GENOMIC TARGETS FOR COPY NUMBER ABNORMALITIES BY PRENATAL TARGETED ARRAY

GeneDx
an OPKO Health Company

Syndrome/Region	Chromosomal Location
1p36 deletion syndrome	1p36
1q21.1 recurrent (TAR syndrome) region	1q21.1
1q21.1 recurrent distal region	1q21.1-q21.2
1q43-q44 microdeletion syndrome	1q43-1q44
2p15p16.1 region	2p16.1-p15
2q11.2 recurrent region	2q11.2
2q13 recurrent region	2q13
2q22.3-q23.3 microdeletion	2q22.3-q23.3
2q37 deletion/Brachydactyly-Mental Retardation syndrome/Albright Hereditary Osteodystrophy-like syndrome	2q37
3q29 microdeletion/microduplication syndrome	3q29
5q14.3 deletion syndrome	5q14.3
7q11.23 recurrent distal region	7q11.23
7q11.23 duplication syndrome	7q11.23
8p23 microdeletion/microduplication syndrome	8p23.1
10q22.3q23.2 deletion syndrome	10q22.3-q23.2
12q14 microdeletion	12q14.2-q15
14q11.2 deletion	14q11.2
15q13.3 microdeletion (BP4-BP5)	15q13.3
15q24 microdeletion	15q24.1-q24.2
15q25.2 microdeletion	15q25.2
16p11.2 recurrent region (distal, BP2-BP3)	16p11.2
16p11.2 recurrent region (proximal, BP4-BP5)	16p11.2
16p12.2 recurrent region	16p12.2
16p13.11 microdeletion/microduplication syndrome	16p13.11
17q12 microdeletion/microduplication syndrome	17q12
17q21.3 microdeletion/microduplication syndrome	17q21.3
22q11.2 recurrent region (distal, D-H)	22q11.21
22q13.3 microdeletion *[SHANK3]*	22q13.3
Agammaglobulinemia; Isolated growth hormone deficiency type III with agammaglobulinemia	Xq22.1
Alagille syndrome	20p12.2
Allan-Herndon-Dudley syndrome	Xq13.2
Alph-thalassemia/mental retardation syndrome (ATRX)	Xq21.1
Alport syndrome 1, X-linked	Xq22.3
Androgen insensitivity syndrome	Xq12
Angelman syndrome (BP1-BP3)	15q11q13
Axenfeld-Rieger syndrome	6p25.3

Syndrome/Region	Chromosomal Location
Basal cell nevus/Gorlin-Goltz/ Holoprosencephaly 7	9q22.32
Beckwith-Wiedemann syndrome	11p15.5
Blepharophimosis/ptosis/epicanthus inversus syndrome (BPES)	3q22.3
Branchio-oto-renal syndrome	8q13.3
Campomelic dysplasia	17q24.3
Cat eye syndrome	22q11.21
Cerebral cavernous malformations, type 2	7p13
Charcot-Marie-Tooth disease, type 1A (CMT1A) *[PMP22]*	17p12
CHARGE syndrome	8q12.1-q12.2
Cleidocranial dysplasia	6p21.1
Coffin-Lowry syndrome	Xp22.12
Congenital adrenal hypoplasia	Xp21.1
Congenital hydrocephalus/MASA syndrome	Xq28
Cornelia de Lange syndrome	5p13.2
Cri du chat syndrome	5p15.33-p15.2
DiGeorge syndrome 2	10p12.31
DiGeorge/velocardiofacial syndrome (proximal, A-D)	22q11.21
Duane radial ray syndrome *[SALL4]*	20q13.2
Duchenne muscular dystrophy	Xp21.2-p21.1
Early infantile epileptic encephalopathy-7	20q13.33
Epileptic encephalopathy, childhood-onset	15q26.2
Epileptic encephalopathy, early infantile, 2/ retinoschisis	Xp22.13
Epileptic encephalopathy, early infantile, 8	Xq11.1-Xq11.2
Epileptic encephalopathy, early infantile, 9 *[PCDH19]*	Xq22.1
Familial adenomatous polyposis (FAP)/ Gardner/Intellectual Disability	5q22.2
Feingold syndrome	2p24.3
FG syndrome 4	Xp11.4
Fragile X syndrome	Xq27.3
GLASS syndrome	2q32-q33
GLUT1 deficiency syndrome	1p34.2
Glycerol kinase deficiency	Xp21.1
Gonadal dysgenesis	Yp11.31
Greig cephalopolysyndactyly	7p14.1
Hemophilia A	Xq28
Hemorrhagic telangiectasia	12q13.13

207 PERRY PARKWAY GAITHERSBURG, MD 20877 • **T** 1 888 729 1206 (TOLL-FREE), 1 301 519 2100 • **F** 1 201 421 2010 • ZEBRAS@GENEDX.COM • WWW.GENEDX.COM

