

Nr	OMIM#*	PMID	Loci	Associated Abnormalites detected by CMA	Cytogenetic Abnormalities	Genes
1				Subtelomeric deletion/ duplication for all chromosome arms	All subtelomeres exluding the short arm of the acrocentric chromosomes	
2				Marker chromosomes that contain a centromere	All pericentromeric regions	
3				Aneuploidy for chromosomes 13,18, 21, X, and Y		
4			1p36	1p36 deletion and/or duplication/ triplication	1p36 deletion and/or duplication	
5	606606		1q25.2	Short stature, Pituitary and cerebellar defects	1q25 deletion	<i>LHX4</i>
6	119300		VWS	Van der Woude syndrome (VWS)	1q32-q41 deletion	<i>IRF6</i>
7	164280		2p24.1	Feingold Syndrome	2p24.1 deletion	<i>MYCN</i>
8	603714		HPE2	Holoprosencephaly 2	2p21 deletion	<i>SIX3</i>
9	256100		NPH1	Nephronophthisis 1	2q13 homozygous deletion	<i>NPHP1</i>
10	165230	14581620	2q14.2	Pituitary Anomalies with Holoprosencephaly	2q14 deletion	<i>GLI2</i>
11	235730		2q22.3	Mowat-Wilson syndrome	2q22.3 deletion	<i>ZFXH1B</i>
12	186000	10364522	SPD1	Synpolydactyly (SPD1)	2q31-q32 deletion	<i>HOXD13</i>
13	119540	16990542	CPI	Cleft Palate, Isolated (CPI)	2q32-q33 deletion	<i>SATB2, SUMO1</i>
14	190182	15235604	3p24.1	HNPCC6	3p24.1 deletion	<i>TGFBR2</i>
		15731757				
15	187395	12073012	3p21.31	Forebrain defects	3p23-p21 deletion	<i>TDGF1 (CRIPTO)</i>
16	193510		WS2A	Waardenburg syndrome, type IIA (WS2A)	3p14.2-p12.3	<i>MITF1</i>
17	110100	15962237	BPES	Blepharophimosis (BPES)	3q23 deletion	<i>FOXL2</i>
18	220200		DWS	Dandy-Walker syndrome	3q24 deletion	<i>ZIC1 ZIC4</i>
19	206900		MCOPS3	Microphthalmia, Syndromic 3 (MCOPS3)	3q26.3-q27	<i>SOX2</i>
20	605289		SHFM4	Split hand/foot malformation 4 (SHFM4)	3q27 deletion	<i>TP73L</i>
21	248250		CLDN16	Hypomagnesemia, primary	3q27	<i>CLDN16</i>
22	605022		3q29	3q29 Microdeletion syndrome	3q29 Microdeletion	<i>PAK2, DLG1</i>
		601014				
23	194190		WHS	Wolf-Hirschhorn syndrome	4p16.3 deletion	
24	180500		RIEG1	Rieger syndrome	4q25 deletion	<i>PITX2</i>
25	123450		CdCS	Cri-du-Chat syndrome 5p15.2	deletion	
26	122470		CdLS	Cornelia de Lange syndrome	5p13.2 deletion	<i>NIPBL</i>
27		15742475	5q21.1-q31.2	5q21.1-q31.2 deletion syndrome	5q21.1-q31.2 deletion	<i>APC</i>
		16365682				
		16470790				
28	154500		TCOF	Treacher-Collins Syndrome (TCOF)	5q32 deletion	<i>TCOF1</i>
29	600584	16470726	5q34-q35.1	Microcephaly, Congenital heart disease (CHD)	5q34-q35.1 deletion	<i>NKX2-5</i>
30	117550		SOS	Sotos syndrome	5q35 deletion	<i>NSD1</i>
31	201910		CAH	Congenital Adrenal Hyperplasia (CAH) due to 21-alpha hydroxylase deficiency	6p21-p21.32 deletions	<i>CYP21A2</i>
