

Disorder / Conditions List

Disorder / Condition	OMIM #	Chromosome
1p36 microdeletion	607872	1p36
1q21 microdeletion (TAR)	274000	1q21.1
1q21.2 deletion/duplication		1q21.2
1q41-q42 microdeletion	229850	1q41
2p15-p16.1 microdeletion		2p15-p16.1
2q22.3 deletion syndrome		2q22.3
2q32.2-q33 microdeletion	119540	2q33.1
2q37 subtelomeric deletion	600430	2q37
3q29 microdeletion/duplication	609425	3q29
5q21.1-q31.2 deletion syndrome		5q21.1-q31.2
6p25.3 microdeletion	601631	6p25.3
7q11.23 microduplication	609757	7q11.23
8p23.1 microdeletion		8p23.1
8p23p22 deletion/duplication syndrome		8p22
9p23 deletion syndrome/Trigonocephaly	158170	9p23
9q22.32-q22.33 microdeletion		9q22.33
9q34 microdeletion *	610253	9q34.3
10q22.3-q23.31 microdeletion		10q22.3-q23.31
10q deletion syndrome	609625	10q26
12q14.1-q15 microdeletion		12q14.3
12q24.21-q24.23 microduplication		12q24.21-q24.23
13q32 deletion *	609637	13q32
14q11.2 syndrome	605463	14q11.2
14q22-q23 microdeletion	600037	14q22-q23
15q11-BP1-BP2 interval deletion & duplication *		15q11.2
15q11.2-15q13.1 microduplication	608636	15q11.2-q13.1
15q13.3 microdeletion	612001	15q13.3
15q21 deletion syndrome	607624	15q21.1-q31.2
15q24.1-q24.3 microdeletion		15q24.1-q24.3
15q26.3 deletion w/ severe intrauterine growth restriction (IUGR)	608183	15q26.3
16p11.2 microdeletion/duplication (AUTS14)	611913	16p11.2
16p11.2-p12.2 microdeletion/ microduplication		16p11.2-p12.2
16p13.1 microdeletion/ microduplication		16p13.1
16p13.3 microdeletion/Severe Rubinstein-Taybi	610543	16p13.3
17q21.31 duplication syndrome; reciprocal 17q21.31 deletion syndrome	610443	17q21.31
18p deletion syndrome	146390	18p11.31
18q deletion syndrome	601808	18q23
22q11.2 distal microdeletion	611867	22q11.2
22q11.2 Duplication	608363	22q11.2
22q13.3 microdeletion	606232	22q13.3
Aarskog-Scott		Xp11.22
Adrenal hypoplasia congenital	300200	Xp21.2
Alagille	118450	20p12.2
Albright hereditary osteodystrophy-like/ Brachydactyl-MR	600430	2q37.3
Alpha thalassemia mental retardation (ATR-16)	141750	16pter-16p13.3
Alport syndrome, X-linked (ATS) *	301050	Xq22.3
Alzheimer disease, early onset with cerebral amyloid angiopathy * (FISH)	104300	21q21.3
Androgen insensitivity syndrome	300068	Xq12
Angelman	105830	15q11.2
Aniridia II	106210	11p13
Atrial septal defect (ASD) with atrioventricular conduction defects *	108900	5q35.1-5q35.2
Axenfeld-Rieger syndrome	602482	6p25
AZFa & AZFb		Yq11.22
Bannayan-Riley-Ruvalcaba (BRRS) *	153480	10q23.31
Bartter 3 (classic) *	607364	1p36.13
Bartter 4 (infantile with sensorineural deafness) *	602522	1p36.13
Bartter with autosomal dominant hypocalcemia	601199	3q21.1
Basal cell nevus	109400	9q22.32
Beckwith- Wiedemann	130650	11p15.5
Blepharophimosis, ptosis, epicanthus inversus	110100	3q22.3
Brachydactyly *		13q31.3
Brachydactyly, type C (BDC)	113100	20q11.2
Branchio-oto-renal (BOR)/Meinick-Fraser *	113650	8q13.3
Bruton agammaglobulinemia tyrosine	300300	Xq22.1
