Supplement Table 1. A total of 285 patients harbored P/LP variants distributed among 115 genes.

|  |  |  |
| --- | --- | --- |
| P/LP variants | No. of cases | Identified variants (no. of patients affected) |
| Variants in genes | 285 | *FGFR3*(31);*PTPN11*(24);*NF1*(13);*PHEX*(11);*KMT2A*(10);*IDS*(9);  *ACAN*(6);*COL2A1*(6);*NIPBL*(6);*GNAS*(6);*SLC12A3*(6);*SHOX*(4);*COL1A1*(4);  *ANKRD11*(4);*EXT1*(4);*RUNX2*(4);*GH1*(3);*GLI2*(3);*COMP*(3);*FGFR2*(3);*KMT2D*(3);  *SOS1*(3);*CASR*(3);*DUOX2*(2);*IGF1R*(2);*FGFR1*(2);*OBSL1*(2);*CHD7*(2);*KDM6A*(2);  *CREBBP*(2);*FAM111A*(2);*MAPK2K1*(2);*KAT6B*(2);*RAF1*(2);*SHOC2*(2);*CLCN7*(2);  *ARSB*(2);*GJB2*(2);*MED12*(2);*OFD1*(2);*POGZ*(2);*MECP2*(2);*TRPS1*(2);*TRVP4*  (2);*GNPTAB*(2);*SOX11*(2);*NPR2*(2);*GALNS*(2);*GHR*(1);*GSHR*(1);*SOX3*(1);*COL3A1*  (1);*COL10A1*(1);*COL11A1*(1);*FBN1*(1);*IHH*(1);*PTHLH*(1);*BLM*(1);*ERCC6*(1);  *FANCE*(1);*CUL7*(1);*HDAC8*(1);*SMC1A*(1);*SMC3*(1);*LMNA*(1);*ORC6*(1);*PCNT*  (1);*FGD1*(1);*HRAS*(1);*BRAF*(1);*KRAS*(1);*RIT1*(1);*RPS7*(1);*BBS2*(1);*NBAS*(1);  *NPHP4*(1);*GPD1*(1);*SBDS*(1);*SOX2*(1);*TRAPPC2*(1);*GHRHR*(1);*COL1A2*(1);  *G6PC*(1);*GRIN1*(1);*WISP3*(1);*TWIST1*(1);*PEX26*(1);*EFNB1*(1);*AMER1*(1);  *TFAP2A*(1);*KDM5C*(1);*POC1A*(1);*GJA1*(1);*TCIRG1*(1);*PAX3*(1);*PAX8*(1);*THRA*(1);  *TPO*(1);*TSHR*(1);*GUSB*(1);*PHKA2*(1);*SGSH*(1);*AR*(1);*ATP6V1B1*(1);*CYP27B1*(1);  *FLNA*(1);*FLNB*(1);*GATA6*(1);*KMT2C*(1);*LMX1B*(1);*MFN2*(1);*NOTCH2*(1);*PLCB4*(1);  *RYR1*(1);*SLC12A1*(1) |

Supplement Table 2. 83 patients were identified with P/LP copy number variations (CNV) or chromosomal abnormalities.

