OMIM Entities	Phenotype MIM number	Chromosome band	Key gene(s)
Chromosome 1p36 deletion syndrome	607872	1p36	multiple
Chromosome 1q21.1 deletion syndrome	612474	1q21.1	multiple
Chromosome 1q21.1 duplication syndrome	612475	1q21.1	multiple
Chromosome 1q41-q42 deletion syndrome	612530	1q41-q42	multiple
Mowat-Wilson syndrome	235730	2q22.3	*ZEB2*
Chromosome 2q37 deletion syndrome	600430	2q37	multiple
Chromosome 3q29 deletion syndrome	609425	3q29	multiple
Chromosome 3q29 duplication syndrome	611936	3q29	unknown
Familial adenomatous polyposis 1 Gardner syndrome; Brain tumor-polyposis syndrome 2; Adenomatous polyposis coli	175100	5q22.2	*APC*
Williams-Beuren region duplication syndrome; Chromosome 7q11.23 duplication syndrome	609757	7q11.23	multiple
8p23.1 deletion	https://rarediseases.info.nih.gov/diseases/3769/chromosome-8p231-deletion; https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2765374/	8p23.1	*GATA4*
KLEEFSTRA SYNDROME 1	610253	9q34.3	*EHMT1*
Witteveen-Kolk syndrome	613406	15q24	*SIN3A*
Chromosome 16p12.2-p11.2 deletion syndrome	613604	16p11.2-p12.1	multiple
Potocki-Lupski syndrome	610883	17p11.2	multiple
Koolen-De Vries syndrome	610443	17q21.31	multiple genes including *KANSL1*
Chromosome 17q21.31 duplication syndrome	613533	17q21.31	multiple
Chromosome 22q11.2 duplication syndrome	608363	22q11.2	multiple
Phelan-McDermid syndrome	606232	22q13.3	multiple genes including *SHANK3*
Adrenal hypoplasia, congenital	300200	Xp21.2	*NR0B1*
Alagille syndrome 1	118450	20p12.2	*JAG1*
Alpha-thalassemia/mental retardation syndrome, type 1	301040	Xq21.1	*ATRX*
Alport syndrome 1, X-linked	301050	Xq22.3	*COL4A5*
Androgen insensitivity syndrome	300068	Xq12	*AR*
Angelman syndrome	105830	15q11.2-q13	*UBE3A*
Aniridia; Cataract with late-onset corneal dystrophy	106210	11p13	*PAX6*

Wilms tumor, aniridia, genitourinary anomalies and mental retardation syndrome	194072	11p13	multiple genes including *PAX6* and *WT1*
Basal cell nevus syndrome	109400	9q22.32	*PTCH1*
Beckwith-Wiedemann syndrome	130650	11p15.4, 11p15.5	*ICR1* *KCNQ1OT1* *CDKN1C*
Blepharophimosis, epicanthus inversus, and ptosis	110100	3q22.3	*FOXL2*
Mental retardation, autosomal recessive 59		8q21.13	*IMPA1*
Branchiootorenal syndrome 1	113650	8q13.3	*EYA1*
Otofaciocervical syndrome	166780	8q13.3	multiple genes including *EYA1*
Agammaglobulinemia, X-linked 1	300755	Xq22.1	*BTK*
Campomelic dysplasia; **Acampomelic campomelic dysplasia;** **Campomelic dysplasia with autosomal sex reversal**	114290	17q24.3	*SOX9*
Cardiofaciocutaneous syndrome	115150	7q34	*BRAF*
Cat eye syndrome	115470	22q11	unknown
Charcot-Marie-Tooth disease, type 1A	118220	17p12	*PMP22*
