
Holoprosencephaly-7 (HPE7)	PTCH1+del
Holt-Oram syndrome (HOS) (HOS1)	TBX5+del+dup
Humerospinal Dysostosis, HSD	CHST3 (=C6ST1)
Hurthle cell thyroid carcinoma (HCTC)	HRAS
Hutchinson-Gilford progeria syndrome (HGPS)	LMNA+del
Hydranencephaly with abnormal genitalia	ARX+del+dup
Hydrolethalus syndrome 2	KIF7
HYP	PHEX+del+dup
Hypercalciuric rickets	SLC34A3
Hyperchylomicronemia, familial	LPL+del+dup
Hyperekplexia, hereditary 1 (HKPX1)	GLRA1+del
Hyperinsulinemic hypoglycemia, familial (HHF)	ABCC8+del, KCNJ11
Hyperinsulinism, congenital	KCNJ11

Hyperkeratosis-contracture syndrome	LMNA+del
Hyperlipemia	LPL+del+dup
Hyperlipidemia, familial combined (FCHL)	LPL+del+dup
Hyperlipoproteinemia	LPL+del+dup, APOC2+del
Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	MAT1A
Hypernephroma	VHL+del
Hyperostosis corticalis deformans juvenilis	TNFRSF11B (=OPG)
Hyperostosis corticalis generalis	SOST
Hyperostosis with Hyperphosphatemia	FGF23
Hyperostosis, cortical, with syndactyly	SOST
Hyperostosis, endosteal, autosomal recessive	SOST
Hyperostosis-Hyperphosphatemia syndrome (HHS)	FGF23
Hyperphenylalaninemia, mild (HPA, mild)	PAH+del
Hyperphenylalaninemia, non-PKU mild (= HPA)	PAH+del
Hyperphosphatasemia tarda	SOST
Hyperphosphatasia	TNFRSF11B (=OPG)
Hypertelorism with esophageal abnormality and hypospadias	MID1+del+dup
Hypertelorism-hypospadias syndrome	MID1+del+dup
Hypertension, salt-resistant (?)	NPR3
Hyperthyroidism, familial, due to inappropriate thyrotropin secretion	THRB
Hyperthyroxinemia, familial euthyroid, secondary to pituitary and peripheral resistance to thyroid hormones	THRB
Hypertrophic osteoarthropathy, primary, autosomal recessive, 1 (PHOAR1)	HPGD
Hypochondrogenesis	COL2A1+del
Hypochondroplasia	FGFR3
Hypoglycemia of infancy, leucine-sensitive	ABCC8+del
Hypogonadism with spermatogenesis	GNRHR
Hypogonadotropic hypogonadism with or without anosmia (HH)	KAL1, FGFR1 (= KAL2), PROKR2, PROK2, CHD7, FGF8, GNRHR, KISS1R, NSMF (= NELF), TAC3, TACR3, GNRH1, KISS1, WDR11, HS6ST1, SEMA3A, SPRY4, IL17RD, DUSP6, FGF17, FLRT3, NR5A1, PIN1
Hypophosphatasia (HOPS)	ALPL (=TNSALP)+del
Hypophosphatemia	PHEX+del+dup; FGF23
Hypophosphatemic rickets	PHEX+del+dup; FGF23
Hypophosphatemic rickets with hypercalciuria (HHRH)	SLC34A3
Hypophosphatemic Vitamin D-resistant rickets (HPDR)	PHEX+del+dup
Hypoplastic left heart syndrome 2 (HLHS2)	NKX2-5
Hyposplenia, isolated congenital	NKX2-5
Hypothalamic hamartoblastoma, hypopituitarism, imperforate anus, and postaxial polydactyly	GLI3+del+dup
Hypothalamic hamartomas, somatic	GLI3+del+dup
Hypothyroidism, congenital nongoitrous, 5 (CHNG5)	NKX2-5
Hystrix-like ichthyosis with deafness (HID syndrome)	GJB2 (=CX26)
IBIDS syndrome	ERCC3 (=XPB)
I-cell disease (ICD)	GNPTAB+del+dup
Ichthyosiform erythroderma with hair abnormality and mental and growth retardation	ERCC3 (=XPB)
Ichthyosis, congenital, with trichothiodystrophy	ERCC3 (=XPB)
Idiopathic hypogonadotropic hypogonadism	KAL1, FGFR1 (= KAL2), PROKR2, PROK2, CHD7, FGF8, GNRHR, KISS1R, NSMF (= NELF), TAC3, TACR3, GNRH1, KISS1, WDR11, HS6ST1, SEMA3A, SPRY4, IL17RD, DUSP6, FGF17, FLRT3, NR5A1, PIN1
Idiopathic scoliosis 3, susceptibility to (IS3)	CHD7 (=KAL5)+del
Illig-type growth hormone deficiency	GH1+del+dup
IMAGE (Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomalies) syndrome	CDKN1C+dup
Immunodeficiency, isolated	IKBKG (=NEMO)+del
Immunodeficiency, severe combined, with hypereosinophilia	DCLRE1C (=ARTEMIS)+del, RAG1+del, RAG2+del
Immunodeficiency-centromeric instability-facial anomalies syndrome (ICF1)	DNMT3B
Inclusion cell disease (ICD)	GNPTAB+del+dup
Incontinentia pigmenti (IP)	IKBKG (=NEMO)+del
Incontinentia pigmenti, type II, formerly (IP2, formerly)	IKBKG (=NEMO)+del
Infantile cortical hyperostosis	COL1A1+del
Infantile epileptic-dyskinetic encephalopathy	ARX+del+dup
Infantile hemiparesis	COL4A1, COL4A2
Infantile spasm syndrome, X-linked (ISSX)	ARX+del+dup, CDKL5+del+dup
Infantile systemic hyalinosis (ISH)	ANTXR2 (=CMG2)
Infiltrative fibromatosis, familial (FIF)	APC+del+dup
Insulin resistance, susceptibility to	PTPN1
Interrupted aortic arch	NKX2-5
Intervertebral disc disease (IDD), susceptibility to	COL9A2, COL9A3
Invasive pneumococcal disease, recurrent isolated, 2 (IPD2)	IKBKG (=NEMO)+del
Iridogoniodysgenesis (IRID)	PITX2+del
Iridogoniodysgenesis syndrome (IGDS)	PITX2+del
Iris hypoplasia and glaucoma	PITX2+del
Isovaleric acidemia (IVA)	IVD
Jackson-Weiss syndrome (JWS)	FGFR1+del (=KAL2), FGFR2+del
Jadassohn nevus phakomatosis (JNP)	KRAS
Jansen metaphyseal chondrodysplasia (JMC)	PTH1R (= PTHR)
Jeune syndrome	WDR34, IFT80
Joubert syndrome (JBTS)	KIF7, TCTN3
JP/HHT syndrome	SMAD4
JPS/HHT	SMAD4
Juberg-Marsidi syndrome (JMS)	ATRX+del+dup (bei Frauen)
Juvenile hyaline fibromatosis (JHF)	ANTXR2 (=CMG2)
Juvenile intestinal polyposis (JIP)	BMPR1A+del
Juvenile polyposis coli	BMPR1A+del
Juvenile polyposis of infancy	BMPR1Adel, PTEN+del
Juvenile polyposis of stomach	BMPR1A+del
Juvenile polyposis syndrome (JPS)	BMPR1A+del, SMAD4, PTEN+del
Juvenile polyposis with hereditary telangiectasia	SMAD4

Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (JPHT)	SMAD4
Kallmann syndrome (-> Hypogonadotropic hypogonadism)	
Keller syndrome	MED12