32	192240		6p21.1	VEGF	6p21.1 deletion	<i>VEGF</i>
33	119600		CCD	Cleidocranial dysplasia	6p21.1 deletion	<i>RUNX2</i>
34	603128		6q16.3	SIM1	6q16-21.1 deletion	<i>SIM1</i>
35	601410		TNDM1	Transient Neonatal diabetes mellitus 1	6q24-q24.2 duplication	<i>PLAGL1/ZAC1</i>
36	101400		SCS	Saethre-Chatzen syndrome	7p21.1 deletion	<i>TWIST</i>
37	175700		GCPS	Greig cephalopolysyndactyly syndrome	7p13 deletion	<i>GLI3</i>
38	194050		WBS	Williams-Beuren syndrome (WBS)	7q11.23 deletion	<i>ELN, LIMK1</i>

39	220600		SHFM1	Split Hand/Foot malformation w. sensorineural hearing loss (Split/hand/foot malformation 1 (SHFM1)	7q21.2-q21.3	<i>SHFM!</i>
	183600					
40	120160		7q21.3	Collagen alpha 2(I) chain	7q21.3 deletions/duplication	<i>COL1A2</i>
41	605317		7q31.1	Speech delay	7q31.1 deletion	<i>FOXP2</i>
42	142945		HPE3	Holoprosencephaly 3	7q36.3 deletion	<i>SHH</i>
43	607941	10096597	8p22-p23.1	8p22 duplication/deletion syndrome	8p22-p23.1 duplication/deletion	<i>GATA4</i>
44	214800		8q12.2	CHARGE syndrome	8q12.2 deletion	<i>CHD7</i>
45	113650		BOR	Branchiootorenal dysplasia syndrome (BOR)	8q13.3 deletion	<i>EYA1</i>
46	602064	14699425	8q21.13	Bipolar disorder	8q21.13 deletion	<i>IMPA1</i>
47	150230		LGS	Langer-Giedion syndrome	8q24.11-q24.13 deletion	<i>TRPS1 and/or EXT1</i>
48	190350		TRPS1	Trichorhinophalangeal syndrome (TRPS), Type 1	8q24.12 deletion	<i>TRPS1</i>
49	154230		SRA2	Sex reversal, Autosomal dominant 2	9p24.3 deletion	<i>TDFA (SRA2)</i>
50		16570072	9p23-p22.3	9p23 microdeletion syndrome, trigonocephaly, overgrowth and mental retardation	9p23 deletion	
		15857417				
51			<i>HSH</i>	Hypomagnesemia with secondary hypocalcemia (HOMG)	<i>9q22 deletion</i>	<i>TRPM6</i>
52	268310		9q22.31	Robinow syndrome	9q22.31 deletion	<i>ROR2</i>
53	113000			Brachydactyly, type B1 (BDB1)		
54	109400		BCNS	Basal cell naevus syndrome (Gorlin syndrome)	9q22.32 deletion	<i>PTCH</i>
55	609192		LDS	Loeys-Dietz syndrome (LDS)	9q22.3 deletion	<i>TGFBR1</i>
56	184757	15546904	ACTs	Adrenocortical tumors (ACTs)	9q33 duplication	<i>NR5A1 (SF1)</i>
57	161200		NPS	Nail-Patella syndrome	9q34.1 deletion	<i>LMX1B</i>
58		16826528	9q34.3	9q34.3 microdeletion	9q34 deletion	<i>EHMT1</i>
59	146255		HDR	Hypoparathyroidism, sensorineural deafness & renal dysplasia	10p14-p15.1 deletion	<i>GATA3</i>
60	601362		DGS2	DiGeorge syndrome 2	10p13-p14 deletion	
61	158350		CD	Cowden syndrome (CD)	10q23.31	<i>PTEN</i>
62	153480		BZS	Bannayan-Zonana syndrome (BZS)		
