## inv(17)(p11.2)

- inversion on Chr17: 16814036(p11.2) 18492887(p11.2)
- $all \ genes \ near \ breakpoints: \ ['ZNF286B', 'EVPLL', 'TNFRSF13B', 'LINC02076', 'KRT17P5', 'SHMT1', 'LINC02076', 'LINC02076'$

'ZNF624', 'KRT17P2', 'SMCR8', 'USP32P1', 'FAM106C', 'TOP3A', 'KRT17P1', 'SCGB1C2', 'LOC284191',

'LINC02091', 'MIR6778', 'TBC1D27P', 'KRT16P2', 'FAM106A', 'TBC1D28', 'LINC02090', 'FOXO3B',

'LGALS9C', 'KRT16P1', 'DOC2B', 'CCDC144A', 'USP32P2', 'CCDC144B']

- genes near breakpoints documented in DDG2P:

TNFRSF13B(604907): IMMUNODEFICIENCY, COMMON VARIABLE, 2(240500)

TOP3A(601243): Bloom Syndrome like Disorder(No disease mim)

## dup(14)(q23.3q24.1)

- tandem duplication on Chr14: 66782235(q23.3) 67429788(q24.1)
- all genes near breakpoints: ['MPP5', 'MIR5694', 'ARG2', 'TMEM229B', 'PIGH', 'GPHN', 'PLEK2', 'PLEKHH1', 'EIF2S1', 'ATP6V1D']
- genes near breakpoints documented in DDG2P:

PIGH(600154): GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT 17(618010)

GPHN(603930): GPHN-related molybdenum cofactor deficiency(615501)

- all genes with +1 CN: ['GPHN', 'FAM71D', 'MPP5', 'ATP6V1D', 'EIF2S1', 'PLEK2']
- genes with +1 CN documented in DDG2P:

GPHN(603930): GPHN-related molybdenum cofactor deficiency(615501)

## del(9)(p21.2)

- deletion on Chr9: 26212045(p21.2) 26499987(p21.2)
- all genes near breakpoints: ['WASHC1', 'PGM5P3-AS1', 'FOXD4', 'DOCK8-AS1', 'FAM138A', 'LOC100506422', 'MIR1302-9', 'MIR1302-11', 'MIR1302-2', 'FAM138F', 'DDX11L5', 'MIR1302-10', 'CBWD1', 'DOCK8', 'FAM138C']
- genes near breakpoints documented in DDG2P:

DOCK8(611432): HYPERIMMUNOGLOBULIN E RECURRENT INFECTION SYNDROME AUTOSOMAL RECESSIVE(243700)

- all genes with -1 CN: []
- genes with -1 CN documented in DDG2P: None