CHARGE syndrome	214800	8q12.2	*CHD7*
Chromosome 10q22.3-q23.2 deletion syndrome	612242	10q22.3-q23.2	multiple
Chromosome 18p deletion syndrome	146390	18p	multiple
Chromosome 18q deletion syndrome	601808	18q	multiple
Cleidocranial dysplasia; **Cleidocranial dysplasia, forme fruste, dental anomalies only;** **Cleidocranial dysplasia, forme fruste, with brachydactyly**	119600	6p21.1	*RUNX2*
Cornelia de Lange syndrome 1	122470	5p13.2	*NIPBL*
CRI-DU-CHAT SYNDROME	123450	5p15.2-p13.3	multiple genes including *TERT*
Menkes disease	309400	Xq21.1	*ATP7A*
Cystinosis, nephropathic; **Cystinosis, atypical nephropathic**	219800	17p13.2	*CTNS*
Dandy-Walker syndrome	220200	3q22-q24	unknown
Hernia, congenital diaphragmatic 1	142340	15q26.1	unknown
DiGeorge syndrome	188400	22q11.21	*TBX1*

DiGeorge syndrome/velocardiofacial syndrome complex-2	601362	10p14-p13	unknown
DOWN SYNDROME	190685	21q22.3	multiple
Duchenne muscular dystrophy	310200	Xp21.2-p21.1	*DMD*
Dyggve-Melchior-Clausen disease	223800	18q21.1	*DYM*
Feingold syndrome 1	164280	2p24.3	*MYCN*
Forebrain defects	187395	3p21.31	*TDGF1*
Fragile X syndrome	300624	Xq27.3	*FMR1*
Glycerol kinase deficiency	307030	Xp21.2	*GK*
Neuropathy, recurrent, with pressure palsies	162500	17p12	*PMP22*
Holoprosencephaly 1	236100	21q22.3	unknown
Holoprosencephaly 2	157170	2p21	*SIX3*
Holoprosencephaly 3	142945	7q36.3	*SHH*
Holoprosencephaly 4	142946	18p11.31	*TGIF*
Holoprosencephaly 5	609637	13q32.3	*ZIC2*
Holoprosencephaly 6	605934	2q37.1-q37.3	*HPE6*
Holoprosencephaly 7	601309	9q22.32	*PTCH1*
Holoprosencephaly 9	610829	2q14.2	unknown
Developmental and epileptic encephalopathy 1; Epileptic encephalopathy, early infantile, 1	308350	Xp21.3	*ARX*
Developmental and epileptic encephalopathy 2	300672	Xp22.13	*CDKL5*
Jacobsen syndrome	147791	11q24-q25	multiple
Nephronophthisis 1, juvenile	256100	2q13	*NPHP1*
Trichorhinophalangeal syndrome, type II	150230	8q24.11-q24.13	multiple genes involving *TRPS1* and *EXT1*
Lesch-Nyhan syndrome	300322	Xq26.2-q26.3	*HPRT1*
Lissencephaly, X-linked; Subcortical laminal heterotopia, X-linked	300067	Xq23	*DCX*
Lissencephaly 1; Subcortical laminar heterotopia	607432	17p13.3	*PAFAH1B1*
Mental retardation, X-linked syndromic, Lubs type	300260	Xq28	*MECP2*
Linear skin defects with multiple congenital anomalies 1	309801	Xp22.2	*HCCS*
Atrial septal defect 7, with or without AV conduction defects	108900	5q35.1	*NKX2-5*

Chromosome 2p16.1-p15 deletion syndrome	612513	2p15-p16.1	multiple
Microphthalmia, syndromic 6	607932	14q22.2	*BMP4*
Optic nerve hypoplasia and abnormalities of the central nervous system; Microphthalmia, syndromic 3	206900	3q26.33	*SOX2*