Keratitis, hereditary (KERH)	PAX6+del
Keratitis-ichthyosis-deafness syndrome (KID syndrome)	GJB2 (=CX26)
Keratoderma, palmoplantar, with deafness (PPKDFN)	GJB2 (=CX26)
Keratosis palmoplantaris with cystic eyelids, hypodontia, and hypotrichosis	WNT10A
Keratosis, seborrheic, somatic	FGFR3
Kinky hair disease	ATP7A+del+dup
Klein-Waardenburg syndrome	PAX3+del
Klippel-Feil syndrome 1, autosomal dominant (KFS1)	GDF6
Kniest dysplasia (KD)	COL2A1+del
Kowarski syndrome (KWKS)	GH1+del+dup
L-2-Hydroxyglutaric acidemia (L2HGA)	L2HGDH+del
L-2-Hydroxyglutaric aciduria (L2HGA)	L2HGDH+del
Lacrimoauriculodentodigital syndrome (LADDS)	FGFR3, FGF10+del, FGFR2+del
LADD syndrome (LADDS)	FGFR3, FGF10+del, FGFR2+del
Lamb syndrome	PRKAR1A+del
Langer mesomelic dysplasia (LMD)	SHOX+del
Langer-Giedion syndrome (LGS)	EXT1+del (=TRPS2), TRPS1del
Larsen syndrome (LRS), autosomal dominant	FLNB
Larsen syndrome, autosomal recessive	CHST3 (=C6ST1)
Lateral cleft, isolated	PTCH2
Lateral incisors, absence of	WNT10A
Lateral incisors, pegged or missing	WNT10A
Legg-Calve-Perthes disease (LCPD; LCP)	COL2A1+del
Leigh syndrome due to pyruvate carboxylase deficiency	PC
Leiomatosis, diffuse, with Alport syndrome (DL-ATS)	COL4A5+del
Leiomatosis, esophageal and vulval, with nephropathy	COL4A5+del
Leiomyoma, uterine, somatic (UL)	HMGA2
Lentiginosis, cardiomyopathic	PTPN11+del+dup, RAF1+dup, BRAF
Lenz microphthalmia syndrome (LMS)	BCOR+del
LEOPARD syndrome	PTPN11+del+dup, RAF1+dup, BRAF
Leri-Weill dyschondrosteosis (LWD)	SHOX+del (erst Del dann Seq)
Lethal short-limbed platyspondylic dwarfism, Torrance type	COL2A1+del
Leukemia, acute lymphoblastic (ALL)	FLI1, LHX4+del
Leukemia, acute lymphocytic	FLI1, LHX4+del
Leukemia, acute myelogenous (AML)	TRIP11, JAK2, CREBBP+del, IDH1, NSD1 (=SOTOS)+del, KIT, KRAS, EP300+del, TERT, GATA2, RUNX1, CEBPA
Leukemia, acute myeloid	TRIP11, JAK2, CREBBP+del, IDH1, NSD1 (=SOTOS)+del, KIT, KRAS, EP300+del, TERT, GATA2, RUNX1, CEBPA
Leukemia, acute T-cell lymphoblastic	NOTCH1+del, SETBP1
Leukemia, juvenile myelomonocytic (JMML)	NF1+del, PTPN11
Leukemia, megakaryoblastic, with or without Down syndrome, somatic	GATA1
Leukodystrophy, hypomyelinating, 2 (HLD2)	GJC2
Leukoencephalopathy with Axenfeld-Rieger anomaly	COL4A1, COL4A2
Levy-Hollister syndrome	FGFR3, FGF10+del, FGFR2+del
Lhermitte-Duclos syndrome (LDD)	PTEN+del
Limb/pelvis-hypoplasia/aplasia syndrome (LPHAS)	WNT7A
Limb-mammary syndrome (LMS)	TP63 (=p63)
Linear sebaceous nevus syndrome	KRAS
Lipase D deficiency	LPL+del+dup
Lipoamide dehydrogenase deficiency, lactic acidosis due to	DLD (=LAD)
Lipoatrophic diabetes	LMNA+del, BSCL2+del
Lipocalcinogranulomatosis	FGF23
Lipodystrophy, Berardinelli-Seip congenital	BSCL2+del, CAV1, PTRF
Lipodystrophy, cephalothoracic type	LMNB2
Lipodystrophy, congenital generalized (CGL)	AGPAT2+del, BSCL2+del, CAV1, PTRF
Lipodystrophy, familial partial, Dunnigan type	LMNA+del
Lipodystrophy, familial partial, type 2 (FPLD2) (FPL2)	LMNA+del
Lipodystrophy, familial, of limbs and lower trunk	LMNA+del
Lipodystrophy, partial, acquired, susceptibility to (APLD) (APLD), susceptibility to	LMNB2
Lipodystrophy, partial, progressive	LMNB2
Lipodystrophy, reverse partial	LMNA+del
Lipodystrophy, total, and acromegaloid gigantism	BSCL2+del
Lipodystrophy, type A, associated with mandibuloacral dysplasia	LMNA+del
Lipoma (LIPO)	HMGA2
Lipomatosis of pancreas, congenital	SBDS+del
Lipomatosis, familial multiple (FML)	HMGA2
Lipomatosis, multiple	HMGA2
Lipoprotein lipase deficiency	LPL+del+dup
Lip-pit syndrome (LPS=PIT)	IRF6+del
Lissencephaly, X-linked 2 (LISX2)	ARX+del+dup
Lissencephaly, X-linked, with ambiguous genitalia (XLAG) (XLISG)	ARX+del+dup
Lobular carcinoma in situ (LCIS)	MLH1+del
Loeys-Dietz syndrome (LDS)	TGFBR1+dup, TGFBR2, SMAD3, TGFB2+del
Low-birth-weight dwarfism with skeletal dysplasia	RNU4ATAC
LPL deficiency	LPL+del+dup
Lujan-Fryns syndrome	MED12
Lumbar disc herniation (LDH), susceptibility to	COL11A1+del, MMP9
Lung cancer	TP63 (=p63)
Lung cancer susceptibility 2 (LNCR2)	CHRNA3
Lung cancer, non-small cell (NSCLC)	STK11
Lung cancer, somatic	KRAS
Lymphangioleiomyomatosis (LAM) (Lymphangiomatosis)	TSC1+del, TSC2+del
Lymphedema and yellow nails (LYYN)	FOXC2+del
Lymphedema with distichiasis (LYD)	FOXC2+del
Lymphedema, hereditary (LMPH)	GJC2, FOXC2+del
Lymphedema-distichiasis syndrome (LYDS)	FOXC2+del

Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus	FOXC2+del
Lynch syndome	MSH2+del, MLH1+del, PMS2+del, MSH6+del, TGFBR2+del, MLH3
Lysosomal glycogen storage disease without acid maltase deficiency, formerly	LAMP2+del
Macrocephaly/autism syndrome (MCEPHAS)	PTEN+del
Macrostomia, isolated	PTCH2
Maffucci syndrome (= Enchondromatosis, multiple, Maffucci type)	IDH1, IDH2
Majeed syndrome (MAJEEDS)	LPIN2
Male germ cell tumor (MGCT)	KIT
Male Turner snydrome	PTPN11+dup
Malignant rhabdoid tumor, somatic (MRT)	SMARCB1 (=INI1)+del
Malouf syndrome	LMNA+del
Mandibuloacral dysplasia with type A lipodystrophy (MADA)	LMNA+del
Mandibuloacral dysplasia with type A lipodystrophy, atypical	LMNA+del
Mandibulofacial dysostosis (MFD1)	TCOF1+del
Maple syrup urine disease, type III	DLD (=LAD)
Marfan syndrome (MFS)	FBN1+del
Marfan syndrome, atypical	COL1A2+del
Marfanoid craniosynostosis syndrome	SKI