Buschke-Ollendorf	166700	12q14.3
Campomelic dysplasia *	114290	17q24.3
Cat-eye	115470	22q11.21

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Cerebellar hypoplasia	224050	Xq12
CHARGE *	214800	8q12.2
Chondrodysplasia *	118650	12q13.11-q13.2
Choroideremia *	303100	Xq21.2
Chronic granulomatous disease, X-linked	306400	Xp11.4
Cleft lip/Cleft palate		Xp11.22
Cleft palate, isolated (CPI) *	119540	2q32
Cleidocranial dysplasia (CCD)	119600	6p12.3
Coffin-Lowry	303600	Xp22.12
Congenital adrenal hyperplasia (CAH)	201910/119600	6p21.32
Congenital diaphragmatic hernia	142340	15q26.1-26.2
Congenital diaphragmatic hernia 2 (CDH2)	222400	8p23.1
Cornelia de Lange	122470	5p13.2
Cornelia de Lange syndrome 2 (CDLS2)	300590	Xp11.22-p11.21
Cowden *	158350	10q23.2-q23.31
Craniosynostosis with 11p15.2 disruption *		11p15.2
Cri-du-Chat	123450	5p15.2-p13.3
Currarino	176450	7q36.3
Dandy-Walker malformation	220200	3q24
Dent disease	300009	Xp11.22
Diabetes insipidus, X-linked	304800	Xp28
Diabetes Mellitus, transient neonatal, 1	601410	6q24.2
DiGeorge 2 *	601362	10p13-p14
DiGeorge/Velocardiofacial	188400/192430	22q11.2
Dosage sensitive sex reversal	300018	Xp21.2
Down Syndrome	190685	21q22
Duane radial ray syndrome *	607323	20q13.2
Duchenne/Becker muscular dystrophy (large del/dup)	310200	Xp21.2
Dyggve-Melchior-Clausen syndrome (DMC) *	223800	18q21.1
Fabry disease	301500	Xq22.1
Familial adenomatous polyposis (FAP)/Gardner/MR	175100	5q22.2
Familial hypocalcaemic hypercalcaemia 1 (HHC1)	145980	3q21.1
Feingold	164280	2p24.3
FG 5 *	300581	Xq22.3
FMR1 microdeletion	300624	Xq27.3
Fryns *	229850	1q41
Generalized epilepsy with febrile seizures plus 2 (GEFS+2) *	604233	2q24.3
Gitelman *	263800	16q13
Glycerol kinase deficiency	300474	Xp21.2
Gorlin Syndrome	610828	9q22.32
Greig cephalopolysyndactyly	175700	7p14.1
HADH2 deficiency	300438	Xp11.22
Hearing Loss, Hereditary		13q12.11
Hemophilia A	306700	Xq28
Hirschsprung	142623	10q11.21
Holoprosencephaly & preaxial polydactyly	605651	5q35.1
Holoprosencephaly 1	236100	21q22.3
Holoprosencephaly 2	157170	2p21
Holoprosencephaly 3	142945	7q36.3
Holoprosencephaly 4	142946	18p11.31
Holoprosencephaly 5 *	609637	13q32.3
Holoprosencephaly 6	605934	2q37.1-q37.3
Holoprosencephaly 7/ Basal cell nevus/ Gorlin-Goltz	601309/109400	9q22.32
Holoprosencephaly 8	609408	14q13.1-13.2
Holoprosencephaly and preaxial polydactyly *	605651	5q35.1
Holt-Oram syndrome-related microdeletion *	142900	12q24.21
Hunter		Xq28
Hypoparathyroidism sensorineural deafness, renal disease	146255	10p14
Infantile spasm syndrome, X-linked (ISSX) *		Xp21.3
Iridogoniodysgenesis anomaly, Axenfeld-Rieger syndrome	601631	6p25.3
Jacobsen/11q terminal deletion disorder	147791	11q25
Joubert 3	608629	6q23.3
Joubert 4	609583	2q13
Joubert 5 *	610188	12q21.32
Juvenile polyposis syndrome (JPS), BMP1/Related	174900	10q23.2
Juvenile polyposis syndrome (JPS), MADH4-related	174900	18q21.2
