inv(7)(q11.23)

- inversion on Chr7: 73049623(q11.23) - 75284230(q11.23)

- all genes near breakpoints: ['NSUN5P1', 'LOC105375115', 'NSUN5P2', 'SPDYE5', 'SPDYE15', 'LOC541473', 'POM121', 'SPDYE10P', 'SPDYE14', 'NCF1C', 'GTF2IRD2B', 'GTF2IP4', 'PMS2P2', 'POM121C', 'SPDYE11', 'LOC100101148', 'TRIM73', 'HIP1', 'SPDYE13', 'SPDYE9', 'TRIM74', 'GTF2IP1', 'STAG3L3', 'NCF1B', 'PMS2P7', 'LOC100507642', 'SPDYE8', 'SPDYE7P', 'LOC102723672', 'STAG3L1', 'PMS2P3', 'FAM20C']

- genes near breakpoints documented in DDG2P:
FAM20C(611061): RAINE SYNDROME(259775)

inv(10)(p15.3p12.1)

- inversion on Chr10: 0(p15.3) - 27458194(p12.1)

- all genes near breakpoints: ['PTCHD3', 'ZMYND11', 'ARMC4P1', 'RAB18', 'LOC102723376', 'TUBB8', 'LINC02564']

- genes near breakpoints documented in DDG2P:
RAB18(602207): WARBURG MICRO SYNDROME TYPE 3(614222)
ZMYND11(608668): INTELLECTUAL DISABILITY(616579)

dup(7)(q11.23)

- tandem duplication on Chr7: 75353606(q11.23) - 77013957(q11.23)

- all genes near breakpoints: ['NSUN5P1', 'DTX2P1-UPK3BP1-PMS2P11', 'SPDYE18', 'SPDYE5', 'SPDYE15', 'LOC541473', 'PMS2P9', 'NCF1C', 'SPDYE10P', 'SPDYE14', 'PMS2P2', 'GTF2IRD2B', 'FAM185BP', 'POM121C', 'LOC100101148', 'SPDYE11', 'TRIM73', 'HIP1', 'SPDYE13', 'SPDYE9', 'FGL2', 'TRIM74', 'GTF2IP1', 'PMS2P7', 'SPDYE17', 'SPDYE8', 'STAG3L1', 'PMS2P3', 'CCDC146']

- genes near breakpoints documented in DDG2P: None

- all genes with +1 CN: ['PMS2P7', 'PMS2P2', 'STAG3L1', 'LOC100101148', 'LOC541473', 'TRIM73', 'TRIM74', 'NSUN5P1', 'POM121C', 'SPDYE5', 'PMS2P3', 'HIP1', 'CCL26', 'CCL24', 'RHBDD2', 'POR', 'MIR4651', 'SNORA14A', 'TMEM120A', 'STYXL1', 'MDH2', 'GTF2IP7', 'SRRM3', 'HSPB1', 'YWHAG', 'SSC4D', 'ZP3', 'DTX2', 'FDPSP2', 'UPK3B', 'SPDYE16', 'LOC100133091', 'POMZP3', 'DTX2P1-UPK3BP1-PMS2P11']

- genes with +1 CN documented in DDG2P:
MDH2(154100): Early-Onset Severe Encephalopathy(No disease mim)
YWHAG(605356): Early-Onset Epilepsy(No disease mim)

del(10)(q26.3p12.1)

- deletion on Chr10: 133785267(q26.3) - 27458194(p12.1)

- all genes near breakpoints: ['PTCHD3', 'ZMYND11', 'LOC102723376', 'ARMC4P1', 'TUBB8', 'LINC02564', 'RAB18', 'FRG2B']

- genes near breakpoints documented in DDG2P:
RAB18(602207): WARBURG MICRO SYNDROME TYPE 3(614222)
ZMYND11(608668): INTELLECTUAL DISABILITY(616579)

- all genes with -1 CN: []

- genes with -1 CN documented in DDG2P: None

del(12)(q24.11)

- deletion on Chr12: 109408269(q24.11) - 109939632(q24.11)

- all genes near breakpoints: ['MYO1H', 'ACACB', 'MMAB', 'LINC01486', 'GIT2', 'LOC574538', 'MIR4497', 'SLC6A12', 'KCTD10', 'FAM222A', 'C12orf76', 'ANKRD13A', 'FOXN4', 'SLC6A12-AS1', 'MVK', 'WASH8P', 'IFT81', 'TCHP', 'GLTP', 'FAM222A-AS1', 'UBE3B', 'FAM138D', 'IQSEC3', 'TRPV4']

- genes near breakpoints documented in DDG2P:
MMAB(607568): METHYLMALONIC ACIDURIA TYPE CBLB(251110)
TRPV4(605427): SPONDYLOMETAPHYSEAL DYSPLASIA, KOZLOWSKI TYPE(184252),METATROPIC DYSPLASIA(156530)
UBE3B(608047): BLEPHAROPHIMOSIS-INTELLECTUAL DEVELOPMENTAL DISORDER(615057)

- all genes with -1 CN: ['MYO1H', 'KCTD10', 'UBE3B', 'MMAB', 'MVK', 'FAM222A', 'FAM222A-AS1', 'TRPV4', 'MIR4497', 'GLTP', 'TCHP', 'GIT2']

- genes with -1 CN documented in DDG2P:
MMAB(607568): METHYLMALONIC ACIDURIA TYPE CBLB(251110)
TRPV4(605427): SPONDYLOMETAPHYSEAL DYSPLASIA, KOZLOWSKI TYPE(184252),METATROPIC DYSPLASIA(156530)
UBE3B(608047): BLEPHAROPHIMOSIS-INTELLECTUAL DEVELOPMENTAL DISORDER(615057)