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	*LHX4*
6	119300		VWS	Van der Woude syndrome (VWS)	1q32-q41 deletion	*IRF6*
7	164280		2p24.1	Feingold Syndrome	2p24.1 deletion	*MYCN*
8	603714		HPE2	Holoprosencephaly 2	2p21 deletion	*SIX3*
9	256100		NPH1	Nephronophthisis 1	2q13 homozygous deletion	*NPHP1*
10	165230	14581620	2q14.2	Pituitary Anomalies with Holoprosencephaly	2q14 deletion	*GLI2*
11	235730		2q22.3	Mowat-Wilson syndrome	2q22.3 deletion	*ZFHX1B*
12	186000	10364522	SPD1	Synpolydactyly (SPD1)	2q31-q32 deletion	*HOXD13*
13	119540	16990542	CPI	Cleft Palate, Isolated (CPI)	2q32-q33 deletion	*SATB2, SUMO1*
14	190182	15235604 15731757	3p24.1	HNPCC6	3p24.1 deletion	*TGFBR2*
15	187395	12073012	3p21.31	Forebrain defects	3p23-p21 deletion	*TDGF1 (CRIPTO)*
16	193510		WS2A	Waardenburg syndrome, type IIA (WS2A)	3p14.2-p12.3	*MITF1*
17	110100	15962237	BPES	Blepharophimosis (BPES)	3q23 deletion	*FOXL2*
18	220200		DWS	Dandy-Walker syndrome	3q24 deletion	*ZIC1 ZIC4*
19	206900		MCOPS3	Microphthalmia, Syndromic 3 (MCOPS3)	3q26.3-q27	*SOX2*
20	605289		SHFM4	Split hand/foot malformation 4 (SHFM4)	3q27 deletion	*TP73L*
21	*248250*		CLDN16	Hypomagnesemia, primary	*3q27*	*CLDN16*
22	605022 601014		3q29	3q29 Microdeletion syndrome	3q29 Microdeletion	*PAK2, DLG1*
23	194190		WHS	Wolf-Hirschhorn syndrome	4p16.3 deletion	
24	180500		RIEG1	Rieger syndrome	4q25 deletion	*PITX2*
25	123450		CdCS	Cri-du-Chat syndrome 5p15.2	deletion	
26	122470		CdLS	Cornelia de Lange syndrome	5p13.2 deletion	*NIPBL*
27		15742475 16365682 16470790	5q21.1-q31.2	5q21.1-q31.2 deletion syndrome	5q21.1-q31.2 deletion	*APC*
28	154500		TCOF	Treacher-Collins Syndrome (TCOF)	5q32 deletion	*TCOF1*
29	600584	16470726	5q34-q35.1	Microcephaly, Congenital heart disease (CHD)	5q34-q35.1 deletion	*NKX2-5*
30	117550		SOS	Sotos syndrome	5q35 deletion	*NSD1*
31	201910		CAH	Congenital Adrenal Hyperplasia (CAH) due to 21-alpha hydroxylase deficiency	6p21-p21.32 deletions	*CYP21A2*
32	192240		6p21.1	VEGF	6p21.1 deletion	*VEGF*
33	119600		CCD	Cleidocranial dysplasia	6p21.1 deletion	*RUNX2*
34	603128		6q16.3	SIM1	6q16-21.1 deletion	*SIM1*
35	601410		TNDM1	Transient Neonatal diabetis mellitus 1	6q24-q24.2 duplication	*PLAGL1/ZAC1*
36	101400		SCS	Saethre-Chotzen syndrome	7p21.1 deletion	*TWIST*
37	175700		GCPS	Greig cephalopolysyndactyly syndrome	7p13 deletion	*GLI3*
38	194050		WBS	Williams-Beuren syndrome (WBS)	7q11.23 deletion	*ELN, LIMK1*

