PRENATAL TARGETED REGIONS

Disorders Tested	OMIM#	Locus	Band(s)
Adrenal hypoplasia congenita (AHC)	300200	NR0B1	Xp21.2
Alagille	118450	JAG1	20p12.2
Albright hereditary osteodystrophy-like/Brachydactyly-MR	600430	(M)	2q37.3
Angelman	105830	UBE3A	15q11.2
Aniridia II	106210	PAX6	11p13
Basal cell nevus/Gorlin-Goltz	109400	PTCH1	9q22.32
Beckwith-Wiedemann	130650	IGF2	11p15.5
Blepharophimosis, ptosis, epicanthus inversus (BPE)	110100	FOXL2	3q22.3
Branchio-oto-renal (BOR)/Melnick-Fraser	113650	EYA1	8q13.3
Campomelic dysplasia (CMPD)	114290	SOX9	17q24.3
Cat-eye	115470	(M)	22q11.1
CHARGE	214800	CHD7	8q12.2
Charcot-Marie-Tooth disease, demyelinating, type 1A	118220	PMP22	17p12
Cleidocranial dysplasia (CCD)	119600	RUNX2	6p12.3
Cornelia de Lange 1	122470	NIPBL	5p13.2
Cri-du-Chat	123450	(M)	5p15.2
Dandy-Walker malformation (DWM)	220200	ZIC1, ZIC4	3q24
DiGeorge/Velocardiofacial (VCF)	188400	HIRA, TBX1	22q11.21
DiGeorge 2	601362	(M)	10p14
DMD	310200	Dystrophin	Xp21.2
Dosage-sensitive sex reversal	300018	NR0B1	Xp21.2
Familial adenomatous polyposis (FAP)/Gardner/MR	175100	APC	5q22.2
Feingold	164280	MYCN	2p24.3
FMR1 Microdeletion	300624	FMR1	Xq27.3
Glycerol kinase deficiency (GKD)	300474	GK	Xp21.2
Greig cephalopolysyndactyly	175700	GLI3	7p14.1
Holoprosencephaly 1	236100	TMEM1	21q22.3
Holoprosencephaly 2	157170	SIX3	2p21
Holoprosencephaly 3	142945	SHH	7q36.3
Holoprosencephaly 4	142946	TGIF	18p11.31
Holoprosencephaly 5	609637	ZIC2	13q32.3
Holoprosencephaly 6	605934	(M)	2q37.1-q37.3
Holoprosencephaly 7	610828	PTCH1	9q22.32
Hypoparathyroidism, sensorineural deafness, renal disease (HDR)	146255	GATA3	10p14
Jacobsen/11q terminal deletion disorder	147791	(M)	11q23-11qter
Joubert 4	609583	NPHP1	2q13
Kallmann 1	308700	KAL1	Xp22.31
Langer-Giedion	150230	TRPS1	8q23.3
		EXT1	8q24.11
Leri-Weill dyschondrosteosis (LWD) (OS)	127300	SHOX	Xpter-Xp22.3 & Ypter-Yp11.32
Leukodystrophy with microdeletion 11q14.3		(M)	11q14.2q14.3
Microphthalmia 7 with linear skin defects	309801	(M)	Xp22.2
Miller-Dieker	247200	PAFAH1B1 (LIS1)	17p13.3
Mowat-Wilson	235730	ZFHX1B	2q22.3
Nail-patella (NPS)	161200	LMX1B	9q33.3
Nephronophthisis 1	256100	NPHP1	2q13
Neurofibromatosis 1 (NF1)/MR	162200	NF1	17q11.2
Neurofibromatosis 2 (NF2)	101000	NF2	22q12.2
Noonan 1	163950	PTPN11	12q24.13
Oto-facio-cervical (OFC)	166780	EYA1	8q13.3
Pallister-Killian	601803	(M)	12p
Pelizaeus-Merzbacher	312080	PLP1	Xq22.2
Polycystic kidney disease 1 (PKD1) (OS)	601313	PKD1	16p13.3
Potocki-Shaffer	601224	EXT2, ALX4	11p11.2
Prader-Willi (PWS)	176270	SNRPN, Necdin	15q11.2
Prader-Willi-like phenotype	176270	SIM1	6q16.3
Retinoblastoma/MR	180200	RB1	13q14.2
Rett syndrome	312750	MECP2	Xq28
Rieger 1 (RIEG1)	180500	PITX2	4q25
Rubinstein-Taybi (RTS)	180849	CREBBP	16p13.3
Saethre-Chotzen	101400	TWIST1	7p21.1
Smith-Magenis (SMS)	182290	RAI1	17p11.2

Sotos	117550	NSD1	5q35.3
Split-hand/foot malformation 1 (SHFM1)	183600	SHFM1	7q21.3
Split-hand/foot malformation 3 (SHFM3)	600095	FBXW4	10q24.32
Split-hand/foot malformation 4 (SHFM4)	605289	TP73L	3q28
Split-hand/foot malformation 5 (SHFM5)	606708	DLX1, DLX2	2q31.1
SRY dosage abnormalities	278850/	SRY	Yp11.31
Steroid sulfatase deficiency	308100	STS	Xp22.31
Synpolydactyly/Syndactyly I	186000	HOXD gene cluster	2q31.1
Tuberous sclerosis 2 (TSC2) (OS)	191100	TSC2	16p13.3
		END3	20q13.2-q13.3
		SOX10	22q13.1
Williams-Beuren	194050	ELN	7q11.23
Wolf-Hirschhorn	194190	(M)	4p16.3
X-linked heterotaxy	306955	ZIC3	Xq26.3
X-linked lissencephaly	300067	DCX	Xq22.3
X-linked mental retardation with isolated growth hormone deficiency	300123	SOX3	Xq27.1
1p36 Microdeletion	607872	(M)	1p36
3q29 Microdeletion	609425	(M)	3q29
8p23.1 Microdeletion		(M)	8p23.1
17q21.3 Microdeletion	610443	(M)	17q21.3
22q13.3 Microdeletion	606232	(M)	22q13.3
All 41 unique subtelomeric regions		(M)	41 sites
All 43 unique pericentromeric regions		(M)	43 sites
Aneuploidy for 24 chromosomes		(M)	24 chromosomes