## del(1)(q21.1)

- deletion on Chr1: 143518643(q21.1) 144085065(q21.1)
- all genes near breakpoints: ['MIR1302-9', 'WASH7P', 'FAM72C', 'FAM138D', 'LOC101060524', 'H2BP2', 'LSP1P5', 'MIR6859-2', 'DDX11L1', 'MIR1302-11', 'LOC101927429', 'SRGAP2D', 'H3-2', 'FAM138C', 'RNVU1-17', 'LOC729737', 'MIR6859-4', 'MIR1302-10', 'MIR6859-3', 'FAM138A', 'FAM138F', 'MIR6859-1', 'DRD5P2', 'OR4F5', 'MIR1302-2', 'DDX11L17']
- genes near breakpoints documented in DDG2P: None
- all genes with -1 CN: ['RNVU1-17', 'RNVU1-18', 'RNU1-3', 'RNU1-4', 'RNU1-2', 'RNU1-1', 'LOC105369140', 'LINC02591', 'LOC644634', 'H2BP2', 'FCGR1CP', 'H3-2', 'FAM72C', 'SRGAP2D']
- genes with -1 CN documented in DDG2P: None

## del(15)(q13.2)

- deletion on Chr15: 30137048(q13.2) 30545741(q13.2)
- all genes near breakpoints: ['ULK4P3', 'GOLGA8R', 'WHAMMP1', 'DNM1P50', 'GOLGA8N', 'LOC100288203', 'GOLGA8IP', 'GOLGA8J', 'GOLGA8H', 'ULK4P1', 'ULK4P2', 'CHRFAM7A', 'LOC100996413', 'LOC102725021', 'GOLGA8Q', 'LINC02249', 'TJP1', 'CHRNA7', 'LOC100288637', 'LINC02256', 'ARHGAP11B', 'GOLGA8T']
- genes near breakpoints documented in DDG2P: None
- all genes with -1 CN: ['LOC102725021', 'GOLGA8T', 'LINC02249', 'CHRNA7', 'CHRFAM7A', 'DNM1P50', 'GOLGA8R', 'LOC100288203', 'WHAMMP1', 'LOC100996413', 'LINC02256']
- genes with -1 CN documented in DDG2P: None

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

- deletion on Chr22: 25200725(q11.23) 25560371(q12.1)
- all genes near breakpoints: ['RIMBP3B', 'THAP7', 'SLC7A4', 'GGT2', 'CRYBB3', 'P2RX6', 'KIAA1671', 'FAM230B', 'CRYBB2P1', 'FAM230H', 'MIR6817', 'KIAA1671-AS1', 'THAP7-AS1', 'MIR649', 'POM121L8P', 'LRP5L', 'TUBA3FP', 'GRK3', 'CRYBB2', 'RIMBP3C', 'MYO18B', 'LZTR1', 'LRRC74B', 'IGLL3P', 'P2RX6P', 'BCRP2']
- 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) MYO18B(607295): KLIPPEL-FEIL SYNDROME 4, AUTOSOMAL RECESSIVE, WITH NEMALINE

## MYOPATHY AND FACIAL DYSMORPHISM(616549)

- all genes with -1 CN: ['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)