inv(10)(q11.22)

- inversion on Chr10: 46991593(q11.22) - 48060267(q11.22)

- all genes near breakpoints: ['NPY4R', 'PTPN20', 'LINC02564', 'FRMPD2', 'FAM25C', 'AGAP13P', 'FRMPD2B', 'BMS1P1', 'TUBB8', 'NPY4R2', 'ZMYND11', 'LOC102723376', 'AGAP12P', 'FAM25G']

- genes near breakpoints documented in DDG2P:

ZMYND11(608668): INTELLECTUAL DISABILITY(616579)

dup(3)(p26.1)

- tandem duplication on Chr3: 4168214(p26.1) - 4532461(p26.1)

- all genes near breakpoints: ['SUMF1', 'SETMAR', 'ITPR1-DT', 'ITPR1', 'CHL1-AS2', 'LINC01986', 'CHL1']

- genes near breakpoints documented in DDG2P:

SUMF1(607939): SULFATIDOSIS, JUVENILE, AUSTIN TYPE(272200)

ITPR1(147265): SPINOCEREBELLAR ATAXIA 29, CONGENITAL NONPROGRESSIVE(117360),Gillespie Syndrome, monoallelic(206700),Gillespie Syndrome(206700)

- all genes with +1 CN: ['SETMAR', 'SUMF1', 'ITPR1-DT', 'ITPR1']

- genes with +1 CN documented in DDG2P:

SUMF1(607939): SULFATIDOSIS, JUVENILE, AUSTIN TYPE(272200)

ITPR1(147265): SPINOCEREBELLAR ATAXIA 29, CONGENITAL NONPROGRESSIVE(117360),Gillespie Syndrome, monoallelic(206700),Gillespie Syndrome(206700)

del(X)(p21.1)

- deletion on ChrX: 31580439(p21.1) - 31836526(p21.1)

- all genes near breakpoints: ['DMD']

- genes near breakpoints documented in DDG2P:

DMD(300377): DUCHENNE MUSCULAR DYSTROPHY(310200)

- all genes with -1 CN: ['DMD']

- genes with -1 CN documented in DDG2P:

DMD(300377): DUCHENNE MUSCULAR DYSTROPHY(310200)