63	608071		SHFM3	Split hand/split foot syndrome 3	10q24 duplication	<i>FBXW4 (SHSF3)</i>
	600095					
64	130650		BWS	Beckwith-Wiedemann syndrome (BWS)	11p15.5 duplication/deletion	<i>IGF2, LIT1, p57, H19, KCNQ1, CDKN1C</i>
65						<i>CALC1, CALC2</i>
66	114130		11p15.2	Behavioral Problems and Autistic Spectrum	11p15.2 deletion	
	114160					
67	607257	16258006	11p15.2-p15.3	Craniosynostosis	11p15 deletion	<i>SOX6</i>
68	106210		AN2	Aniridia type 2	11p13 deletion	<i>PAX6</i>
69	607102		WT1	Wilm's tumor 1 gene	11p13 deletion	<i>WT1</i>
70	194072		WAGR	Wilm's tumor-aniridia-genitourinary abnormalities (WAGR) syndrome	11p13 deletion	<i>WT1</i>
71	601224		PSS	Potocki-Shaffer syndrome	11p11.2 deletion	<i>ALX4 and/or EXT2</i>
72	252010		11q13.2	Mitochondrial Complex 1 deficiency	11q13.3 duplication	<i>NDUFV1</i>
73		14722582	11q14.2	Leukodystrophy	11q14.2-q14.3 deletion	
74	147791	15857417	JBS	Jacobsen syndrome	11q23-q25 deletion	
75	120140	15362574	12q13.11	Collagen, Type II, Alpha-1	12q13.11 deletion	<i>COL2A1</i>
76	142900		HOS	HOLT-ORAM SYNDROME; HOS	12q24.1 deletion	<i>TBX5</i>
77	181450		UMS	ULNAR-MAMMARY SYNDROME; UMS	12q24.1 deletion	<i>TBX3</i>

78	163950	7747795	NS1	Noonan syndrome	12q24.13 deletion	<i>PTPN11</i>
79		16760730	12q24	12q24 microduplication syndrome	12q24.21-q24.23 duplication	<i>THRAP2, NOS1, RFC5</i>
		16411218				<i>RB1</i>
80	180200		RB1	Retinoblastoma	13q14 deletion	<i>GPC5, GPC6</i>
81	602446		13q31.3	Brachydactyly	13q32 deletion	
82	604404		13q32			
83	603073		HPE5	Holoprosencephaly 5	13q32.3 deletion	<i>ZIC2</i>
84	164874		14q12	Forkhead box G1B	14q12 deletion	<i>FOXC1B</i>
85	112262		14q22.2-q22.3	Anophthalmia, Pituitary hypoplasia	14q22.2-q22.3 deletion	<i>BMP4</i>
86	176270		PWS	Prader Willi syndrome (PWS) Paternal	Paternal 15q11.2-q13 deletion	<i>SNRPN</i>
87	105830		AS	Angelman syndrome (AS) Maternal	Maternal 15q11.2-q13 deletion	<i>UBE3A</i>
88	209850		AUTS4	Autism	Maternal 15q11.2-q13 duplication	
89	154700		MFS	Marfan syndrome (MFS)	15q21.1 deletion	<i>FBN1</i>
90	102578	16199540	15q22	PML	15q22 deletion	<i>PML</i>
91	142340		HCD	Congenital diaphragmatic hernia	15q26.1-q26.2 deletion	<i>DIH1</i>
92	147370	1849352	15q26.3	Severe intrauterine growth restriction (IUGR)	15q26.3 deletion	<i>IGF1R</i>
93	141750		ATR	Alpha thalassemia/MR syndrome (ATR)	16pter-p13.3 deletion	<i>HBA1, HBA2</i>
94	605923	15949865	16p13.3	Sex reversal	16p13.3 deletion	<i>SOX8</i>
95	191092		TS 2	Tuberous Sclerosis 2	16p13.3 deletion	<i>TSC2</i>
