inv(7)(q11.23)

- inversion on Chr7: 73061114(q11.23) - 75291965(q11.23)

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

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

dup(4)(p16.1)

- tandem duplication on Chr4: 8929610(p16.1) - 9144574(p16.1)

- all genes near breakpoints: ['USP17L29', 'USP17L28', 'FAM90A26', 'USP17L12', 'USP17L18', 'USP17L21', 'USP17L24', 'USP17L30', 'USP17L27', 'USP17L26', 'USP17L20', 'USP17L11', 'USP17L25', 'USP17L5', 'USP17L17', 'USP17L19', 'USP17L10', 'USP17L22', 'USP17L13', 'HMX1', 'ZNF718', 'ZNF595', 'ZNF876P', 'USP17L15']

- genes near breakpoints documented in DDG2P:
HMX1(142992): OCULOAURICULAR SYNDROME(612109)

- all genes with +1 CN: []

- genes with +1 CN documented in DDG2P: None

dup(17)(p11.2)

- tandem duplication on Chr17: 21366522(p11.2) - 21692279(p11.2)

- all genes near breakpoints: ['LINC02693', 'MAP2K3', 'NATD1', 'KCNJ12', 'DOC2B', 'RPH3AL', 'TMEM11', 'LINC02091', 'DHRS7B', 'SCGB1C2', 'KCNJ18']

- genes near breakpoints documented in DDG2P: None

- all genes with +1 CN: ['KCNJ12', 'LINC02693']

- genes with +1 CN documented in DDG2P: None

del(22)(q11.23q12.1)

- deletion on Chr22: 25127033(q11.23) - 25515764(q12.1)

- all genes near breakpoints: ['SLC7A4', 'CRYBB2P1', 'BCRP2', 'RIMBP3C', 'CRYBB2', 'LRP5L', 'RIMBP3B', 'LRRC74B', 'FAM230H', 'IGLL3P', 'MIR6817', 'LZTR1', 'KIAA1671-AS1', 'MIR649', 'TUBA3FP', 'POM121L8P', 'SGSM1', 'P2RX6', 'KIAA1671', 'CRYBB3', 'THAP7-AS1', 'GGT2', 'GRK3', 'TMEM211', 'P2RX6P', 'THAP7', 'FAM230B']

- genes near breakpoints documented in DDG2P:

CRYBB2(123620): CATARACT, COPPOCK-LIKE(604307),CATARACT, CONGENITAL, CERULEAN TYPE, 2(601547)

CRYBB3(123630): CATARACT, CONGENITAL NUCLEAR, AUTOSOMAL RECESSIVE 2(609741)

LZTR1(600574): Noonan syndrome(No disease mim),NOONAN SYNDROME 10(616564)

- all genes with -1 CN: ['KIAA1671', 'CRYBB3', 'CRYBB2', 'IGLL3P', 'LRP5L', 'CRYBB2P1', 'MIR6817']

- genes with -1 CN documented in DDG2P:

CRYBB2(123620): CATARACT, COPPOCK-LIKE(604307),CATARACT, CONGENITAL, CERULEAN TYPE, 2(601547)

CRYBB3(123630): CATARACT, CONGENITAL NUCLEAR, AUTOSOMAL RECESSIVE 2(609741)