Kallmann 1	308700	Xp22.31
Langer- Giedion	150230	8q23.3-q24.11

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Langer mesomelic dysplasia (LMD)	249700	Xpter-Xp22.3/Ypter-Yp11.32
Leber congenital amaurosis X (LCAX) *	610142	12q21.32
Leri-Weill dyschondrosteosis	127300	Xpter-p22.32/Yp11.32
Leukodystrophy with 11q14.2-q14.3		11q14.2-q22.3
Li-Fraumeni 1 (LFS)	151623	17p13.1
Lissencephaly 1	607432	17p13.3
Lissencephaly with cerebellar hypoplasia	257320	7q22.1
Lissencephaly, X-linked (LISX) *	300067	Xq22.3-q23
Loeys-Dietz (LDS), TGFBR2-related *	609192	3p24.1
Lowe	309000	Xq25
Macrocephaly-autism *	605309	10q23.31
Marfan 1 (MFS1) *	154700	15q21.1
Marfan 2 (MFS2) *	154705	3p24.1
Meckel 4 *	611134	12q21.32
Microcephaly, congenital heart disease *	601355	5q35.1
Microphthalmia 3 *	206900	3q26.33
Microphthalmia with linear skin defects	309801	Xp22.2
Microphthalmia, syndromic 7 (MCOPT7)/MLS/MIDAS	309801	Xp22.2
Miller Dieker/ Lissencephaly 1	247200	17p13.3
Mohr-Tranebjaerg	304700	Xq22.1
Monosomy 9p	158170	9p22.3-p23
Mowat-Wilson syndrome *	235730	2q22.3
Mucopolysaccharidosis, type II (MPS2)	309900	Xq28
Nablu mask-like facial	608156	8q21.3-q22.1
Nail-patella *	161200	9q33.3
Nance-Horan	302350	Xp22.13
Nebulette *	605491	10p12.31
Neonatal severe primary hypoparathyroidism(NSHPT)	239200	3q21.1
Nephrophthisis 1 *	258100	2q13
Neurofibromatosis 1/ MR	162200	17q11.2
Neurofibromatosis 2	101000	22q12.2
NFIA Haploinsufficiency *	600727	1p31.3
Noonan 1	163950/176876	12q24.13
Noonan 4 *	610733	2p22.1
Noonan syndrome	163950	3p25.1
Oculocutaneous albinism 2 (OCA2) *	203200	15q13.1
Okhiro *	607323	20q13.2
Opitz	300000	Xp22.2
Ornithine transcarbamylase deficiency *	311250	Xp11.4
Oro-facio-digital 1 (OFD1) *	311200	Xp22.2
Oto-facio-cervical (OFC) *	166780	8q13.3
Pallister-Killian	601803	12p
Pelizaeus-Merzbacher	312080	Xq22.2
Pitt-Hopkins *	610954	18q21.2
Polycystic kidney disease 1 (PKD1)	601313	16p13.3
Potocki-Lupski syndrome	610883	17p11.2
Potocki-Shaffer	601224	11p11.2
Prader-Willi	176270	15q11.2-q12
Prader-Willi-like phenotype *	176270	6q16.3
Proteus/Proteus-like *	176920	10q23.31
PTEN hamartoma tumor *	158350	10q23.31
Pyruvate dehydrogenase deficiency	312170	Xp22.12
Renal cyst and diabetes	137920	17q12
Retinoblastoma/ MR	180200	13q14.2
Rett Syndrome/ MECP2 male duplication	312750	Xq28
Rieger 1 (RIEG1)	180500	4q25
Rubenstein-Taybi	180849	16p13.3
SRY dosage abnormalities	278850/306100	Yp11.31
Sacral/anorectal malformation syndrome		6q25.3
Saethre-Chotzen	101400	7p21.1
Schizencephaly	269160	10q26.11
Schizophrenia & epilepsy *		7q35
Senior-Loken 6 *	610189	12q21.32
Sensorineural deafness and male infertility	611102	15q15.3
Severe myoclonic epilepsy of infancy *	607208	2q24.3
Sex reversal, autosomal dominant 2	154230	9p24.3
Sex reversal, SOX8 related		16p13.3
Short stature, idiopathic, X-linked;see Leri Weill	300582	Xp22.33/Yp11.32

