Genetic Disorders	OMIM	Gene(s)/Locus	Detection Rate	Location
1p36 deletion syndrome	607872	multiple	>95%	1p36
1q21.1 deletion/duplication	612474	multiple	>95%	1q21.2
1q41-q42 microdeletion	612530	multiple	>95%	1q41-q42
2q22.3 deletion syndrome	-	multiple	>95%	2q22.3
2q37 deletion syndrome	605314	*HDAC4*	>95%	2q37
3q29 microdeletion syndrome	609425	multiple	>95%	3q29
3q29 microduplication syndrome	611936	multiple	>95%	3q29
5q21.1-q31.2 deletion syndrome	-		>95%	5q21.1-q31.2
7q11.23 duplication (Williams-Beuren region)	609757	multiple	>95%	7q11.23
8p23.1 deletion syndrome	-		>95%	8p23.1
9q34.3 deletion syndrome	610253	*EHMT1*		9q34.3
11q11-q13.3 duplication	164950; 164980	*FGF3, FGF4*	>95%	11q11-q13.3
15q24 microdeletions syndrome	613406	*HCN4*	>95%	15q24
16p11.2-p12.2 deletion syndrome	611913; 613604	multiple	>95%	16p11.2-p12.1
17p11.2 duplication; reciprocal SMS region	-	*RAI1*	>95%	17p11.2
17q21.31 microdeletion syndrome	610443	*MAPT, CRHR1, KANSL1*	>95%	17q21.31
17q21.31 microduplication syndrome	613533	multiple	>95%	17q21.31
22q11.2 duplication;reciprocal DGS1 region	608363	*TBX1*	>95%	22q11.2
22q13.3 microdeletion syndrome	606232	*SHANK3*	>95%	22q13.3
Adrenal Hypoplasia Congenita (AHC), X-Linked	300200	*NROB1*	~18%	Xp21.2
Alagille sydrome (AGS)	118450	*JAG1*	3-7%	20p12.2
Alpha Thalassemia, Mental Retardation syndrome	301040, 141750	*ATRX, SOX8*	Unknown	Xq21.1, 16p13.3
Alport syndrome, X-linked (ATS)	301050	*COL4A5*	~5%	Xq22.3
Androgen insensitivity syndrome (AIS)	300068	*AR*	Very low	Xq12
Aneuploidy (for 24 chromosomes)		multiple	>95%	24 chromosomes
Angelman syndrome (AS)	105830	*UBE3A*	~70%	15q11.2-q13
Aniridia II & WAGR syndrome	106210	*PAX6*	20-35%	11p13
Bannayan-Riley-Ruvalcaba syndrome (BRRS)	153480	*PTEN*	~2-11%	10q23.31
Basal cell nevus syndrome (BCNS)	109400	*PTCH1*	6-21%	9q22.32
Beckwith-Wiedemann syndrome (BWS)	130650	*IGF2, KCNQ1*	Unknown	11p15.5
Blepharophimosis, ptosis, epicanthus inversus (BPES)	110100	*FOXL2*	unknown	3q22.3
Branchiootorenal dysplasia syndrome (BOR)/Melnick-Frazer Syndrc	113650/166780	*EYA1*	7-11%	8q13.3
Bruton agammaglobulinemia (hypogammaglobulinemia)	300755	*BTK*	~8%	Xq22.1
Campomelic dysplasia (CMPD)	114290	*SOX9*	Low	17q24.3
Cardio-Facio-Cutaneous syndrome	115150	*BRAF*	Unknown	7q34
Cat-eye syndrome (CES)	115470	multiple	Supernumary marker chromosome is detectable	22q11.21
Charcot-Marie-Tooth disease type 1A (CMT1A)	118220	*PMP22*	>95%	17p12

CHARGE syndrome	214800	*CHD7*	~10%	8q12.2
Chromosome 10q22.3-q23.31 microdeletion	-		>95%	10q22.3-q23.31
Chromosome 18p deletion syndrome	146390	multiple	>95%	18p
Chromosome 18q deletion syndrome	601808	multiple	>95%	18q
Cleidocranial dysplasia (CCD)	119600	*RUNX2*	~10%	6p21.1
Cornelia de Lange syndrome (CDLS)	122470	*NIPBL*	Deletions uncommon	5p13.2
Cowden syndrome (CD)	158350	*PTEN*	~1% have a detectable deletion	10q23.31
Cri-du-Chat syndrome	123450	multiple	>95%	5p15.2-p15.33
Cutis laxa, X-linked/ Occipital horn syndrome	304150	*ATP7A*	~15%	Xq21.1