Syndrome/Region	Chromosomal Location
Hereditary neuropathy with liability to pressure palsies (HNPP) *[PMP22]*	17p12
Hirschsprung disease	10q11.21
Holoprosencephaly type 2	2p21
Holoprosencephaly type 3	7q36.3
Holoprosencephaly type 4	18p11.31
Holoprosencephaly type 5	13q32.3
Holoprosencephaly type 9	2q14.2
Holt-Oram syndrome *[TBX5]*	12q14
Hypoparathyroidism, sensorineural deafness, and renal disease (HDR syndrome)	10p14
Jacobsen syndrome	11q23.1-q24.1
Kallmann syndrome	Xp22.31
KBG syndrome *[ANKRD11]*	16q24.3
Kleefstra syndrome *[EHMT1]*	9q34.4
Langer-Giedion syndrome	8q23.3
Leri-Weill dyschondrosteosis	Xp22.33/Yp11.32
Lesch-Nyhan syndrome	Xq26.2-q26.3
Linear skin defects with multiple congenital anomalies 1	Xp22.2
Lissencephaly, X-linked	Xq23
Lowe syndrome	Xq25-q26.1
Marfan syndrome *[FBN1]*	15q21.1
Mental retardation and distinctive facial features with or without cardiac defects *[MED13L]*	12q24.13
Mental retardation syndrome, X-linked, Siderius type	Xp11.22
Mental retardation, X-linked *[IL1RAPL1]*	Xp21.3-p21.2
Mental retardation, X-linked syndromic, Cabezas type *[CUL4B]*	Xq24
Microphthalmia, syndromic 2 *[BCOR]*	Xp11.4
Miller-Dieker syndrome	17p13.3
Mowat-Wilson syndrome	2q22.3
Mucopolysaccharidosis II (Hunter syndrome)	Xq28
Nail-patella syndrome	9q33.3
Neurofibromatosis type 1	17q11.2
Neurofibromatosis, type 2	22q12.2
Noonan syndrome 1	12q24.13
Norrie disease	Xp11.3
Opitz syndrome	Xp22.2
Oral-facial-digital sydrome, type 1	Xp22.2
Ornithine transcarbamylase deficiency	Xp11.4

Syndrome/Region	Chromosomal Location
Pelizaeus-Merzbacher syndrome	Xq22.2
Pitt-Hopkins syndrome	18q21.1
Pitt-Hopkins-like syndrome 2 *[NRXN1]*	2p16.3
Polycystic kidney disease	16p13.3
Potocki-Lupski syndrome	17p11.2
Potocki-Shaffer syndrome	11p11.2
Prader-Willi syndrome (BP1-BP3)	15q11q13
PTEN-related disorders	10q23.31
Retinoblastoma	13q14.2
RETT syndrome/Lubs X-linked mental retardation syndrome *[MECP2]*	Xq28
Rieger 1 syndrome	4q25
Rubenstein-Taybi syndrome	16p13.3
Saethre-Chotzen syndrome	7p21.1
Simpson-Golabi-Behmel syndrome, type 1	Xq26.2
Smith-Magenis syndrome	17p11.2
Sotos syndrome	5q35.2-q35.3
Split-hand/foot malformation 1	7q21.2-q21.3
Syndromic microphthalmia 3	3q26.33
Synpolydactyly	2q31.1
Townes-Brocks syndrome	16q12.1
Van der Woude syndrome	1q32.2
Waardenburg syndrome, type 2A	3p13
Waardenburg syndrome, types 1 and 3	2q36.1
Williams syndrome	7q11.23
Wilms tumor, aniridia, genitourinary anomalies and mental retardation (WAGR) syndrome	11p13
Wolf-Hirschhorn syndrome	4p16.3
X-linked chondrodysplasia punctata	Xp22.33
X-linked chronic granulomatous disease	Xp11.4
X-linked heterotaxy	Xq26.3
X-linked ichthyosis	Xp22.31
X-linked mental retardation	Xp22.32-p22.31
X-linked mental retardation with isolated growth hormone deficiency	Xq27.1
X-linked periventricular heterotopia	Xq28
Xp11.22-linked intellectual disability	Xp11.2
Xp11.23 region *[MAOA, MAOB]*	Xp11.3
Xp11.23-p11.22 duplication syndrome	Xp11.23-p11.22
Xq25 microduplication syndrome	Xq25
Xq28 duplication syndrome	Xq28
XY sex reversal	9q33.3

207 PERRY PARKWAY
GAITHERSBURG, MD 20877
T 1 888 729 1206 (TOLL-FREE), 1 301 519 2100 • **F** 1 201 421 2010
ZEBRAS@GENEDX.COM • WWW.GENEDX.COM

GeneDx
an OPKO Health Company