Marfanoid disorder with craniosynostosis, type I	SKI
Marshall syndrome (MARSHS)	COL11A1+del
Marshall-Smith syndrome (MSS)	NFIX
MASS syndrome	FBN1+del
Mast cell disease	KIT
Mastocytosis	KIT
Meacham syndrome (MEACHS)	WT1+del
MECP2 duplication syndrome	MECP2+del
Medium chain Acyl-CoA dehydrogenase deficiency (MCAD)	ACADM (=MCAD)+del
Medulloblastoma (MDB)	CTNNB1
Medulloblastoma (MDB), somatic	APC+del+dup, PTCH1+del, PTCH2
Medulloblastoma with extensive nodularity (MBEN)	SUFU+del
Medulloblastoma, desmoplastic (MDB, desmoplastic)	SUFU+del
Meige disease	FOXC2+del
Meige lymphedema	FOXC2+del
Melanoma, cutaneous malignant, 9	TERT
Melanoma, desmoplastic neurotrophic (DNM)	NF1+del
Melanoma, malignant sporadic	STK11
Melanoma, malignant, somatic	BRAF, PTEN+del
Meningioma	PTEN+del
Menkes disease (MK) (MNK)	ATP7A+del+dup
Menkes syndrome	ATP7A+del+dup
Mental retardation and muscular atrophy	SLC16A2 (=MCT8)+del
Mental retardation, autosomal dominant 20 (MRD20)	MEF2C
Mental retardation, large head, imperforate anus, congenital hypotonia, and partial	MED12
Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	MEF2C
Mental retardation, X-linked 19 (MRX19)	RPS6KA3 (= RSK2) +del+dup
Mental retardation, X-linked 29/32/33/38/43/54/76/87	ARX+del+dup
Mental retardation, X-linked 36 (MRX36)	ARX+del+dup
Mental retardation, X-linked syndromic 1 (MRXS1)	ARX+del+dup
Mental retardation, X-linked syndromic 13 (MRXS13)	MECP2+del
Mental retardation, X-linked syndromic 16 (MRXS16)	FGD1+del
Mental retardation, X-linked syndromic, Lubs type (MRXSL)	MECP2+del
Mental retardation, X-linked, with creatine transporter deficiency	SLC6A8+del
Mental retardation, X-linked, with dystonic movements, ataxia, and seizures	ARX+del+dup
Mental retardation, X-linked, with growth retardation, deafness, and microgenitalism	ATRX+del+dup
Mental retardation, X-linked, with hypotonia	SLC16A2 (=MCT8)+del
Mental retardation, X-linked, with marfanoid habitus	MED12
Mental retardation, X-linked, with or without seizures, ARX-related (MRXARX)	ARX+del+dup
Mental retardation, X-linked, with seizures, short stature, and midface hypoplasia	SLC6A8+del
Mental retardation-hypotonic facies syndrome, X-linked (MRXHF1)	ATRX+del+dup
Mesomelic dwarfism of the hypoplastic ulna, fibula, and mandible type	SHOX+del
Mesothelioma, malignant; susceptibility to (MESOM)	CTNNB1
Mesothelioma, somatic (MESOM)	WT1+del
Metachondromatosis (METCDS)	PTPN11+del+dup
Metaphyseal anadysplasia (MANDP)	MMP13, MMP9
Metaphyseal chondrodysplasia, Jansen type	PTH1R (= PTHR)
Metaphyseal chondrodysplasia, McKusick type	RMRP
Metaphyseal chondrodysplasia, Murk Jansen type	PTH1R (= PTHR)
Metaphyseal chondrodysplasia, Schmid type (MCDS)	COL10A1
Metaphyseal chondromatosis with urinary excretion of D-2-hydroxy-glutaric acid (MC-HGA)	IDH1, IDH2
Metaphyseal dysplasia without hypotrichosis	RMRP
Metatropic dwarfism (MD)	TRPV4
Metatropic dysplasia (MTD)	TRPV4
Methionine adenosyltransferase (MAT) deficiency, autosomal recessive	MAT1A
Methylmalonic acidemia and homocystinuria, cblC type	MMACHC
Methylmalonic aciduria and homocystinuria, cblC type	MMACHC
Methylmalonic aciduria and homocystinuria, Vitamin B12-responsive	MMACHC
Microcephalic osteodysplastic primordial dwarfism, type I (MOPD1) (MOPD I) (MOPD)	RNU4ATAC
Microcephalic osteodysplastic primordial dwarfism, type II (MOPD2)	PCNT (=MOPD2)+del
Microphthalmia	VAX1
Microphthalmia and cataract 2	SIX6
Microphthalmia and esophageal atresia syndrome	SOX2+del
Microphthalmia with brain and digital anomalies	BMP4
Microphthalmia, cataract, and nystagmus	SIX6
Microphthalmia, isolated 4 (MCOP4)	GDF6
Microphthalmia, isolated, with cataract 2 (MCOPCT2)	SIX6
Microphthalmia, isolated, with coloboma 6, digenic (MCOPCB6)	GDF6
Microphthalmia, syndromic (MCOPS)	BCOR+del, SOX2+del, OTX2+del, BMP4

Microvascular complications of diabetes, susceptibility to, 1 (MVCD1)	VEGFA
Mismatch repair cancer syndrome (MMRCS)	MLH1+del, MSH2+del, MSH6+del, PMS2+del
Mohr-Majewski syndrome	TCTN3
Mohr-Wriedt type brachydactyly	GDF5 (=CDMP1), BMP2+del+dup, BMPR1B
Monocarboxylate transporter 8 (MCT8) deficiency	SLC16A2 (=MCT8)+del
Monocytopenia and mycobacterial infection syndrome (MONOMAC)	GATA2
Monocytopenia with susceptibility to mycobacterial, fungal, and papillomavirus infections and myelodysplasia	GATA2
MonoMAC (monocytopenia, NK- and B- lymphocytopenia, severe infections with M. avium complex (MAC), and risk of progression to MDS/AML) (GATA2 deficiency)	GATA2
Morbus Teutschlaender	FGF23
Morning glory disc anomaly	PAX6+del
Morquio A syndrome	GALNS+del
Morquio syndrome B	GLB1+del+dup
Motor and sensory neuropathy, hereditary, 4 (HMSN4)	PEX7+del
Mowat-Wilson syndrome (MWIS)	ZEB2+del
Moyamoya disease 5 (MYMY5)	ACTA2
Muckle-Wells syndrome (MWS)	NLRP3 (=NALP3)
Mucolipidosis II alpha/beta (ML II)	GNPTAB+del+dup
Mucolipidosis III alpha/beta (ML III)	GNPTAB+del+dup
Mucopolysaccharidosis type 4A (MPS4A)	GALNS+del
Mucopolysaccharidosis type IVB (MPS IVB) (MPS4B)	GLB1+del+dup
Muenke syndrome	FGFR3
Muir-Torre syndrome (MRTES)	MLH1+del, MSH2+del
Mullerian aplasia and hyperandrogenism (MULLAPL)	WNT4
Mullerian duct failure and hyperandrogenism	WNT4
Multicentric carpotarsal osteolysis syndrome (MCTO)	MAFB