Cystinosis, Nephropathic (CTNS)	219800	*CTNS*	~50%	17p13.2
Dandy-Walker syndrome (DWS)	220200	*ZIC1, ZIC4*	Deletions uncommon	3q22-q24
Diaphragmatic hernia, congenital (HCD/DIH1)	142340	*NR2F2, CHD2*	~1-2%	15q26.1-q26.3
DiGeorge syndrome 1 (DGS1)/Velocardiofacial (VCFS)	188400	*TBX1*	>95%	22q11.21
DiGeorge syndrome 2 (DGS2)	601362	multiple	<1%	10p13-14
Down syndrome critical regions (DSCR)	602917	multiple	Unknown	21q22.12
Duchenne muscular dystrophy (DMD)	310200	*DMD*	50-65%	Xp21.1-p21.2
Dyggve-Melchior-Clausen syndrome (DMC)	223800	*DYM*	Deletions uncommon	18q21.1
Feingold syndrome	164280	*MYCN*	~10%	2p24.3
Forebrain defects	187395	*TDGF1(CRIPTO)*	Unknown	3p21.31
Fragile-X mental retardation syndrome (FMR1)	300624	*FMR1*	<1%	Xq27.3
Glycerol kinase deficiency (GKD)	300474	*GK*	Deletions uncommon	Xp21.2
Greig cephalo-polysyndactyly syndrome (GCPS)	175700	*GLI3*	5-10%	7p14.1
Hereditary Neuropathy with Liability to Pressure Palsies	162500	*PMP22*	~80%	17p12
Holoprosencephaly 1 (HPE1)	236100	TRAPPC10, PWP2	>95%	21q22.3
Holoprosencephaly 2 (HPE2)	157170	*SIX3*	~1%	2p21
Holoprosencephaly 3 (HPE3)	142945	*SHH*	1-2%	7q36.3
Holoprosencephaly 4 (HPE4)	142946	*TGIF1*	<1%	18p11.31
Holoprosencephaly 5 (HPE5)	609637	*ZIC2*	<1%	13q32.3
Holoprosencephaly 6 (HPE6)	605934	*HPE6*	Unknown	2q37.1-q37.3
Holoprosencephaly 7 (HPE7)	610828	*PTCH1*	Unknown	9q22.32
Holoprosencephaly 9/ Pituitary anomalies with Holoprosencephaly	610829	*GLI2*	Unknown	2q14.2
Infantile spasm syndrome, X-linked (ISSX)	300419	*ARX*	Deletion uncommon	Xp21.3
Infantile spasm syndrome, X-linked (ISSX)	300672	*CDKL5*	Unknown	Xp22.13
Jacobsen Syndrome	147791	multiple	Majority of reported deletions are detectable by array CGH	11q23-q25

Joubert syndrome 4	609583	*NPHP1*	Unknown	2q13
Kallman syndrome 1 (KAL1)	308700	*KAL1*	10-12%	Xp22.31
Langer-Giedion syndrome (LGS)	150230	*EXT1, TRPS1*	~75%	8q23.3-q24.11
Lesch-Nyhan syndrome (LNS)	300322	*HPRT1*	21-24%	Xq26.2-q26.3
Leukodystrophy with 11q14.2-q14.3			Rare	11q14.2-q22.3
Lissencephaly, X-linked (LISX)	300067	*DCX*	Unknown	Xq23
Lissencephaly 1	607432	*PAFAH1B1*	~27%	17p13.3
MECP2 duplication syndrome	300260	*MECP2*	>95%	Xq28
Menkes disease (MNK)	309400	*ATP7A*	~1%	Xq21.1
Microphthalmia with linear skin defects	309801	*HCCS*	Majority of reported deletions are detectable by array CGH.	Xp22.2
Microcephaly, congenital heart disease	108900	*NKX2-5*	Uncertain	5q35.1
Microdeletion 2p15-p16.1	612513	*MTIF2, VRK2*	>95%	2p15-p16.1
Microphthalmia syndrome 6, pituitary hypoplasia	607932	*BMP4*		14q22.2
Microphthalmia, syndrome 3	206900	*SOX2*	~2%	3q26.33
Microphthalmia, syndromic 7	309801	multiple	Uncertain	Xp22.2
Miller-Dieker Lissencephaly syndrome (MDLS)	247200	*YWHAE, LIS1*	85-90%	17p13.3
Monosomy 9p syndrome	158170	multiple	>95%	9p
Mowat-Wilson syndrome	235730	*ZEB2*	~15%	2q22.3
Mucopolysaccharidosis, type II (MPS2)	309900	*IDS*	~9%	Xq28
Nail-patella syndrome (NPS)	161200	*LMX1B*	~5%	9q33.3