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Simpson-Golabi Behmel *	312870	Xq26.2
Smith-Lemli-Opitz (SLOS) *	270400	11q13.4
Smith-Magenis	182290	17p11.2
Sotos syndrome	117550	5q35.3
Speech delay with FOXP2 deletion	602081	7q31.1
Split hand/foot malformation 1 (SHFM1) *	183600	7q21.3
Split hand/foot malformation 3 (SHFM3)	600095	10q24.32
Split hand/foot malformation 4	605289	3q28
Split hand/foot malformation 5	606708	2q31.1
Steroid sulfatase deficiency *	308100	Xp22.31
Stickler I *	108300	12q13.11
Stocco dos Santos X-Linked Mental Retardation	300434	Xp11.22-p11.21
Synpolydactyly (SPD1)	186000	2q31q32
Synpolydactyly/Syndactyly II	186000	2q31.1
Tetralogy of Fallot *	187500	8q23.1
Thrombocytopenia-Absent Radius syndrome	274000	1q21.1
Timothy syndrome (TS)	601005	12p13.33
Townes-Brocks *	107480	16q12.1
Trichorhinophalangeal 1	190350	8q23.3
Tuberous Sclerosis 1 (TSC1)	191100	9q34
Tuberous sclerosis 2 (TSC2)	191100	16p13.3
Ulnar-Mammary *	181450	12q24.21
Van der Woude syndrome *	119300	1q32.2
Vascular endothelial growth factor related disorders *	192240	6p21.1
Waardenburg syndrome IIA	193510	3p14.1
Waardenburg syndrome type 1	193500	2q36.1
WAGR	194072	11p13
West, Proud, XLAG, Partington syndromes *		Xp21.3
Williams-Beuren	194050	7q11.23
Wilms Tumor	194070	11p13
Wolf-Hirschhorn	194190	4p16.3
XIST deletion on ring X chromosomes	314670	Xq13.2
X-linked chronic granulomatous disease	306400	Xp11.4
X-Linked Cornelia De Lange syndrome	300590	Xp11.22
X-linked dyskeratosis congenita	305000	Xq28
X-linked heterotaxy *	306955	Xq2.3
X-linked hypohidrotic ectodermal dysplasia	305100	Xq13.1
X-linked idiopathic short stature (ISSX)	300582	Xpter-Xp22.3/Ypter-Yp11.32
X-linked infantile spasms *	300203	Xq22.13
X-linked infantile spasms, ARX-related *	308350	Xp21.3
X-linked lissencephaly with ambiguous genitalia *	300215	Xp21.3
X-linked lymphoproliferative syndrome	308240	Xq25
X-Linked Mental Retardation (XLMR)	300075	Xp22.2
X-Linked Mental Retardation (XLMR)	300143	Xp21.2-p21.3
X-Linked Mental Retardation (XLMR) *	300534	Xp11.22
X-Linked Mental Retardation (XLMR) *		Xp11.4
X-Linked Mental Retardation (XLMR)	300578	Xp11.3
X-Linked Mental Retardation (XLMR)		Xq13.1
X-Linked Mental Retardation (XLMR)		Xq13.3
X-Linked Mental Retardation (XLMR)	300354	Xq23
X-Linked Mental Retardation (XLMR) *	312180	Xq25
X-Linked Mental Retardation (XLMR)	300055	Xq28
X-linked Mental Retardation 30 *	300558	Xq22.3
X-linked Mental Retardation 54 *	300419	Xp21.3
X-linked Mental Retardation MRX21	300143	Xp21.3
X-linked recessive chondrodysplasia	302950	Xq22.33
XLMR retinitis pigmentosa	300578	Xp11.3
Xp11.3 microdeletion	300578	Xp11.3
Xp22.31 duplication; reciprocal STS *		Xp22.31
XX male	278850	Yp11.31
XY gonadal dysgenesis	306100	Yp11.31
>70 Autism Spectrum Disorder- associated loci		
All 41 unique subtelomeric regions		
All 43 unique pericentromeric regions		
Aneuploidy for 24 chromosomes		

\* Best detected by Oligo HD Scan