

inv(1)(q21.1q21.2)

- inversion on Chr1: 146327105(q21.1) - 149352353(q21.2)
- all genes near breakpoints: ['SEC22B2P', 'RNU1-1', 'NUDT4P2', 'NOTCH2NLC', 'NBPF19', 'RNU1-2', 'RNU1-3', 'PPIAL4F', 'HYDIN2', 'NUDT4B', 'RNVU1-18', 'DRD5P2', 'PPIAL4H', 'NBPF10', 'NBPF15', 'SRGAP2-AS1', 'RNU1-4', 'LOC101060524', 'RNVU1-2A', 'LOC653513', 'NOTCH2NLA', 'LOC105371215', 'PPIAL4E', 'LSP1P5', 'PPIAL4D', 'LOC101927429', 'SEC22B3P']
- genes near breakpoints documented in DDG2P: None

inv(1)(q21.1q21.2)

- inversion on Chr1: 146327105(q21.1) - 149352353(q21.2)
- all genes near breakpoints: ['SEC22B2P', 'RNU1-1', 'NUDT4P2', 'NOTCH2NLC', 'NBPF19', 'RNU1-2', 'RNU1-3', 'PPIAL4F', 'HYDIN2', 'NUDT4B', 'RNVU1-18', 'DRD5P2', 'PPIAL4H', 'NBPF10', 'NBPF15', 'SRGAP2-AS1', 'RNU1-4', 'LOC101060524', 'RNVU1-2A', 'LOC653513', 'NOTCH2NLA', 'LOC105371215', 'PPIAL4E', 'LSP1P5', 'PPIAL4D', 'LOC101927429', 'SEC22B3P']
- genes near breakpoints documented in DDG2P: None

del(6)(p21.32)

- deletion on Chr6: 32906203(p21.32) - 33210303(p21.32)
- all genes near breakpoints: ['RING1', 'HLA-DQA2', 'PFDN6', 'HLA-DPB2', 'HLA-DMA', 'PSMB8', 'MIR6873', 'MIR6834', 'RXRB', 'HLA-DOB', 'TAPBP', 'RPS18', 'HLA-DPA1', 'HLA-DPB1', 'SLC39A7', 'BRD2', 'HLA-DOA', 'LOC285766', 'VPS52', 'HSD17B8', 'PSMB8-AS1', 'COL11A2', 'HCG25', 'ZBTB22', 'WDR46', 'LINC00266-3', 'HCG24', 'RGL2', 'HLA-DMB', 'MIR3135B', 'B3GALT4', 'TAP2', 'LOC100294145', 'DAXX', 'PSMB9', 'SMIM40', 'HLA-DQB2', 'TAP1', 'KIFC1', 'MIR219A1']
- genes near breakpoints documented in DDG2P:
COL11A2(120290): AUTOSOMAL RECESSIVE OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA(215150),DEAFNESS AUTOSOMAL DOMINANT TYPE 13(601868),DEAFNESS AUTOSOMAL RECESSIVE TYPE 53(609706),STICKLER SYNDROME TYPE 3(184840)
PSMB8(177046): NAKAJO SYNDROME(256040)
- all genes with -1 CN: ['HLA-DMB', 'HLA-DMA', 'BRD2', 'HLA-DOA', 'HLA-DPA1', 'HLA-DPB1', 'HLA-DPB2', 'HCG24', 'COL11A2', 'RXRB', 'SLC39A7', 'HSD17B8', 'MIR219A1', 'RING1']
- genes with -1 CN documented in DDG2P:
COL11A2(120290): AUTOSOMAL RECESSIVE OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA(215150),DEAFNESS AUTOSOMAL DOMINANT TYPE 13(601868),DEAFNESS AUTOSOMAL RECESSIVE TYPE 53(609706),STICKLER SYNDROME TYPE 3(184840)