Multicentric osteolysis, autosomal dominant	MAFB
Multicentric osteolysis, nodulosis, and arthropathy (MONA)	MMP14, MMP2
Multiple acyl-CoA dehydrogenase deficiency (MADD)	ETFDH, ETFA, ETFB
Multiple basal cell nevi, odonogenic keratocysts, and skeletal anomalies	PTCH1+del
Multiple hamartoma syndrome (MHAM)	PTEN+del
Multiple lentigines syndrome	PTPN11+del+dup, RAF1+dup, BRAF
Multiple lentigines, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonic stenosis, abnormal genitalia, retardation of growth, and sensorineural deafness (LEOPARD)	PTPN11+del+dup, RAF1+dup, BRAF
Multiple myeloma (MM)	FGFR3
Multiple self-healing squamous epithelioma, susceptiblity to (MSSE)	TGFBR1+dup
Multiple synostoses syndrome (SYNS)	NOG, GDF5 (=CDMP1), FGF9
Multisystem inflammatory disease, neonatal onset (NOMID)	NLRP3 (=NALP3)
Multisystemic smooth muscle dysfunction syndrome	ACTA2
Muscular dystrophy with early contractures and cardiomyopathy, autosomal dominant	LMNA+del
Muscular dystrophy, congenital, LMNA-related (MDCL)	LMNA+del
Muscular dystrophy, limb-girdle, type 1B (LGMD1B)	LMNA+del
Muscular dystrophy, proximal, type 1B	LMNA+del
Mycobacterial disease, susceptibility to, X-linked, type 1	IKBKG (=NEMO)+del
Myelodysplastic syndrome, susceptibility to	GATA2
Myelofibrosis with myeloid metaplasia, somatic (MMM)	MLP (=C-MLP)
Myelofibrosis, somatic	JAK2
Myelokathexis, isolated	CXCR4
Myhre syndrome (MYHRS)	SMAD4
Myopathy, congenital, with excess of muscle spindles (CMEMS)	HRAS
Myxoma, intracardiac (INTMYX)	PRKAR1A+del
Myxoma, spotty pigmentation, and endocrine overactivity	PRKAR1A+del
Naegeli syndrome	KRT14
Naegeli-Franceschetti-Jadassohn syndrome (NFJS)	KRT14
Nail-patella syndrome (NPS)	LMX1B+del
Nail-patella syndrome with primary open angle glaucoma (POAG)	LMX1B+del
Najjar syndrome	LMNA+del
Name syndrome	PRKAR1A+del
Nance-Insley syndrome	COL2A1+del, COL11A2
Nance-Sweeney chondrodysplasia	COL2A1+del, COL11A2
Navajo brainstem syndrome	HOXA1
Neck cancer	TP63 (=p63)
Nemaline myopathy 4 (NEM4)	TPM2
Nephroblastoma	WT1+del
Nephrolithiasis/osteoporosis, hypophosphatemic, 1 (NPHLOP1)	SLC34A1
Nephropathy, Wilms tumor, and genital anomalities	WT1+del
Nephrotic syndrome, type 4 (NPHS4)	WT1+del
Nesidioblastosis	KCNJ11
Netherton disease	SPINK5
Netherton syndrome (NETH) (NS)	SPINK5
Neurilemmomatosis, congenital cutaneous	SMARCB1 (=INI1)+del
Neurofibromatosis	NF1+del, SMARCB1 (=INI1)+del
Neurofibromatosis, type I, with leukemia (DD zu MMRCS)	MSH2+del
Neurofibromatosis-Noonan syndrome (NFNS)	NF1+del
Neuronopathy, distal hereditary motor, type V (HMN5)	BSCL2+del
Neuronopathy, distal hereditary motor, type VA (HMN5A) (HMN VA)	BSCL2+del
Neuropathy, distal hereditary motor, type VA (DHMN5A) (DHMN VA)	BSCL2+del
Neuropathy, hereditary sensory, type ID (HSN1D)	ATL1+del
Nevo syndrome	PLOD1+del+dup
Nevoid basal cell carcinoma syndrome (NBCCS)	PTCH1+del
Nevus sebaceous of Jadassohn	KRAS
Nevus, keratinocytic, nonepidermolytic (KNEN)	FGFR3
Nodulosis-arthropathy-osteolysis (NAO) syndrome	MMP14, MMP2
Nonproliferative retinopathy, diabetic, susceptibility to	VEGFA
Nonsmall cell lung cancer, somatic	BRAF
Noonan syndrome	PTPN11+dup, RAF1+del+dup, SOS1, RIT1, KRAS, MAP2K1, BRAF

Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSL)	CBL
Noonan-Neurofibromatosis syndrome	NF1+del
Obesity, adrenal insufficiency, and red hair	POMC
Obesity, autosomal dominant (OBESITY)	MC4R
Obesity, early-onset, susceptibility to	POMC
Obesity, morbide	CEP19
Occipital horn syndrome (OHS)	ATP7A+del+dup
Ocular coloboma	GDF6
Oculoauriculovertebral dysplasia (OAVD)	TCOF1+del
Oculoauriculovertebral spectrum (OAVS)	TCOF1+del
Oculofaciocardiodental (OFCD) syndrome	BCOR+del
O'Donnell-Papas syndrome	PAX6+del
Odontochondrodysplasia	N.N.
Odontohypophosphatasia	ALPL (=TNSALP)+del
Odontoonychodermal dysplasia (OODD)	WNT10A
OFD syndrome	TCTN3
Ohdo syndrome, X-linked (OHDOX)	MED12
Ohtahara syndrome, X-linked	ARX+del+dup
Oligodendroglioma	PTEN+del
Ollier disease	IDH1, IDH2, PTH1R (= PTHR)
Omenn syndrome (OS)	DCLRE1C (=ARTEMIS)+del, RAG1+del, RAG2+del
Ommunodeficiency NEMO-related without anhidrotic ectodermal dysplasia (NEMOID)	IKBKG (=NEMO)+del
Ondine curse	EDN3
Onychoosteodysplasia	LMX1B+del
Opitz BBBG syndrome, type 1 (BBBG1)	MID1+del+dup
Opitz GBBB syndrome, type I (GGGB1)	MID1+del+dup
Opitz GBBB syndrome, X-linked	MID1+del+dup
Opitz syndrome (OS)	MID1+del+dup
Opitz syndrome, X-linked (OSX)	MID1+del+dup
Opitz trigonocephaly syndrome	CD96
Opitz trigonocephaly-like syndrome	CD96
Opitz-G syndrome, type 1 (OGS1)	MID1+del+dup
Opitz-Kaveggia syndrome (OKS)	MED12
Optic nerve aplasia, bilateral (BONA)	PAX6+del
Optic nerve coloboma with renal disease	PAX2+del
Optic nerve head pits, bilateral, congenital	PAX6+del
Optic nerve hypoplasia and abnormalities of the central nervous system	SOX2+del
Optic nerve hypoplasia, bilateral (BONH)	PAX6+del
Oral-facial-digital syndrome, type IV	TCTN3
Organoid nevus phakomatosis	KRAS
Orofacial cleft 6, susceptibility to (OFC6)	IRF6+del, TP63 (=p63), BMP4
Orofaciodigital syndrome IV (OFD4) (OFDS IV)	TCTN3
OSMED, heterozygous (WZS)	COL11A2
Osteoarthritis of distal interphalangeal joints (OADIP)	MATN3
Osteoarthritis with mild chondrodysplasia (OACD)	COL2A1+del