Miller-Dieker lissencephaly syndrome	247200	17p13.3	multiple
Chromosome 9p deletion syndrome	158170	9p	multiple
Mucopolysaccharidosis II	309900	Xq28	*IDS*
Nail-patella syndrome	161200	9q33.3	*LMX1B*
Noonan syndrome 1	163950	12q24.13	*PTPN11*
Opitz GBBB syndrome, type I	300000	Xp22.2	*MID1*
Orofaciodigital syndrome I	311200	Xp22.2	*OFD1*
Pallister-Killian syndrome	601803	12p	unknown
Pelizaeus-Merzbacher disease	312080	Xq22.2	*PLP1*
Potocki-Shaffer syndrome	601224	11p11.2	multiple
Prader-Willi syndrome	176270	15q11.2	multiple genes including *SNRPN*, *NDN*
Rett syndrome	312750	Xq28	*MECP2*
Axenfeld-Rieger syndrome, type 1	180500	4q25	*PITX2*
Rubinstein-Taybi syndrome 1	180849	16p13.3	*CREBBP*
sacral/anorectal malformations	https://doi.org/10.1038/sj.ejhg.5201635	6q25.3	unknown
Saethre-Chotzen syndrome with or without eyelid anomalies	101400	7p21.1	*TWIST1*
Smith-Magenis syndrome	182290	17p11.2	multiple genes including *RAI1*
Sotos syndrome 1	117550	5q35.3	*NSD1*
Split-hand/foot malformation 1	183600	7q21.3	multiple genes including *DSS1, DLX5, DLX6*
Split-hand/foot malformation 3	246560	10q24	multiple
Split-hand/foot malformation 5	606708	2q31	unknown
Subtelomeric regions (41 regions)		41 sites	_
Synpolydactyly 1	186000	2q31.1	*HOXD13*
Ichthyosis, X-linked	308100	Xp22.31	*STS*
Thrombocytopenia-absent radius syndrome	274000	1q21.1	*RBM8A*
Treacher Collins syndrome 1	154500	5q32-q33	*TCOF1*
Trichorhinophalangeal syndrome, type I	190350	8q23.3	*TRPS1*
van der Woude syndrome 1	119300	1p32.2	*IRF6*
Waardenburg syndrome, type 1	193500	2q36.1	*PAX3*

Waardenburg syndrome, type 2A	193510		3p13	*MITF*
Williams-Beuren syndrome	194050		7q11.23	multiple
Wilms tumor	194070		11p13	*WT1*
Wolf-Hirschhorn syndrome	194190		4p16.3	multiple
X-inactivation, familial skewed	300087		Xq13.2	*XIST*
Heterotaxy, visceral, 1, X-linked; Congenital heart defects, nonsyndromic, 1, X-linked	306955		Xq26.3	*ZIC3*
Lymphoproliferative syndrome, X-linked, 1	308240		Xq25	*SH2D1A*
Chromosome Xp11.3 deletion syndrome	300578		Xp11.3	multiple genes including *RP2*
Trisomy 13				
Trisomy 18				
Trisomy 21				
Turner Syndrome(Monosomy X)				
Jacob's Syndrome(XYY)				
Klinefelter Syndrome(XXY)				
Triple X Syndrome(XXX)				
MENTAL RETARDATION, AUTOSOMAL DOMINANT 1; MRD1	156200		2q23.1	*MBD5*
MICROCORIA, CONGENITAL	156600		13q32	no specific
OTODENTAL DYSPLASIA	166750		11q13	*FGF3*
RECOMBINANT CHROMOSOME 8 SYNDROME	179613		8q22.1-qter, 8pter-p23.1	no specific
CHROMOSOME 2q35 DUPLICATION SYNDROME	185900		2q34-q36	no specific
VELOCARDIOFACIAL SYNDROME; VCFS	192430		22q11.21	*TBX1*
BARAITSER-WINTER SYNDROME 1; BRWS1	243310		7p22.1	*ACTB*
46,XX SEX REVERSAL 2; SRXX2	278850		17q24.3-q25.1	*SOX9*
