

60 deletion and duplication syndromes (Nifty Plus)

Alpha Thalassemia, mental retardation syndrome

Androgen insensitivity syndrome (AIS)

Angelman syndrome/Prader-Willi syndrome

Bannayan-Riley-Ruvalcaba syndrome (BRRS)

Branchiootorenal dysplasia syndrome (BOR)/ Melnick-Fraser syndrome

Cat eye syndrome (CES)

Chromosome 10q deletion syndrome

Chromosome 10q22.3–q23.31 microdeletion syndrome

Chromosome 18p deletion syndrome

Chromosome 18q deletion syndrome

Cornelia de Lange syndrome (CDLS)

Cowden syndrome (CD)

Cri du Chat (5p deletion) syndrome

Dandy-Walker syndrome (DWS)

DiGeorge syndrome type 2 (DGS2)

Distal arthrogryposis type 2B (DA2B)

Duchenne and Becker muscular dystrophy (DMD/BMD)

Dyggve-Melchior-Clausen syndrome (DMC)

Feingold syndrome

Holoprosencephaly type 1 (HPE1)

Holoprosencephaly type 4 (HPE4)

Holoprosencephaly type 6 (HPE6)

Jacobsen syndrome

Langer-Giedion syndrome (LGS)

Leukodystrophy with 11q14.2–q14.3

Mental retardation, X-linked growth hormone deficiency (MRGH)

Microphthalmia, syndromic type 6, pituitary hypoplasia

Microphthalmia with linear skin defects

Monosomy 9p syndrome

Orofaciodigital syndrome

Panhypopituitarism, X-linked

Potocki-Lupski syndrome (17p11.2 duplication syndrome)

Prader-Willi-like syndrome (SIM1 syndrome)

Rieger syndrome type 1 (RIEG1)

Saethre-Chotzen syndrome (SCS)

Sensorineural deafness and male infertility

Smith-Magenis syndrome

Split-hand/foot malformation type 3 (SHFM3)

Split-hand/foot malformation type 5 (SHFM5)

Syndrome diaphragmatic hernia, congenital (HCD/DIH)

Trichorhinophalangeal syndrome type I (TRPS1)

Van der Woude syndrome (VWS)

WAGR syndrome and aniridia II

Wilms tumor 1 (WT1)

X-linked lymphoproliferative syndrome (XLP)

Xp11.22–p11.23 microduplication syndrome

1p36 microdeletion syndrome

1q41–q42 microdeletion syndrome

2q33.1 deletion syndrome/Glass syndrome

5q21.1–q31.2 deletion syndrome

8p23.1 deletion syndrome

8p23.1 duplication syndrome

11q11–q13.3 duplication syndrome

12q14 microdeletion syndrome

14q11–q22 deletion syndrome

15q26 overgrowth syndrome

16p11.2–p12.2 microdeletion syndrome

16p11.2–p12.2 microduplication syndrome

17q21.31 deletion syndrome

17q21.31 duplication syndrome