GENOMIC REGIONS ASSAYED FOR COPY NUMBER ABNORMALITIES BY PRENATAL TARGETED ARRAY

Syndrome	Locus
1p36 deletion syndrome	1p36
1q43-1q44 microdeletion	1q43-1q44
2p15 microdeletion	2p15-2p16.1
2q33 microdeletion	2q32.2-2q33
2q37 microdeletion	2q37
3q29 microdeletion	3q29
6p25 microdeletion (including FOXC1)	6p25
7q11 Williams region duplication	7q11.32
8p23 microdeletion	8p23.1
9q34 subtelomeric deletion	9q34
10q22-q23 microdeletion	10q22.3-q23.31
12q14.13 microdeletion	12q14.13
12q24.21 microduplication	12q24.21
14q22-14q23 microdeletion	14q22.1-q23.1
15q11.2-15q13.1 microduplication	15q11.2-q13.1
15q24 microdeletion	15q24.1-q24.3
16p11.2-16p12.2 microdeletion	16p11.2-p12.2
16p13.3 deletion, Rubenstein-Taybi syndrome	16p13.3
16q24.3 microdeletion	16q24.3
17p13.3 deletion	17p13.3
17p13.3 duplication	17p13.3
17q12 microdeletion	17q12
17q21 microdeletion/duplication	17q21.3
22q11 VCF/DGS deletion and reciprocal duplication	22q11.2
22q11.2 distal microdeletion	22q11.2
22q13.3 microdeletion	22q13.3
Alagille syndrome	20p12.2
Alpha thalassemia/mental retardation	16p13.3
Androgen insensitivity syndrome	Xq12
Angelman syndrome	15q11.2-q13.1
Aniridia type 2	11p13
Basal cell nevus syndrome (PTCH1)	9q22.32
Beckwith-Wiedemann syndrome	11p15.5
BPE syndrome	3q22.3
Branchio-oto-renal	8q13.3
Campomelic dysplasia	17q24.3
Cat-eye syndrome	22q11.21
Charcot-Marie-Tooth disease	17p11.2
CHARGE syndrome	8q12.2
Cleidocranial dysplasia	6p21.1
Cornelia de Lange syndrome	5p13.2
Cri-du-chat syndrome	5p15.2
Dandy-Walker malformation	3q24
Duane-radial ray syndrome (SALL4)	20q13.2
Esophageal atresia syndrome	16q24.1
Gonadal dysgenesis (SRY deletion)	Yp11.3
Greig cephalopolysyndactyly syndrome	7p14.1
Hemophilia A	Xq28
Hereditary neuropathy with pressure palsies	17p11.2
Holoprosencephaly type 2	2p21
Holoprosencephaly type 3	7q36

Syndrome	Locus
Holoprosencephaly type 4	18p11.3
Holoprosencephaly type 5	13q32
Holt-Oram-related disorders	12q24.21
Hypoparathyroidism, sensorineural deafness, and renal disease (HDR syndrome)	10p14
Jacobsen syndrome	11q23-11q25
Kallmann syndrome	Xp22.31
Langer-Giedion syndrome	8q24.11
Leri-Weill dyschondrosteosis	Xp22.33
Lowe syndrome	Xq25
MECP2 duplication MR syndrome	Xq28
Miller-Dieker syndrome	17p13.3
Mowat-Wilson syndrome	2q22.3
Nablus mask-like facial syndrome	8q21.3-8q22.1
Nail-patella syndrome	9q33.3
Neurofibromatosis type 1	17q11.2
Neurofibromatosis type 2	22q12.2
Opitz syndrome	Xp22
Oral-facial-digital syndrome type 1	Xp22.2
Ornithine transcarbamylase deficiency	Xp11.4
Pallister-Killian syndrome	12p arm
Pelizaeus-Merzbacher syndrome	Xq22.2
Polycystic kidney disease	16p13.3
Potocki-Lupski syndrome	17p11.2
Potocki-Shaffer syndrome	11p11.2
Prader-Willi syndrome	15q11.2-15q13.1
PTEN-related disorders	10q23.31
Retinoblastoma	13q14.2
Rett syndrome (MECP2 deletions)	Xq28
Rieger syndrome type 1	4q25
Rubenstein-Taybi syndrome	16p13.3
Saethre-Chotzen syndrome	7p21.1
Smith-Magenis syndrome	17p11.2
Smith-Lemli-Opitz syndrome	11q13.4
Sotos syndrome	5q35.3
Syndromic microphthalmia 3	3q26.33
Synpolydactyly	2q31.1
Townes-Brocks syndrome	16q12.1
Trichorhinophalangeal syndrome type 1	8q24.12
Tuberous sclerosis type 2	16p13.3
Ulnar-mammary syndrome	12q24.21
Van der Woude syndrome	1q32.2
Waardenburg syndrome type 2/2E	22q13.1
WAGR syndrome	11p13
Williams-Beuren syndrome	7q11.23
Wolf-Hirschhorn syndrome	4p16.3
X-linked hydrocephalus/MASA/CRASH	Xq28
X-linked ichthyosis	Xp22.31
Xp21 microdeletion (NR0B1, IL1RAPL1, GK)	Xp21
Xp11.3 microdeletion	Xp11.3
Xq28 duplication (GDI1)	Xp21

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