MESOMELIA-SYNOSTOSES SYNDROME	600383		8q13	no specific
NABLUS MASK-LIKE FACIAL SYNDROME; NMLFS	608156		8q22.1	no specific
SPINOCEREBELLAR ATAXIA 20; SCA20	608687		11q12	no specific
CHROMOSOME 10q26 DELETION SYNDROME	609625		10q26	no specific
FRIAS SYNDROME	609640		14q22.1-q22.3	*BMP4*
CHROMOSOME 22q11.2 DELETION SYNDROME, DISTAL		611867	22q11.2	no specific
GLASS SYNDROME; GLASS	612313		2q33.1	*SATB2*

CHROMOSOME 17p13.3, TELOMERIC, DUPLICATION SYNDROME	612576	17p13.3-p13.1	no specific
CORNEAL DYSTROPHY, POSTERIOR AMORPHOUS; PACD	612868	12q21.33	no specific
CHROMOSOME 19q13.11 DELETION SYNDROME, DISTAL	613026	19q13.11	no specific
MENTAL RETARDATION, AUTOSOMAL DOMINANT 20; MRD20	613443	5q14.3	*MEF2C*
CHROMOSOME 16p13.3 DUPLICATION SYNDROME	613458	16p13.3	*CREBBP*
CHROMOSOME 17q23.1-q23.2 DUPLICATION SYNDROME	613618	17q23.1-q23.2	no specific
CHROMOSOME 17q11.2 DELETION SYNDROME, 1.4-MB	613675	17q11.2	*NF1*
CHROMOSOME 2q31.1 DUPLICATION SYNDROME	613681	2q31.1	no specific
BRAIN MALFORMATIONS WITH OR WITHOUT URINARY TRACT DEFECTS; BRMUTD	613735	1p31.3	*NFIA*
CHROMOSOME 17p13.1 DELETION SYNDROME	613776	17p13.1	no specific
CHROMOSOME 3pter-p25 DELETION SYNDROME	613792	3pter-p25	no specific
CHROMOSOME 13q14 DELETION SYNDROME	613884	13q14	no specific
CHROMOSOME 8q21.11 DELETION SYNDROME	614230	8q21.11	no specific
CHROMOSOME 15q25 DELETION SYNDROME	614294	15q25	no specific
CHROMOSOME 17q12 DUPLICATION SYNDROME	614526	17q12	no specific
CHROMOSOME 17q12 DELETION SYNDROME	614527	17q12	no specific
CHROMOSOME 3q13.31 DELETION SYNDROME	615433	3q13.31	*ZBTB20*
CHROMOSOME 15q11.2 DELETION SYNDROME	615656	15q11.2	*TUBGCP5* (608147)*, NIPA1 (*608145*),* *NIPA2 (*608146*),* and *CYFIP1 (*606322*)*
CHROMOSOME 5q12 DELETION SYNDROME	615668	5q12	no specific
CHROMOSOME 15q14 DELETION SYNDROME	616898	15q14	*MEIS2*
CHROMOSOME 11p13 DELETION SYNDROME, DISTAL	616902	11p13	*ELP4* (606985) and *PAX6* (607108) genes

2q13 deletion syndrome	——	2q13	no specific
15q13.2-13.3 deletion (CHRNA7)	——	15q13.2-13.3	no specific
RCAD syndrome (renal cysts and diabetes) (HNF1B or ACACA)	137920	17q12	*HNF1B; ACACA*
22q11.2 distal deletion (BCR, MAPK1)	611867	22q11.2	*BCR; MAPK1*
1q44 deletion syndrome (AKT3)	615937	1q44	*AKT3*
NRXN1 deletion syndrome	614332	2p16	*NRXN1*
5q14.3 deletion syndrome (MEF2C)	612881	5q14.3	*MEF2C*
15q26 deletion syndrome (IGF1R)	612626	15q26	*IGF1R*
Pitt-Hopkins syndrome (TCF4)	610954	18q21.2	*TCF4*
DYRK1A deletion syndrome	600855	21q22.13	*DYRK1A*
22q13.3 duplication syndrome (SHANK3/PROSAP2)	——	22q13.3	*SHANK3*
16p13.11 duplication	——	16p13.11	no specific
11p13 duplication/triplication syndrome (PAX6)	——	11p13	*PAX6*