Osteoarthritis, familial early-onset (FOA), susceptibility to	IDH1
Osteoarthritis, hand, (HOA)	MATN3
Osteoarthritis, susceptibility (OS)	GDF5 (=CDMP1), MATN3
Osteochondritis dissecans, short stature, and early-onset osteoarthritis (OD)	ACAN
Osteochondromas, multiple	EXT1+del (=TRPS2), EXT2+del
Osteochondromatosis	IDH1, IDH2, PTH1R (= PTHR)
Osteodysplastic primordial dwarfism, type I	RNU4ATAC
Osteodysplastic primordial dwarfism, type II	PCNT (=MOPD2)+del
Osteoectasia, familial	TNFRSF11B (=OPG)
Osteogenesis imperfecta (OI), autosomal recessive	SERPINF1, CRTAP, LEPRE1+del, PPIB, SERPINH1, FKBP10, SP7, BMP1, TMEM38B+del, WNT1, CREB3L1, PLOD2
Osteogenesis imperfecta (OI), autsomal dominant	COL1A1+del, COL1A2+del, IFITM5
Osteogenesis imperfecta with congenital joint contractures	PLOD2
Osteoglophonic dysplasia (OGD)	FGFR1+del (=KAL2)
Osteolysis, hereditary multicentric	MMP14, MMP2
Osteolysis, hereditary, of carpal bones with or without nephropathy	MAFB
Osteomalacia, tumor-induced	FGF23
Osteonecrosis of femoral head	COL2A1+del
Osteoporosis, postmenopausal	COL1A2+del
Osteoporosis, susceptibility to (OSTEOP)	COL1A1+del
Otospondylomegaepiphyseal dysplasia (OSMED)	COL2A1+del, COL11A2
Ovarian cancer (OC)	TP63 (= p63), CTNNB1
Ovarian cancer, endometrioid type	MSH6+del
Ovarian cancer, somatic (OC)	AKT1
Overlap connective tissue disease (OCTD)	FBN1+del
Ovotesticular disorder of sex development	SRY
Ovotesticular DSD	SRY
Oxycephaly	TWIST1+del
Pachydermoperiostosis (PDP), autosomal recessive	HPGD
Paget disease, juvenile (JPD)	TNFRSF11B (=OPG)
PAH deficiency	PAH+del
Pallister-Hall syndrome (PHS)	GLI3+del+dup
Pancreatic cancer (PNCA)	SMAD4
Pancreatic cancer, somatic (= Pancreatic carcinoma)	KRAS
Pancreatic cancer, sporadic	STK11
Pancreatic insufficiency and bone marrow dysfunction	SBDS+del
Pancreatitis, chronic	PRSS1
Pancreatitis, hereditary (PCTT) (HPC) (HP)	PRSS1
Panhypopituitarism	PROP1+del
Papillorenal syndrome	PAX2+del
Parastremmatic dwarfism (PSTD)	TRPV4
Parietal foramina 1 (PFM1)	MSX2+del, ALX4

Paris-Trousseau thrombocytopenia (TCPT)	FLI1
Partington syndrome	ARX+del+dup
Partington X-linked mental retardation syndrome (PRTS)	ARX+del+dup
PC deficiency	PC
Pearson	long range PCRs an mitochondrialer DNA
Pelizaeus-Merzenbacher-like disease, 1 (PMLD1)	GJC2
Pelviscapular dysplasia	TBX15
Pendred syndrome (PDS)	FOXI1
Perrault syndrome (PRS)	HSD17B4+del (= DBP)
Persistent hyperinsulinemic hypoglycemia of infancy (PHHI)	KCNJ11
Persistent truncus arteriosus (PTA)	NKX2-5
Peters anomaly (PAN)	PAX6+del, PITX2+del
Peters-plus syndrome (PpS)	B3GALTL+del
Peutz-Jeghers syndrome (PJS)	STK11+del
Pfeiffer syndrome (PS)	FGFR1+del (=KAL2), FGFR2+del
Phenylketonuria (PKU)	PAH+del
Pheochromocytoma	VHL+del
PHO, autosomal recessive	HPGD
Phytanic acid oxidase deficiency	PEX7+del
Piebald trait (PBT)	KIT+del, SNAI2+del
Piebaldism	KIT+del, SNAI2+del
Pigmented micronodular adrenocortical disease, primary, 1	PRKAR1A+del
Pigmented nodular adrenocortical disease, primary, 1 (PPNAD1)	PRKAR1A+del
Pilomatricoma (PTR)	CTNNB1
Pitt-Hopkins-like syndrome 2 (PTHSL2)	NRXN1+del
Pituitary dwarfism	GH1+del+dup, PROP1+del
Pituitary hormone deficiency, combined (CPHD)	POU1F1+del, PROP1+del, LHX3+del, LHX4+del, HESX1+del, OTX2+del
Pituitary hormone deficiency, combined, with rigid cervical spine or sensorineural deafness with pituitary dwarfism	LHX3+del
Platelet disorder, familial, with associated myeloid malignancy (FPDMM)	RUNX1
Platyspondylic lethal skeletal dysplasia Sand Diego type (PLSD-SD)	FGFR3
Platyspondylic lethal skeletal dysplasia, Luton type (PLSD-L)	COL2A1+del
Platyspondylic skeletal dysplasia, Torrance type (PLSD-T)	COL2A1+del
Poikiloderma with neutropenia (PN)	USB1 (= C16orf57)
Poikiloderma with neutropenia, Clericuzio-type	USB1 (= C16orf57)
Polycythemia rubra vera (PRV)	JAK2
Polycythemia vera (PV)	JAK2
Polycythemia, Chuvash type	VHL+del
Polycythemia, VHL-dependent	VHL+del
Polydactyly with neonatal chondrodystrophy, type III	WDR34
Polydactyly, postaxial and preaxial	GLI3+del+dup
Polyposis syndrome, hereditary mixed, 2 (HMPS2)	BMPR1A+del
Polyposis, familial, of enire gastrointestinal tract	BMPR1A+del, SMAD4
Polyposis, generalized juvenile, with pulmonary arteriovenous malformation	SMAD4
Polyposis, hamartomatous intestinal	STK11+del
Polyposis, juvenile intestinal (PJI)	BMPR1A+del, SMAD4
Polyps-and-spots syndrome	STK11+del
Polysyndactyly with peculiar skull shape	GLI3+del+dup
Polysyndactyly, uncomplicated	GLI3+del+dup
Popliteal pterygium syndrome 1 (PPS)	IRF6+del
POR deficiency	POR+del
Porencephaly, familial (PCEPH)	COL4A1
Porencephaly, type 1, autosomal dominant (ADT1P)	COL4A1
Postaxial polydactyly, type A (PAPA)	GLI3+del+dup
Posterior openbite, familial	PTH1R (= PTHR)
Potocki-Shaffer syndrome (PSS)	EXT2+del, ALX4
Prader-Willi syndrome (PWS)	Deletions-/Duplikations- und Methylierungsanalyse der PWS-kritischen Region in 15q11-q13 (insbesondere SNRPN-Gen)
Precocious puberty, central	KISS1R
Premature ovarian failure	FOXL2+del+dup, NR5A1
Preterm premature rupture of the membranes, susceptibility to (PPROM)	SERPINH1
Primary failure of eruption, nonsyndromic	PTH1R (= PTHR)
Primary retention of teeth	PTH1R (= PTHR)
Primordial dwarfism	GH1+del+dup
Progeria	LMNA+del