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

78	163950	7747795	NS1	Noonan syndrome	12q24.13 deletion	*PTPN11*
79		16760730 16411218	12q24	12q24 microduplication syndrome	12q24.21-q24.23 duplication	*THRAP2, NOS1, RFC5*
80	180200		RB1	Retinoblastoma	13q14 deletion	*RB1*
81	602446		13q31.3	Brachydactyly	13q32 deletion	*GPC5, GPC6*
82	604404		13q32			
83	603073		HPE5	Holoprosencephaly 5	13q32.3 deletion	*ZIC2*
84	164874		14q12	Forkhead box G1B	14q12 deletion	*FOXG1B*
85	112262		14q22.2-q22.3	Anophthalmia, Pituitary hypoplasia	14q22.2-q22.3 deletion	*BMP4*
86	176270		PWS	Prader Willi syndrome (PWS) Paternal	Paternal 15q11.2-q13 deletion	*SNRPN*
87	105830		AS	Angelman syndrome (AS) Maternal	Maternal 15q11.2-q13 deletion	*UBE3A*
88	209850		AUTS4	Autism	Maternal 15q11.2-q13 duplication	
89	154700		MFS	Marfan syndrome (MFS)	15q21.1 deletion	*FBN1*
90	102578	16199540	15q22	PML	15q22 deletion	*PML*
91	142340		HCD	Congenital diaphragmatic hernia	15q26.1-q26.2 deletion	*DIH1*
92	147370	1849352	15q26.3	Severe intrauterine growth restriction (IUGR)	15q26.3 deletion	*IGF1R*
93	141750		ATR	Alpha thalassemia/MRsyndrome (ATR)	16pter-p13.3 deletion	*HBA1, HBA2*
94	605923	15949865	16p13.3	Sex reversal	16p13.3 deletion	*SOX8*
95	191092		TS 2	Tuberous Sclerosis 2	16p13.3 deletion	*TSC2*
96	600273		PKDTS	Polycystic kidney disease/tuberous sclerosis 2	16p13.3 deletion	*PKD1 and/or TSC2*
97	180849		RSTS	Rubinstein-Taybi syndrome	16p13.3 deletion	*CREBBP*
98	107480		TBS	Townes-Brocks syndrome, TBS	16q12.1 deletion	*SALL1*
99	247200		MDLS	Miller-Dieker lissencephaly syndrome	17p13.3 deletion	*LIS1 and/or YWHAE*
100	219800		CTNS	Cystinosis, Nephropathic (CTNS)	17p13.3-p.13.2 deletion	*CTNC*
101	118220		CMT1A	Charcot-Marie-Tooth disease type 1A	17p12 duplication	*PMP22*
102	162500		HNPP	Hereditary neuropathy with liability to pressure palsies	17p12 deletion	*PMP22*
103	182290		SMS	Smith-Magenis syndrome	17p11.2 deletion	*RAI1*
104		10615134	17p11.2	dup(17)(p11.2p11.2) syndrome	17p11.2 duplication	
105	162200		NF1	Neurofibromatosis 1	17q11.2 deletion	*NF1*
106	137920		RCAD	Renal cysts and diabetes (RCAD)	17q21.3 deletion	*TCF2*
107	122561 157140	16906164 16906163	17q21.31	17q21.31 microdeletion syndrome	17q31.31 deletion	*CRHR1, MAPT*
108	120150		17q21.33	Collagen, type I, alpha-1	17q21.33 deletion	*COL1A1*
109	114290		CMPD	Campomelic dysplasia	17q24.3 deletion	*SOX9*
110	142946		HPE4	Holoprosencephaly 4	18p11.31 deletion	*TGIF*
111	223800		DMC	Gyggve Melchior-Clausen syndrome	18q21.1 deletion	*DYM*
112	608696		20p12.2	Coloboma	20p12.2 deletion	*SNAP25*
113	118450		ALGS1	Alagille syndrome 1 (ALGS1)	20p12.2 deletion	*JAG1*
114	113100		BDC	Brachydactyly, type C (BDC)	20q11.2 deletion	*GDF5*
115	607323		DRRS	Duane-Radial Ray syndrome, Okihiro syndrome	20q13.13-q13.2	*SALL4*
116	190685		DS	Down syndrome critical region	21q22 duplication	
117	236100		HPE1	Holoprosencephaly 1	21q22.3 deletion	*TMEM1, LSS*
118	115470		CES	Cat eye syndrome	inv dup(22q11.2)	
119	600237		22q11.2	DiGeorge syndrome critical region gene 1	22q11.2 deletion	*TUPLE1 (DGCR1)*
120	192430		VCFS	Velocardiofacial syndrome / DiGeorge syndrome 1	22q11.2 deletion	*TBX1*

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