96	600273		PKDTS	Polycystic kidney disease/tuberous sclerosis 2	16p13.3 deletion	<i>PKD1 and/or TSC2</i>
97	180849		RSTS	Rubinstein-Taybi syndrome	16p13.3 deletion	<i>CREBBP</i>
98	107480		TBS	Townes-Brocks syndrome, TBS	16q12.1 deletion	<i>SALL1</i>
99	247200		MDLS	Miller-Dieker lissencephaly syndrome	17p13.3 deletion	<i>LIS1 and/or YWHAE</i>
100	219800		CTNS	Cystinosis, Nephropathic (CTNS)	17p13.3-p.13.2 deletion	<i>CTNC</i>
101	118220		CMT1A	Charcot-Marie-Tooth disease type 1A	17p12 duplication	<i>PMP22</i>
102	162500		HNPP	Hereditary neuropathy with liability to pressure palsies	17p12 deletion	<i>PMP22</i>
103	182290		SMS	Smith-Magenis syndrome	17p11.2 deletion	<i>RAI1</i>
104		10615134	17p11.2	dup(17)(p11.2p11.2) syndrome	17p11.2 duplication	
105	162200		NF1	Neurofibromatosis 1	17q11.2 deletion	<i>NF1</i>
106	137920		RCAD	Renal cysts and diabetes (RCAD)	17q21.3 deletion	<i>TCF2</i>
107	122561	16906164	17q21.31	17q21.31 microdeletion syndrome	17q31.31 deletion	<i>CRHR1, MAPT</i>
	157140	16906163				
108	120150		17q21.33	Collagen, type I, alpha-1	17q21.33 deletion	<i>COL1A1</i>
109	114290		CMPD	Campomelic dysplasia	17q24.3 deletion	<i>SOX9</i>
110	142946		HPE4	Holoprosencephaly 4	18p11.31 deletion	<i>TGIF</i>
111	223800		DMC	Gyggve Melchior-Clausen syndrome	18q21.1 deletion	<i>DYM</i>
112	608696		20p12.2	Coloboma	20p12.2 deletion	<i>SNAP25</i>
113	118450		ALGS1	Alagille syndrome 1 (ALGS1)	20p12.2 deletion	<i>JAG1</i>
114	113100		BDC	Brachydactyly, type C (BDC)	20q11.2 deletion	<i>GDF5</i>
115	607323		DRRS	Duane-Radial Ray syndrome, Okihiro syndrome	20q13.13-q13.2	<i>SALL4</i>
116	190685		DS	Down syndrome critical region	21q22 duplication	
117	236100		HPE1	Holoprosencephaly 1	21q22.3 deletion	<i>TMEM1, LSS</i>
118	115470		CES	Cat eye syndrome	inv dup(22q11.2)	
119	600237		22q11.2	DiGeorge syndrome critical region gene 1	22q11.2 deletion	<i>TUPLE1 (DGCR1)</i>
120	192430		VCFS	Velocardiofacial syndrome / DiGeorge syndrome 1	22q11.2 deletion	<i>TBX1</i>

	188400	DGS1				
121		15800846	22q11.2	dup(22)(q11.2q11.2) syndrome	22q11.2 duplication	
122	607379	NF2		Neurofibromatosis 2	22q12.2 deletion	<i>NF2</i>
123	127300	LWD		Leri-Weill dyschondrosteosis	Xp22.33/Yp11.32 deletion	<i>SHOX</i>
124	300495	12669065	AUTSX2	Autism, X-linked, susceptibility to, 2	Xp22.32 deletion	<i>NLGN4</i>
		14963808				
125	308100	STS		Steroid sulfatase deficiency	Xp22.31 deletion	<i>STS</i>
126	308700	KMS		Kallmann syndrome 1	Xp22.31 deletion	<i>KAL1</i>
127	309801	MLS		Micophthalmia with linear skin defects	Xp22.2 deletion	