Progeria syndrome, childhood onset	LMNA+del
Progressive pseudorheumatoid dysplasia (PPD)	WISP3
Progressive pseudorheumatoid dysplasia with hypoplastic toes	COL2A1+del
Proliferative retinopathy, diabetic, susceptibility to	VEGFA
Proopiomelanocortin deficiency	POMC
Prostate cancer, somatic (PC)	PTEN+del
Proteus syndrome (PROTEUSS)	AKT1, PTEN+del
Proteus-like syndrome	PTEN+del
Proud syndrome	ARX+del+dup
Proximal 11p Deletion Syndrome (P11pDS)	EXT2+del, ALX4
Pseudoachondroplasia (PSACH)	COMP+del, COL9A3
Pseudoglycogenosis II	LAMP2+del
Pseudo-Hurler polydystrophy	GNPTAB+del+dup
Pseudo-Morquio syndrome, type 2	TRPV4
Pseudorheumatoid dysplasia, progressive (PPD)	WISP3
Pseudorheumatoid dysplasia, progressive, with hypoplastic toes	COL2A1+del
Pseudoxanthoma elasticum (PXE), modifier of severity of	XYLT1
PTEN hamartoma tumor syndrome (PHTS) (includes Cowden syndrome (CS), Bannayan-Riley-Ruvalcaba syndrome (BRRS), Proteus syndrome (PS), and Proteus-like syndrome)	PTEN+del
Pterygium colli syndrome	PTPN11
Pulmonary fibrosis, idiopathic, susceptibility to (IPF)	TERC+del,TERT+del
Pulmonary fibrosis, telomere-related, 1	TERT
Pulmonary hypertension, primary, 3 (PPH3)	CAV1

Pulmonic stenosis with cafe-au-lait spots	NF1+del
Puretic Syndrome	ANTXR2 (=CMG2)
Pycnodysostosis (PKND)	CTSK
Pyruvate carboxylase (PC) deficiency	PC
Radioulnar synostosis with amegakaryocytic thrombocytopenia	HOXA11
Rapp-Hodgkin syndrome (RHS)	TP63 (=p63)
Rear syndrome	SALL1+del
Refetoff syndrome	THRB
Refsum disease, adult (RDA) / classic	PEX7+del
Renal cell carcinoma, somatic (RCC)	VHL+del
Renal glucosuria (GLYS1)	SLC5A2+del
Renal hypoplasia, isolated	PAX2+del
Renal-coloboma syndrome	PAX2+del
Renal-ear-anal-radial syndrome	SALL1+del
Restrictive dermopathy, lethal	LMNA+del
Reticuloendotheliosis, familial, with eosinophilia	DCLRE1C (=ARTEMIS)+del, RAG1+del, RAG2+del
Retinal arteriolar tortuosity, infantile	COL4A1, COL4A2
Retinal detachment, rhegmatogenous, autosomal dominant (DRRD)	COL2A1+del
Retinal dystrophy, early-onset, and pituitary dysfunction	OTX2+del
Retinopathy, exudative. with bone marrow failure (ERBMF)	TINF2
Rett syndrome (RTT; RTS)	MECP2+del
Rett syndrome, atypical, CDKL5-related	CDKL5+del+dup, FOXG1+del
Revesz syndrome	TINF2
Rhabdoid predisposition syndrome 1 (RPS1)	SMARCB1 (=INI1)+del
Rhabdoid tumor (RDT)	SMARCB1 (=INI1)+del
Rhabdoid tumor predisposition syndrome (RTPS)	SMARCB1 (=INI1)+del, SMARCA4
Rhabdomyosarcoma 2 (RMS2)	PAX3+del
Rhabdomyosarcoma, alveolar (RMSA)	PAX3+del
Rhegmatogenous retinal detachment, autosomal dominant (DRRD)	COL2A1+del
Rhizomelic chondrodysplasia punctata, type 1 (RCDP1)	PEX7+del
Riley-Smith syndrome	PTEN+del
Ring dermoid of cornea (RDC)	PITX2+del
Rubinstein-Taybi syndrome (RSTS)	CREBBP+del, EP300+del
Ruvalcaba-Myhre-Smith syndrome (RMSS)	PTEN+del
Saethre-Chotzen syndrome (SCS)	FGFR2+del, TWIST1+del
Saethre-Chotzen syndrome with eyelid anomalies	TWIST1+del
Scalp-ear-nipple syndrome (SENS)	KCTD1
Scaphocephaly	TWIST1+del
Scaphocephaly and Axenfeld-Rieger anomaly	FGFR2+del
Scaphocephaly syndrome, familial (FSPC)	FGFR2+del
Scaphocephaly, maxillary retrusion, and mental retardation	FGFR2+del
Scapuloilioperoneal atrophy with cardiopathy	LMNA+del
Scapuloperoneal spinal muscular atrophy (SPSMA)	TRPV4
Schimmelpfennig-Feuerstein-Mims syndrome (SFM), somatic mosaic	KRAS
Schinzel phocomelia syndrome	WNT7A
Schinzel-Giedion midface retraction syndrome (SGS)	SETBP1
Schizophrenia, susceptibility to	AKT1, NRXN1del
Schmid-type metaphyseal chondrodysplasia (SMCD)	COL10A1
Schöpf-Schulz-Passarge syndrome (SSPS)	WNT10A
Schwannomatosis	SMARCB1 (=INI1)+del
Schwartz-Jampel syndrome (SJS)	HSPG2+del, LIFR
Sclerosteosis 1 (SOST1) (SOST)	SOST
Scoliosis, congenital, with unilateral unsegmented bar	FLNB
Sebaceous nevus syndrome, linear	KRAS
Seckel syndrome	PCNT (=MOPD2)+del
SED congenita	COL2A1+del
SED tarda, X-linked	TRAPPC2 (= SEDLIN)+del
SED, Maroteaux type	TRPV4
SED, Namaqualand type	COL2A1+del
Seip syndrome	BSCL2+del
SEN syndrome	KCTD1
Sensenbrenner syndrome	IFT122, WDR35+del, IFT43
Septooptic dysplasia	HESX1+del
SERKAL syndrome	WNT4
severe combined immunodeficiency Athabaskan-type (SCIDA)	DCLRE1C (= ARTEMIS)+del
severe combined immunodeficiency with sensitivity to ionizing radiation (RS-SCID)	DCLRE1C (= ARTEMIS)+del
Severe combined immunodeficiency, autosomal recessive, T-cell-negative, B-cell-negative, NK-cell-positive (T-B-NK+ SCID)	RAG1+del, RAG2+del
Severe combined immunodeficiency, B cell-negative (= B- SCID)	RAG1+del, RAG2+del
Sex reversal, XY, with or without adrenal failure	NR5A1
Sex-reversing locus on X (SRVX), formerly)	SRY
Sexual ateleiotic dwarfism	GH1+del+dup
SFM syndrome	KRAS
Sheldon-Hall syndrome (SHS)	TNNT3, TNNI2, MYH3, TPM2
Short rib-polydactyly syndrome (SRPS)	DYNC2H1, WDR34, WDR35+del, WDR60
Short stature syndrome, autosomal recessive, with intellectual disability	XYLT1
Short stature, idiopathic, X-linked (ISS)	SHOX+del
Short stature, pituitary and cerebellar defects, and small sella turcica	LHX4+del
Shprintzen-Goldberg craniosynostosis syndrome (SGS)	SKI
Shwachman-Bodian syndrome (SBS)	SBDS+del
Shwachman-Bodian-Diamond syndrome (SBDS)	SBDS+del
Shwachman-Diamond syndrome (SDS)	SBDS+del
Silver snydrome	BSCL2+del
