

dup(1)(p13.3)

- tandem duplication on Chr1: 108908921(p13.3) - 109192502(p13.3)
- all genes near breakpoints: ['SORT1', 'WDR47', 'MIR6859-4', 'MIR6859-1', 'CLCC1', 'SARS1', 'AKNAD1', 'MIR1302-10', 'MYBPHL', 'STXBP3', 'SPATA42', 'GPSM2', 'FAM138C', 'CELSR2', 'FAM138F', 'DDX11L1', 'TAF13', 'ELAPOR1', 'MIR1302-11', 'FAM138A', 'WASH7P', 'DDX11L17', 'MIR1302-9', 'FNDC7', 'FAM138D', 'LOC729737', 'PSRC1', 'TMEM167B', 'C1orf194', 'MIR6859-3', 'MIR1302-2', 'MIR6859-2', 'SCARNA2', 'OR4F5']
- genes near breakpoints documented in DDG2P:
GPSM2(609245): CHUDLEY-MCCULLOUGH SYNDROME(604213)
TAF13(600774): Autosomal-Recessive Intellectual Disability and Microcephaly(No disease mim)
SARS1(607529): SARS1-related neurodevelopmental disorder with microcephaly, ataxia, and seizures(617709),Autosomal dominant SARS1-related neurodevelopmental disorder(No disease mim)
- all genes with +1 CN: ['GPSM2', 'CLCC1', 'WDR47', 'TAF13', 'TMEM167B', 'SCARNA2', 'C1orf194', 'ELAPOR1']
- genes with +1 CN documented in DDG2P:
GPSM2(609245): CHUDLEY-MCCULLOUGH SYNDROME(604213)
TAF13(600774): Autosomal-Recessive Intellectual Disability and Microcephaly(No disease mim)

del(8)(q24.23)

- deletion on Chr8: 136620058(q24.23) - 136875709(q24.23)
- all genes near breakpoints: ['OR4F21', 'RPL23AP53', 'LOC101927506', 'ZNF596', 'LINC02055']
- genes near breakpoints documented in DDG2P: None
- all genes with -1 CN: ['LINC02055']
- genes with -1 CN documented in DDG2P: None