

dup(4)(q31.3)

- tandem duplication on Chr4: 150746214(q31.3) - 151129847(q31.3)
- all genes near breakpoints: ['SH3D19', 'LOC729558', 'SNORD73B', 'ZNF718', 'ZNF595', 'LRBA', 'MAB21L2', 'RPS3A', 'PRSS48', 'ZNF876P', 'SNORD73A']
- genes near breakpoints documented in DDG2P:
LRBA(606453): CHILDHOOD-ONSET HYPOGAMMAGLOBULINEMIA(614700)
MAB21L2(604357): MICROPHTHALMIA, SYNDROMIC 14(615877),MICROPHTHALMIA, SYNDROMIC 14(615877)
- all genes with +1 CN: ['LRBA', 'RPS3A', 'SNORD73B', 'SNORD73A', 'SH3D19']
- genes with +1 CN documented in DDG2P:
LRBA(606453): CHILDHOOD-ONSET HYPOGAMMAGLOBULINEMIA(614700)

del(2)(q14.1)

- deletion on Chr2: 117154946(q14.1) - 117670484(q14.1)
- all genes near breakpoints: ['DDX18', 'SH3YL1', 'FAM110C', 'LOC107985939']
- genes near breakpoints documented in DDG2P: None
- all genes with -1 CN: []
- genes with -1 CN documented in DDG2P: None

del(8)(p23.2)

- deletion on Chr8: 2366171(p23.2) - 2696469(p23.2)
- all genes near breakpoints: ['OR4F21', 'RPL23AP53', 'LOC105377785', 'LOC101927506', 'ZNF596', 'LOC101927815']
- genes near breakpoints documented in DDG2P: None
- all genes with -1 CN: ['LOC101927815']
- genes with -1 CN documented in DDG2P: None