Silver spastic paraplegia syndrome	BSCL2+del
Silver-Russel syndrome	Deletions-/Duplikations- und Methylierungsanalyse der differentiell methylierten Regionen KvDMR (u.a. KCNQ1-Gen) und H19DMR (u.a. H19-Gen) sowie des CDKN1C-Gens in der SRS-kritischen Region in 11p15; maternal uniparental disomy of chromosome 7; CDKN1C-Sequenzierung
Simpson dysmorphia syndrome (SDYS)	GPC3+del, GPC4del+dup

Simpson-Golabi-Behmel syndrome, type 1 (SGBS1) (SGBS)	GPC3+del, GPC4del+dup
SMD, Kozlowski type	TRPV4
Smith-Fineman-Myers syndrome type 1 (SFM1)	ATRX+del+dup
Smith-Lemli-Opitz syndrome (SLOS)	DHCR7+del
Smith-McCort dysplasia (SMC)	DYM+dup
Sotos syndrome (SOTOSS)	NSD1 (=SOTOS)+del, NFIX
Spastic paraplegia with amyotrophy of hands and feet	BSCL2+del
Spastic paraplegia, autosomal dominant (SPG)	ATL1+del, SPAST, BSCL2+del
Spastic paraplegia, autosomal recessive (SPG)	CYP7B1, SPG7 (=PGN)+del, GJC2
Spermatocytic seminoma, somatic	FGFR3
Spermatogenic failure 8 (SPGF8)	NR5A1
Spinal muscular atrophy, congenital	TRPV4
Spinal muscular atrophy, distal (DSMA)	BSCL2+del, TRPV4
Spinal muscular atrophy, distal, X-linked (SMAX)	ATP7A+del+dup
Splenic hypoplasia	NKX2-5
Split-hand/foot malformation (SHFM)	TP63 (=p63), WNT10B
Spondylism, congenital	FLNB
Spondylocarpotarsal syndrome	FLNB
Spondylocarpotarsal synostosis syndrome (SCT)	FLNB
Spondylocostal dysostosis, autosomal dominant (SCDO)	TBX6+dup
Spondylocostal dysostosis, autosomal recessive (SCDO)	DLL3, MESP2, LFNG, HES7
Spondylodysplasia and premature pubarche	PAPSS2
Spondyloenchondrodysplasia with D-2-hydroxyglutaric aciduria	IDH1, IDH2
Spondyloenchondrodysplasia with immune dysregulation (SPENCDI)	ACP5
Spondyloepimetaphyseal dysplasia (SEDM), aggrecan type	ACAN
Spondyloepimetaphyseal dysplasia (SEMD)	MATN3
Spondyloepimetaphyseal dysplasia with joint laxity (SEMDJL)	B3GALT6, KIF22
Spondyloepimetaphyseal dysplasia with multiple dislocations, Hall type	KIF22
Spondyloepimetaphyseal dysplasia, Missouri type (SEMD-MO)	MMP13
Spondyloepimetaphyseal dysplasia, Pakistani type (SEMD, PA)	PAPSS2
Spondyloepimetaphyseal dysplasia, Strudwick type (SEMD-STR)	COL2A1+del
Spondyloepiphyseal dysplasia congenita (SEDC)	COL2A1+del
Spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA)	WISP3
Spondyloepiphyseal dysplasia tarda, X-linked (SEDT)	TRAPPC2 (= SEDLIN)+del
Spondyloepiphyseal dysplasia with congenital joint dislocations	CHST3 (=C6ST1)
Spondyloepiphyseal dysplasia with precocious osteoarthritis	COL2A1+del
Spondyloepiphyseal dysplasia, congenital type	COL2A1+del
Spondyloepiphyseal dysplasia, Kimberley type (SEDK)	ACAN
Spondyloepiphyseal dysplasia, late	TRAPPC2 (= SEDLIN)+del
Spondyloepiphyseal dysplasia, Maroteaux type (SEDM)	TRPV4
Spondyloepiphyseal dysplasia, Omani type	CHST3 (=C6ST1)
Spondylohumerofemoral hypoplasia	FLNB
Spondylometaepiphyseal dysplasia, anauxetic type	RMRP, POP1
Spondylometaepiphyseal dysplasia, Menger type	RMRP, POP1
Spondylometaphyseal chondrodysplasia, japanese type (SMD)	COL10A1
Spondylometaphyseal dysplasia (SMD)	COL2A1+del
Spondylometaphyseal dysplasia with dentinogenesis imperfecta	N.N.
Spondylometaphyseal dysplasia, Kozlowski type (SMDK)	TRPV4
Spondyloperipheral dysplasia (SPD)	COL2A1+del
Squamous cell carcinoma, head and neck, somatic (HNSCC)	PTEN+del
St. Helena dysplasia	NPR2
Stapes ankylosis with broad thumb and toes (SABTS)	NOG
Steely hair disease	ATP7A+del+dup
Stickler sydrome	COL2A1+del, COL11A1+del, COL11A2, COL9A1, COL9A2
Stickler syndrome, atypical	COL2A1+del
Stickler syndrome, vitreous type	COL2A1+del, COL11A1+del
Stiff skin syndrome (SSKS)	FBN1+del
Strudwick syndrome	COL2A1+del
Strumpell disease	ATL1+del
Strumpell-Lorrain syndrome	ATL1+del
Stub thumb	HOXD13+del
Stueve-Wiedemann syndrome (SWS)	LIFR
Succedaneous teeth, agenesis of	WNT10A
Sugio-Kajii syndrome	TRPS1+del
Supravalvar aortic stenosis (SVAS)	ELN+del
Supravalvar aortic stenosis, Eisenberg type	ELN+del
Swyer syndrome	SRY
Symphalangism, proximal (SYM)	GDF5 (=CDMP1), NOG
Symphalangism-brachydactyly syndrome	NOG
Syndactyly with metacarpal and metatarsal fusion	HOXD13+del
Syndactyly, type 2 (SDTY2)	HOXD13+del
Syndactyly, type V (SDTY5)	HOXD13+del
Synostoses, multiple, with brachydactyly	NOG
Synostosis of talus and calcaneus with short stature	NOG
Synpolydactyly 1 (SPD1)	HOXD13+del
Synpolydactyly with foot anomalies	HOXD13+del
Systemic juvenile hyalinosis	ANTXR2 (=CMG2)
T3 resistance	SLC16A2 (=MCT8)+del
Tarsal-carpal coalition syndrome (TCC)	NOG
Tay syndrome	ERCC3 (=XPB)
Taybi-Linder syndrome (TALS)	RNU4ATAC
Telangiectasia, hereditary hemorrhagic, with juvenile polyposis coli	SMAD4
Telecanthus-hypospadias syndrome	MID1+del+dup
Temple-Syndrom (maternale uniparentale Disomie des Chromosoms 14)	u.a. MEG3
Teratoid tumor, atypical	SMARCB1 (=INI1)+del
Testicular germ cell tumor (TGCT)	STK11, KIT
Testicular tumor, sporadic	STK11
Testis-determining factor, X-chromosomal (TDFX), formerly	SRY
Tetralogy of Fallot (TOF)	JAG1+del+dup, NKX2-5, GATA4+del

Teunissen-Cremers syndrome	NOG
Teutschlaender disease, familial	FGF23
Thanatophoric dwarfism	FGFR3
Thanatophoric dysplasia, Luton variant	COL2A1+del
Thanatophoric dysplasia, Torrance variant	COL2A1+del
Thanatophoric dysplasia, type I (TD1)	FGFR3
Thanatophoric dysplasia, type II	FGFR3