|  |  |  |
| --- | --- | --- |
| P/LP variants | No. of cases | Identified variants (no. of patients affected) |
| Chromosomal abnormalities and CNVs | 83 | 22q11.2 deletion syndrome (7)  Williams–Beuren syndrome (6)  1q24q25 deletion syndrome (4)  Chromosome 18p deletion syndrome (3)  Miller–Dieker lissencephaly syndrome (3)  Smith–Magenis syndrome (2)  del(1)(p36.33-p36.31)[hg19,(chr1:1,950,768-6,551,761)] (over 4500kb)(1)  del(1)(p36.33-p36.23 )[hg19,(chr1:955,503-7,829,766)](over 6800kb)(1)  dup(1)(q42.13-q44)[hg19,(chr1:229,566,993-247,612,406)](over 18000kb)(1)  dup(1)(q42.2-q44)[hg19,(chr1:230,838,270-247,612,406)](over 16000kb(1)  del(2)(p21-p16.3) [hg19,(chr2:47,596,287-49,381,666)](over 1800kb) (1)  del(2)(q24.2-q31.1)[hg19(chr2: (157205510-170457666)](13252kb) (1)  del(2)(q32.3-q33.2)[hg19(chr2: 193,730,505-204,701,813)](10971kb) (1)  del(2)(q37.3)[hg19,(chr2:239,152,679-242,708,231)](over 3500kb) (1)  del(2)(q37.3)[hg19,(chr2:239,969,864-242,708,231)](over 2700kb) (1)  dup(2)(p11.2-p13.2)[hg19,(chr2:71,693,832-86,565,206)] (over 15000kb) (1)  dup(2)(p25.3)[hg19,(chr2:1,417,233-3,750,260)](over 2300kb) (1)  dup(2)(p24.1-16.3)[hg19,(chr2:20,110,029-51,259,674)](over 31000kb) (1)  del(4)(p16.3)[hg19,(chr1:168,250,278-173,886,516)](over 5000kb) (1)  del(5)(p15.33)[hg19,(chr5:218,338-1,816,167)](over 1600kb) (1)  del(5)(p15.33-p15.1)[hg19,(chr5:218,338-16,617,094)](over 16000kb) (1)  dup(6)(p25.3-p22.3)[hg19,(chr6:391,739-18,155,400)](over 17000kb) (1)  del(6)(p21.1)[hg19,(chr6:41,126,244-43,021,683)](over 1900kb）(1)  del(6)(q22.1)[hg19,(chr6:116,440,085-117,923,705)](over 1400kb) (1)  del(6)(q25.1-q25.2)[hg19,(chr6:149,539,060-152,958,497)](over 3400kb) (1)  del(7)(q33-q35)[hg19,(chr7:137,761,205-144,533,146)](over 6700kb) (1)  del(7)(q33-q34)[hg19,(chr7:137,761,178-140,624,728)](over 2900kb) (1)  del(7)(q36.1-q36.3)[hg19,(chr7:150,642,044-157,210,133)](over 6500kb) (1)  del(8)(q23.3-q24.11)[hg19,(chr8:116,420,724-119,124,058)](over 2700kb) (1)  del(9)(q21.11-q21.31)[hg19,(chr9:71000154-83236029)](12236kb) (1)  del(9)(q31.2-q31.3)[hg19,(chr9:108,320,411-113,563,278)](over 5200kb) (1)  del(9)(q33.3-q34.11)[hg19,(chr9:129,376,722-131,395,944)](over 2000kb) (1)  dup(9)(q34.3)[hg19,(chr9:140,149,752-140,730,578)](over 500kb) (1)  del(10)(q26.13-26.2)[hg19,(chr10:124,221,041-127,511,837)] (over 3200kb) (1)  del(11)(q24.1-q25)[hg19,(chr11:122,942,714-134,135,746)](over 1200kb) (1)  del(12)(q24.31)[hg19,(chr12:122,755,981-124,246,301)](over 1400kb）(1)  del(12)(p13.33-p13.31) [hg19,(chr12:389,223-6,484,729)](over 6000kb) (1)  del(13)(q31.1-q32.1)[hg19,(79,314,118-96,544,277)](17230kb) (1)  del(13)(q34)(over 3000kb) (1)  del(15)(q11.2-q13.1)[hg19(23,707,494-28,525,454)](4818kb) (1)  del(15)(q26.3)[hg19,(chr15:99,191,768-101,792,137)] (over 2600kb) (1)  del(15)(q26.3)[hg19,(chr15:99,191,768-101,792,253)](over 1600kb) (1)  dup(16)(p13.11)[hg19,(chr16:15,737,124-16,317,328)](over 500kb) (1)  del(16)(p13.11)[hg19,(chr16:15,737,124-16,317,328)(over 500kb) (1)  dup(16)(p11.2)(over 300kb) (1)  dup(17)(p11.2)(over 3000kb) (1)  dup(18)(q23)[hg19,(chr18:77,439,801- 77,514,510)](over 200kb) (1)  dup(19)(p13.3)[hg19,(chr19:852,303-6,720,661)](over 5800kb) (1)  del(20)(p13)[hg19,(chr20:939,096-2,413,399)](over 1400kb) (1)  dup(21)(q22.3)[hg19,(chr21:46,305,869-47,865,682)](over 1500kb）(1)  del(22)(q13.2)[hg19,(chr22:41,488,614-41,924,993)](over 400kb) (1)  del(22)(q12.3-q13.1)[hg19,(chr22:36,649,117-38,380,539)](over 1700kb) (1)  dup(22)(q11.23)[(hg19)chr22: 23,648,768-24,995,964](1347kb）(1)  dup(22)(q11.23)[hg19,(chr22:23,915,313-24,924,358)](over 1000kb）(1)  del(X)(p22.33-p22.32)[(hg19chrX: 60,701- 4,664,247)] (over 4600kb)(include SHOX gene) (1)  del(X)(p22.33-p22.31)[(hg19chrX: 60,701- 6445238)] (6385kb)(include SHOX gene) (1)  dup(X)(q26.3-q28) [hg19,(chrX:135,067,586-149,841,616)] (over 14800kb)(1)  large deletion of Xp,large duplication of Xq(2)  45X(6)  47XXY(1)  45X/46XY mosaicisms(2)  duplication of ARID1B (1) |