128	300000	Xp22.2		Opitz syndrome	Xp22.2 deletion	<i>MID1</i>
129	311200	OFD1		Orofaciodigital syndrome (OFD1)	Xp22.2 deletion	<i>CXORF5</i>
130	312700	RS1		X-linked juvenile retinoschisis (RS1)	Xp22.13 deletion	<i>RS1</i>
131	312750	RTT		Rett syndrome, Infantile spasms (RTT)	Xp21.3-p22.1 deletion	<i>STK9 (CDKL5)</i>
132	308350	12379852	ISSX	Infantile spasms (ISSX)	Xp21.3-p22.11 deletion	<i>ARX</i>
133	300143	14610352	MRX21	X-linked mental retardation (MRX21)	Xp21.3 deletion	<i>IL1RAPL1</i>
134	300200	AHC		Congenital adrenal hypoplasia sex reversal	Xp21.2 deletion/duplication	<i>NROB1 (DAX1)</i>
135	300474	GKD		Glycerol kinase deficiency	Xp21 deletion	<i>GK</i>
136	311250	2983225	Xp21.1	Ornithine transcarbamylase deficiency	Xp21.1 deletion (rare)	<i>OTC</i>
137	300573	16385466	Xp11.3	X-linked mental retardation	Xp11.3 deletion (rare)	<i>ZNF674</i>
138	300590	CdLS		X-linked Cornelia de Lange syndrome (CdLS)	Xp11.2 deletion	<i>SMC1L1</i>
139	300068	AIS		Androgen insensitivity syndrome (AIS)	Xq12 deletion	<i>AR</i>
140	314670	Xq13.2-q13.3		X-inactivation specific transcript	Xq13.2-q13.3	<i>XIST</i>
141	309580	Xq13.2-q21.1		X-linked mental retardation / Hypotonic Facies syndrome	Xq13.2-q21.1	<i>ATRX</i>
142	309400	12485192	MNK	Menkes Disease (MNK)	Xq12 deletion	<i>ATP7A</i>
143	307200	Xq22.1		Hypogammaglobulinemia, Isolated growth hormone	Xq22.1 deletion	<i>BTK</i>
144	300300			Deficiency		
145	312080	PMD		Pelizaeus-Merzbacher disease	Xq22 duplication or deletion	<i>PLP1</i>
146	301050	ATS		X-linked Alport syndrome (ATS)	Xq22.3 deletion	<i>Col4A5</i>
147	300067	LISX		X-linked Lissencephaly (LISX)	Xq22.3-q23 deletion	<i>DCX</i>
148	308240	XLP		X-linked Lymphoproliferate syndrome (XLP)	Xq25 deletion	<i>SH2D1A</i>
149	300322	LNS		Lesch-Nyhan syndrome (LNS)	Xq26-26.3 deletion	<i>HPRT</i>
150	306955	HTX		X-Linked heterotaxy	Xq26.3 deletion	<i>ZIC3</i>
151	300123	MRGH		Mental retardation X-linked	Xq27.1 deletion or duplication	<i>SOX3</i>
152	309900	MPS2		Mucopolysaccharidosis, type II (MSP2)	Xq28 deletion	<i>IDS</i>
153	300123	XLMR		Mental retardation X-linked	Xq28 duplication	<i>MECP2</i>
154	312750	RTT		Rett syndrome	Xq28 deletion	<i>MECP2</i>
155	300321	Xq28		FG syndrome 2	Xq28 deletion	<i>FGS2</i>
156	300133	VBP1		Von Hippel-Lindau binding protein 1 (VBP1)	Xq28 deletion	<i>VBP1</i>
157	306100	GDXY		Gonadal Dygenesis, XY female Type (GDXY)	Yp11.31 deletion	<i>SRY</i>
158	415000	AZFa		Azospemia factor a	Yq11 deletion	<i>AZFa</i>
159		AZFb		Azospemia factor b	Yq11 deletion	<i>AZFb</i>