balanced trans skipped: (1, 'balanced_translocation', ['45.1.mt(42+).45+.46+', '46.0.wt(42+).p-ter.43+'])

- x

- x

- x

inv(16)(p12.2)

- inversion on Chr16: 21502978(p12.2) - 22436123(p12.2)

- all genes near breakpoints: ['RHBDF1', 'SMG1P1', 'METTL9', 'HBQ1', 'SNRNP25', 'HBM', 'LOC100190986', 'MIR6859-3', 'WASIR2', 'SLC7A5P2', 'MIR6859-1', 'POLR3E', 'HBZ', 'OTOAP1', 'MFSD13B', 'NPIPB3', 'MPG', 'HBA2', 'DDX11L10', 'MIR3680-1', 'RRN3P3', 'OTOA', 'LOC101927814', 'CRYM-AS1', 'NPIPB5', 'SMG1P3', 'MIR3680-2', 'EEF2K', 'CRYM', 'NPRL3', 'LOC440346', 'MIR6859-2', 'CDR2', 'IGSF6', 'SNX29P1', 'MIR6859-4', 'HBA1', 'LUC7L', 'POLR3K']

- genes near breakpoints documented in DDG2P: None

inv(17)(p11.2)

- inversion on Chr17: 16806184(p11.2) - 18509508(p11.2)

- all genes near breakpoints: ['ZNF286B', 'EVPLL', 'TNFRSF13B', 'LINC02076', 'KRT17P5', 'SHMT1', 'ZNF624', 'KRT17P2', 'SMCR8', 'USP32P1', 'FAM106C', 'TRIM16L', '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)

del(Y)(p11.32p11.2)

- deletion on ChrY: 0(p11.32) - 1439001(p11.2)

- all genes near breakpoints: ['IL3RA', 'ASMTL', 'AKAP17A', 'ASMTL-AS1', 'ASMT', 'SLC25A6', 'MIR3690', 'P2RY8', 'CSF2RA', 'LINC00106']

- genes near breakpoints documented in DDG2P: None

- all genes with -1 CN: ['PLCXD1', 'GTPBP6', 'LINC00685', 'PPP2R3B', 'SHOX', 'CRLF2', 'CSF2RA', 'MIR3690', 'IL3RA', 'SLC25A6', 'LINC00106', 'ASMTL-AS1', 'ASMTL']

- genes with -1 CN documented in DDG2P:
SHOX(312865): LANGER MESOMELIC DYSPLASIA(249700),LERI-WEILL DYSCHONDROSTEOSIS(127300)

del(Y)(p11.2q12)

- deletion on ChrY: 1439002(p11.2) - 57212132(q12)

- all genes near breakpoints: ['IL3RA', 'IL9R', 'ASMTL', 'AKAP17A', 'VAMP7', 'ASMTL-AS1', 'ASMT', 'SLC25A6', 'WASIR1', 'MIR3690', 'DDX11L16', 'P2RY8', 'CSF2RA', 'LINC00106']

- genes near breakpoints documented in DDG2P: None

- all genes with -1 CN: ['ASMTL', 'P2RY8', 'AKAP17A', 'ASMT', 'DHRSX', 'ZBED1', 'MIR6089', 'CD99P1', 'LINC00102', 'CD99', 'XGY2', 'SRY', 'RPS4Y1', 'ZFY', 'ZFY-AS1', 'LINC00278', 'TGIF2LY', 'MIR9985', 'PCDH11Y', 'TTTY23B', 'TTTY23', 'TSPY2', 'FAM197Y9', 'LINC00280', 'TTTY1', 'TTTY1B', 'TTTY2', 'TTTY2B', 'TTTY21', 'TTTY21B', 'TTTY7B', 'TTTY7', 'TTTY8B', 'TTTY8', 'AMELY', 'TBL1Y', 'PRKY', 'TTTY16', 'TTTY12', 'LINC00279', 'TTTY18', 'TTTY19', 'TTTY11', 'RBMY1A3P', 'TTTY20', 'TSPY1', 'TSPY10', 'TSPY3', 'TSPY4', 'TSPY8', 'FAM197Y4', 'FAM197Y2', 'FAM197Y7', 'FAM197Y5', 'FAM197Y8', 'FAM197Y6', 'FAM197Y3', 'RBMY3AP', 'TTTY22', 'GYG2P1', 'TTTY15', 'USP9Y', 'DDX3Y', 'UTY', 'MIR12120', 'TMSB4Y', 'VCY1B', 'VCY', 'NLGN4Y', 'NLGN4Y-AS1', 'FAM41AY1', 'FAM41AY2', 'FAM224B', 'FAM224A', 'XKRY2', 'XKRY', 'CDY2B', 'CDY2A', 'HSFY2', 'HSFY1', 'TTTY9B', 'TTTY9A', 'TTTY14', 'BCORP1', 'TXLNGY', 'KDM5D', 'TTTY10', 'EIF1AY', 'RPS4Y2', 'PRORY', 'RBMY2EP', 'RBMY1D', 'RBMY1A1', 'RBMY1B', 'RBMY1E', 'TTTY13', 'PRY2', 'PRY', 'LOC101929148', 'TTTY6', 'TTTY6B', 'RBMY1F', 'RBMY1J', 'TTTY5', 'RBMY2FP', 'LOC100652931', 'TTTY17C', 'TTTY17B', 'TTTY17A', 'TTTY4', 'TTTY4C', 'TTTY4B', 'BPY2', 'BPY2B', 'BPY2C', 'DAZ4', 'DAZ1', 'DAZ3', 'DAZ2', 'TTTY3B', 'TTTY3', 'CDY1', 'CDY1B', 'CSPG4P1Y', 'GOLGA2P3Y', 'GOLGA2P2Y', 'SPRY3', 'VAMP7', 'IL9R', 'WASIR1']

- genes with -1 CN documented in DDG2P:
SRY(480000): 46XY SEX REVERSAL 1(400045)