Thin membrane nephropathy (TMN)	COL4A3, COL4A4
Thin-basement-mambrane nephropathy	COL4A3, COL4A4
Thrombocythemia (THCYT)	MLP (=C-MLP), JAK2
Thrombocytopenia with beta-thalassemia, X-linked (XLTT)	GATA1
Thrombocytopenia, amegakaryocytic, with radioulnar synostosis	HOXA11
Thrombocytopenia, congenital amegakaryocytic (CAMT)	MLP (=C-MLP)
Thrombocytopenia, familial, with propensity to acute myelogenous leukemia	RUNX1
Thrombocytopenia, X-linked, with or without dyserythropoietic anemia (XLTDA)	GATA1
Thrombocytosis	JAK2
Thyroid carcinoma, follicular (FTC), somatic	HRAS, PTEN+del
Thyroid carcinoma, papillary, somatic (PTC)	PRKAR1A+del
Thyroid hormone metabolism, abnormal	SECISBP2+del
Thyroid hormone resistance, generalized (GRTH) (GTHR)	THRB
Thyroid hormone resistance, selective pituitary (PRTH)	THRB
Thyroid hormone unresponsiveness	THRB
Tight skin contracture syndrome, lethal	LMNA+del
Tooth agenesis, selective, 4 (STHAG4)	WNT10A
Torg syndrome	MMP14, MMP2
Torg-Winchester syndrome	MMP14
Torg-Winchester syndrome, formerly	MMP2
Touraine-Solente-Gole syndrome	HPGD
Townes-Brocks branchiootorenal-like syndrome	SALL1+del
Townes-Brocks syndrome (TBS)	SALL1+del
Transverse cleft, isolated	PTCH2
Treacher Collins syndrome (TCS)	TCOF1+del
Treacher Collins syndrome 1 (TCS1)	TCOF1+del
Treacher Collins-Franceschetti syndrome (TCOF)	TCOF1+del
Trichorhinophalangeal syndrome (TRPS)	TRPS1+del, EXT1+del (=TRPS2)
Trichothiodystrophy with congenital ichthyosis	ERCC3 (=XPB)
Trichothiodystrophy, photosensitive (TTDP)	ERCC3 (=XPB)
Trigonocephaly syndrome	CD96
Trigonocephaly, nonsyndromic (TRICEPH)	FGFR1+del (=KAL2)
Triiodothyronine resistance	SLC16A2 (=MCT8)+del
Truncus arteriosus communis	NKX2-5
Trypsinogen deficiency	PRSS1
Tuberous sclerosis (TSC)	TSC2+del, TSC1+del
Tumoral calcinosis, hyperphosphatemic, familial (HFTC)	FGF23
Tumoral calcinosis, primary hyperphosphatemic (PHPTC)	FGF23
Turcot syndrome	MLH1+del, MSH2+del, MSH6+del, PMS2+del
Turner phenotype with normal karyotype	PTPN11+dup
Ulna and fibula, absence of, with severe limb deficiency	WNT7A
Unerupted second primary molar	PTH1R (= PTHR)
UPD14	Methylierungsanalyse der Region 14q32.2 (u.a. MEG3-Gen)
UPD7	Methylierungsanalyse der Region 7p12.1 und 7q32.2 (u.a. MEST-Gen)
Urticaria pigmentosa	KIT+del
Urticaria-deafness-amyloidosis (UDA) syndrome	NLRP3 (=NALP3)
VACTERL association	HOXD13+del
Vacuolar cardiomyopathy and myopathy, X-linked	LAMP2+del
Van Buchem disease (VBCH)	SOST
Van der Woude syndrome 1 (VWS1) (VDWS)	IRF6+del
VATERL association with hydrocephalus (VATERL-H)	PTEN+del
VATERL association with macrocephaly and ventriculomegaly	PTEN+del
Ventricular septal defect (VSD)	GATA4+del, NKX2-5
Verma-Naumoff syndrome	DYNC2H1, WDR34
Vertebral anomalies, anal atresia, congenital cardiac disease, tracheoesophageal fistula, renal anomalies, radial dysplasia, and other limb defects with macrocephaly and ventriculomegaly (VATERL)	PTEN+del
Vertebral defects (V), anal atresia (A), cardiac malformations (C), tracheoesophageal	HOXD13+del
Vertebral fusion with carpal coalition	FLNB
Very long chain Acyl-CoA dehydrogenase deficiency	ACADVL (=VLCAD)
Vitamin B12 metabolic defect with combined deficiency of mathylmalonyl-CoA mutase and homocysteine:methyltetrahydrofolate methyltransferase	MMACHC
Vitamin D-resistant rickets	PHEX+del+dup, FGF23
Vitreoretinopathy with phalangeal epiphyseal dysplasia	COL2A1+del
Vohwinkel syndrome (VS)	GJB2 (=CX26)
Von Gierke disease	G6PC
von Hippel-Lindau syndrome (VHLS)	VHL+del
Von Recklinghausen disease	NF1+del
Waardenburg syndrome with dystopia canthorum	PAX3+del, SNAI2del, EDN3
Wagner syndrome type 2 (WGN2)	COL2A1+del
WAGR syndrome	PAX6del, WT1del
WAGRO syndrome	PAX6del, WT1del
Walt Disney dwarfism	GORAB (=SCYL1BP1)
Watson syndrome	NF1+del
Weaver syndrome (WES)	EZH2, (NSD1)
Weaver-Smith syndrome (WSS)	EZH2, (NSD1)
Weill-Marchesani syndrome (WMS)	FBN1+del
Weissenbacher-Zweymueller syndrome (WZS)	COL11A2
West syndrome, X-linked	ARX+del+dup
Weyers acrodental dysostosis (WAD)	EVC (=EVC1)+del, EVC2+del
Weyers acrofacial dysostosis	EVC (=EVC1)+del, EVC2+del
WHIM (Warts, hypogammaglobulinemia, infections, and myelokathexis) syndrome	CXCR4

White forelock with malformations	PAX3+del
Wiedemann-Beckwith syndroms (WBS)	CDKN1C+dup; NSD1 (=SOTOS)+del
Williams-Beuren syndrome (WBS)	ELNdel
Wilms tumor and pseudo- or true hermaphroditism	WT1+del
Wilms tumor, aniridia, genitourinary anomalies, and mental retardation syndrome	PAX6del, WT1del
Wilms tumor, aniridia, genitourinary anomalies, mental retardation, and obesity syndrome	PAX6del, WT1del
Wilms tumor, somatic	GPC3
Wilms tumor, type 1 (WT1)	WT1+del
Winchester syndrome (WNCHRS)	MMP14
Wolfram syndrome 1 (WFS1) (WFS)	WFS1+del
Wolfram-like syndrome, autosomal dominant (WFSL)	WFS1+del
Wrinkly skin syndrome (WSS)	ATP6V0A2+del
WS4B with Hirschsprung disease	EDN3
Xeroderma pigmentosum, group B (XBP)	ERCC3 (=XPB)
Xeroderma pigmentosum/Cockayne syndrome (XPB/CS)	ERCC3 (=XPB)
X-linked recessive chondrodysplasia punctata type 1 (CPXR1)	ARSE+del bei Frauen
XLMR-Hypotonic facies syndrome	ATRX+del+dup (bei Frauen)
XMESID	ARX+del+dup
XP, group B (XPBC)	ERCC3 (=XPB)
XX Male, SRY-positive	SRY
Yellow nail syndrome (YNS)	FOXC2+del