

inv(2)(q12.2q12.3)

- inversion on Chr2: 106462406(q12.2) - 107833526(q12.3)
- all genes near breakpoints: ['LOC100133920', 'RGPD4-AS1', 'RGPD4', 'CD8B2', 'SLC5A7', 'RGPD3', 'LOC100509620', 'ANAPC1P6', 'GACAT1', 'PLGLA']
- genes near breakpoints documented in DDG2P:
SLC5A7(608761): Congenital Myasthenic Syndrome with Episodic Apnea(No disease mim)

inv(9)(q22.32q22.33)

- inversion on Chr9: 94438791(q22.32) - 96949317(q22.33)
- all genes near breakpoints: ['MFSD14C', 'LOC101928195', 'ZNF510', 'CBWD5', 'LOC101929583', 'NUTM2F', 'LOC101928381', 'MFSD14B', 'FBP1', 'ZNF782', 'ZNF169', 'CTSV', 'GAS2L1P2', 'LOC100132077', 'PTMAP11', 'FOXD4L4', 'LINC02603', 'FBP2', 'LOC100132781', 'PCAT7', 'LOC105379443', 'NUTM2G']
- genes near breakpoints documented in DDG2P:
FBP1(611570): FRUCTOSE 1,6 BISPHOSPHATASE DEFICIENCY(229700)

inv(10)(q11.22)

- inversion on Chr10: 46991593(q11.22) - 48060267(q11.22)
- all genes near breakpoints: ['NPY4R2', 'ZMYND11', 'LOC102723376', 'BMS1P1', 'TUBB8', 'NPY4R', 'FRMPD2', 'FAM25C', 'AGAP12P', 'LINC02564', 'AGAP13P', 'FRMPD2B', 'PTPN20', 'FAM25G']
- genes near breakpoints documented in DDG2P:
ZMYND11(608668): INTELLECTUAL DISABILITY(616579)

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', 'NP1PB5', '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', 'NP1PB3', 'MIR6859-2', 'OTOA', 'SLC7A5P2', 'LOC100190986', 'DDX11L10']
- genes near breakpoints documented in DDG2P: None

inv(17)(p11.2)

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

- all genes near breakpoints: ['LINC02090', 'TBC1D28', 'TOP3A', 'ZNF286B', 'CCDC144B', 'EVPLL', 'LOC284191', 'KRT17P5', 'FAM106A', 'USP32P1', 'MIR6778', 'LINC02076', 'KRT17P1', 'SHMT1', 'SMCR8', 'DOC2B', 'TRIM16L', 'KRT17P2', 'KRT16P1', 'FAM106C', 'FOXO3B', 'LINC02091', 'ZNF624', 'CCDC144A', 'TNFRSF13B', 'LGALS9C', 'KRT16P2', 'USP32P2', 'TBC1D27P', 'SCGB1C2']

- genes near breakpoints documented in DDG2P:

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

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

dup(2)(q14.3)

- tandem duplication on Chr2: 124318296(q14.3) - 125307772(q14.3)

- all genes near breakpoints: ['RGP4-AS1', 'CNTNAP5', 'SLC5A7', 'RGP4', 'GACAT1']

- genes near breakpoints documented in DDG2P:

SLC5A7(608761): Congenital Myasthenic Syndrome with Episodic Apnea(No disease mim)

- all genes with +1 CN: ['CNTNAP5']

- genes with +1 CN documented in DDG2P: None

del(9)(p21.1)

- deletion on Chr9: 28498341(p21.1) - 28758302(p21.1)

- all genes near breakpoints: ['DDX11L5', 'DOCK8-AS1', 'CBWD1', 'MIR1302-11', 'WASHC1', 'MIR873', 'FAM138A', 'MIR1302-10', 'MIR1302-9', 'FOXO4', 'LINGO2', 'PGM5P3-AS1', 'FAM138C', 'MIR1302-2', 'MIR876', 'DOCK8', 'FAM138F']

- genes near breakpoints documented in DDG2P:

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

- all genes with -1 CN: ['LINGO2']

- genes with -1 CN documented in DDG2P: None

del(15)(q14)

- deletion on Chr15: 34346216(q14) - 34614876(q14)

- all genes near breakpoints: ['GOLGA8B', 'SLC12A6', 'LPCAT4', 'ACTC1', 'EMC4', 'LOC100288637', 'OTUD7A', 'GOLGA8A', 'MIR1233-2', 'LINC02252', 'GJD2', 'MIR1233-1', 'NUTM1', 'LOC101928174', 'KATNBL1', 'NOP10']

- genes near breakpoints documented in DDG2P:

NOP10(606471): DYSKERATOSIS CONGENITA, AUTOSOMAL RECESSIVE 1(224230)

SLC12A6(604878): AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL

NEUROPATHY(218000)

OTUD7A(612024): 15q13.3 deletions phenocopy(No disease mim)

- all genes with -1 CN: ['NUTM1', 'LPCAT4', 'GOLGA8A', 'MIR1233-1', 'MIR1233-2', 'GOLGA8B']

- genes with -1 CN documented in DDG2P: None