Nephronophthisis 1 (NPH1)/2q13 duplication	256100	*NPHP1*	60-70% homozygous deletion	2q13
Noonan syndrome (NS1)	163950	*PTPN11*	Precise detection rate unknown	12q24.13
Opitz GBBB, X-linked	300000	*MID1*	~2%	Xp22.2
Orofaciodigital syndrome	311200	*OFD1*	~5%	Xp22.2
Pallister-Killian	601803	multiple	unknown (tetrasomy 12p is detectable)	12p
Pelizaeus-Merzbacher disease (PMD)	312080	*PLP1*	50-75%	Xq22.2
Pericentromeric regions (43 regions)	-	multiple	Detection rate variable depending upon the marker chromosome 43 sites	
Potocki-Shaffer syndrome	601224	*EXT2, ALX4*	>95%	11p11.2
Prader-Willi syndrome (PWS)	176270	multiple	~70%	15q11.2-q13
Prader-Willi-like phenotype	176270	*SIM1*	Deletions uncommon	6q16.1-q16.3
Rett syndrome (RTT)	312750	*MECP2*	3-8%	Xq28
Rieger syndrome, type 1 (RIEG1)	180500	*PITX2*	Precise detection rate unknown	4q25
Rubinstein-Taybi syndrome (RSTS)	180849	*CREBBP*	10-20%	16p13.3

Sacral/anorectal malformation syndrome		multiple		6q25.3
Saethre-Chotzen syndrome (SCS)	101400	*TWIST1*	11-28%	7p21.1
Smith-Magenis syndrome (SMS)	182290	*RAI1*	>95%	17p11.2
Sotos syndrome (SOS)	117550	*NSD1*	10-15% (50% in Japanese)	5q35.2-q35.3
Split-hand/foot malformation-1 (SHFM1)	183600	*SHFM1*	Precise detection rate unknown	7q21.3
Split-hand/foot malformation-3 (SHFM3)	246560	*FBXW4*	Precise detection rate unknown	10q24.32
Split-hand/foot malformation (SHFM5)	606708	multiple	Precise detection rate unknown	2q31.1
Subtelomeric regions (41 regions)		multiple	0.5-7% have a detectable deletion	41 sites
Synpolydactyly (SPD1)	186000	*HOXD cluster*	Low	2q31.1
Steroid sulfatase deficiency, X-linked/Ichthyosis, X-linked	308100	*STS*	80-90%	Xp22.31
Thrombocytopenia-Absent Radius syndrome	274000	*RBM8A*	>95%	1q21.1
Treacher Collins-Franceschetti syndrome (TCOF)	154500	*TCOF1*	Precise detection rate unknown	5q32
Trichorhinophalangeal syndrome 1 (TRPS1)	190350	*TRPS1*	<20%	8q23.3
Van der Woude syndrome (VWS)	119300	*IRF6*	2-3%	1q32.2
Waardenburg syndrome, Type I	193500	*PAX3*	~6%	2q36.1
Waardenburg syndrome, Type IIA (WS2A)	193510	*MITF*	Precise detection rate unknown	3p13-p14.1
Williams-Beuren syndrome (WBS)	194050	*ELN*	>95%	7q11.23
Wilms tumor-aniridia-genital anomalies-retardation (WAGR)	194072	*PAX6, WT1*	60-70%	11p13
Wilms tumor 1 (WT1)	194070	*WT1*	Deletions uncommon	11p13
Wolf-Hirschhorn syndrome	194190	multiple	>95%	4p16.3
XIST deficiency (Kabuki syndrome-like features)		*XIST*	Very low	Xq13.2
X-linked heterotaxy (HTX)	306955	*ZIC3*	Deletions uncommon	Xq26.3
X-linked lymphoproliferative syndrome (XLP)	308240	*SH2D1A*	4-9%	Xq25
Xp11.3 deletion with mental retardation	300578	*multiple*	Precise detection rate unknown	Xp11.3
X-linked ichthyosis (XLI)			>95%	Xp22.31
Uniparental isodisomy				
chromosomes 1-22, X		multiple	Detect uniparental isodisomy*	multiple

* To rule out uniparental heterodisomy or mat/ pat UPD, additional PCR for specific UPD is required. Please contact lab for test availability.