

balanced trans skipped: (1, 'balanced_translocation', ['45.1.mt(44+).46+.47+', '46.0.wt(44+).p-ter.45+'])

- X
- X
- X

inv(16)(p12.2)

- inversion on Chr16: 21502978(p12.2) - 22436123(p12.2)
- all genes near breakpoints: ['POLR3K', 'CRYM', 'MIR3680-1', 'OTOAP1', 'METTL9', 'MIR6859-4', 'MIR6859-1', 'HBM', 'LOC440346', 'CDR2', 'NPIP5', 'POLR3E', 'HBQ1', 'MFSD13B', 'SMG1P1', 'HBA1', 'HBA2', 'WASIR2', 'RHBDF1', 'SMG1P3', 'NPRL3', 'HBZ', 'CRYM-AS1', 'LUC7L', 'SNRNP25', 'EEF2K', 'IGSF6', 'MPG', 'RRN3P3', 'LOC101927814', 'MIR6859-3', 'MIR3680-2', 'SNX29P1', 'NPIPB3', 'MIR6859-2', 'OTOA', 'SLC7A5P2', 'LOC100190986', 'DDX11L10']
- genes near breakpoints documented in DDG2P: None

del(9)(p11.2p11.1)

- deletion on Chr9: 41293386(p11.2) - 42559661(p11.1)
- all genes near breakpoints: ['CBWD3', 'LOC440896', 'LOC102724238', 'LOC107984035', 'LOC101927827', 'LOC103908605', 'MIR4477B', 'CBWD5', 'FGF7P3', 'FOXD4L6', 'CBWD6', 'GLIDR', 'LOC554249', 'MIR4477A']
- genes near breakpoints documented in DDG2P: None
- all genes with -1 CN: ['LOC107984035', 'FAM27E2', 'FAM242F', 'CNTNAP3B', 'SPATA31A6', 'FAM74A7']
- genes with -1 CN documented in DDG2P: None

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

- deletion on ChrY: 0(p11.32) - 1439001(p11.2)
- all genes near breakpoints: ['CSF2RA', 'ASMTL-AS1', 'SLC25A6', 'LINC00106', 'ASMT', 'AKAP17A', 'ASMTL', 'IL3RA', 'MIR3690', 'P2RY8']
- 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: ['CSF2RA', 'ASMTL-AS1', 'SLC25A6', 'IL9R', 'LINC00106', 'ASMT', 'AKAP17A', 'DDX11L16', 'ASMTL', 'WASIR1', 'IL3RA', 'MIR3690', 'VAMP7', 'P2RY8']
- 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', 'RBM1A3P', 'TTTY20', 'TSPY1', 'TSPY10', 'TSPY3', 'TSPY4', 'TSPY8', 'FAM197Y4', 'FAM197Y2', 'FAM197Y7', 'FAM197Y5', 'FAM197Y8', 'FAM197Y6', 'FAM197Y3', 'RBM1A3P', 'TTTY22', 'GYG2P1', 'TTTY15', 'USP9Y', 'DDX3Y', 'UTY', 'MIR12120', 'TMSB4Y', 'VCY1B', 'VCY', 'NLGN4Y', 'NLGN4Y-AS1', 'FAM41A1', 'FAM41A2', 'FAM224B', 'FAM224A', 'XKRY2', 'XKRY', 'CDY2B', 'CDY2A', 'HSFY2', 'HSFY1', 'TTTY9B', 'TTTY9A', 'TTTY14', 'BCORP1', 'TXLNGY', 'KDM5D', 'TTTY10', 'EIF1AY', 'RPS4Y2', 'PRORY', 'RBM1A2EP', 'RBM1A1D', 'RBM1A1A', 'RBM1A1B', 'RBM1A1E', 'TTTY13', 'PRY2', 'PRY', 'LOC101929148', 'TTTY6', 'TTTY6B', 'RBM1A1F', 'RBM1A1J', 'TTTY5', 'RBM1A2FP', '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)

ins(9)(p11.2p11.1p12)

- insertion of Chr9: 42559661(p11.1) - 39875146(p12) into Chr9: 41293386
- all genes near breakpoints: ['LOC554249', 'LOC101927827', 'LOC440896', 'LOC107984035', 'LOC102724238', 'LOC103908605', 'MIR4477B', 'CBWD5', 'FOXD4L6', 'FGF7P3', 'CBWD6', 'GLIDR', 'CBWD3', 'MIR4477A']
- genes near breakpoints documented in DDG2P: None
- all genes with +1 CN: []
- genes with +1 CN documented in DDG2P: None