Supplement Table 3. 39 male patients (46 XY) were diagnosed with cryptorchidism, 69.23% (27/39) patients were identified with P/LP variants.

|  |  |  |
| --- | --- | --- |
| P/LP variants | No. of cases | Identified variants (no. of patients affected) |
| Variants in genes | 20 | *PTPN11*(8)*,MAP2K1*(1)*,RAF1*(1),*SOS1*(1),*GH1*(1),*GLI2*(1),  *NIPBL*(1), *CREBBP*(1),*SOX11*(1),*KMT2A*(1),*BBS2*(1),  *PEX26*(1),*RYR1*(1) |
| Chromosomal abnormalities and CNVs | 7 | 47XXY (1)  del(18)(p11.31-p11.21)[hg19,(chr18:2,916,992-12,884,236)] (over10000kb)(1)  del(17)(p13.3) [hg19,(chr17:1,247,834-1,680,868)] (over 400kb)(1)  del(17)(p12-p11.2)[hg19(chr17:15,801,183-20,274,157)](4473kb)(1)  del(1)(q24.3-q25.3)[hg19(chr1:171,130,803-185,746,128)(14615kb)(1)  del(5)(p15.33-p15.1)[hg19,(chr5:218,338-16,617,094)](over 16000kb)(1)  dup(22)(q11.23)[(hg19)chr22: 23,648,768-24,995,964](1347kb)(1) |

Supplement Table 4. 71.43% (100/140) patients with intellectual disability or developmental delay were identified with P/LP variants. 48%(48/100) patients were identified with CNVs.

|  |  |  |
| --- | --- | --- |
| P/LP  variants | No. of cases | Identified variants (no. of patients affected) |
| CNV | 48 | Williams–Beuren syndrome (4)  22q11.2 deletion syndrome (3)  1q24q25 deletion syndrome (3)  Chromosome 18p deletion syndrome (2)  Smith–Magenis syndrome (2)  1p36 deletion syndrome (2)  dup(1)(q42.13-q44)[hg19,(chr1:229,566,993-247,612,406)](over 18000kb);del(15)(q26.3)[hg19,(chr15:99,191,768-101,792,253)](over 1600kb)(1)  dup(1)(q42.2-q44)[hg19,(chr1:230,838,270-247,612,406)](over 16000kb）(1)  del(2)(q24.2-q31.1)[hg19(chr2: (157205510-170457666)](13252kb) (1)  del(2)(q37.3)[hg19,(chr2:239,152,679-242,708,231)](over 3500kb) (1)  del(2)(q37.3)[hg19,(chr2:239,969,864-242,708,231)](over 2700kb);dup(9)(q34.3)[hg19,(chr9:140,149,752-140,730,578)](over 500kb) (1)  del(2)(p21-p16.3) [hg19,(chr2:47,596,287-49,381,666)](over 1800kb) (1)  dup(2)(p24.1-16.3)[hg19,(chr2:20,110,029-51,259,674)](over 31000kb) (1)  del(4)(p16.3)[hg19,(chr1:168,250,278-173,886,516)](over 5000kb);dup(21)(q22.3)[hg19,(chr21:46,305,869-47,865,682)](over 1500kb)(1)  del(5)(p15.33)[hg19,(chr5:218,338-1,816,167)](over 1600kb)(1)  del(5)(p15.33-p15.1)[hg19,(chr5:218,338-16,617,094)](over 16000kb)(1)  dup(6)(p25.3-p22.3)[hg19,(chr6:391,739-18,155,400)](over 17000kb) (1)  del(6)(q22.1)[hg19,(chr6:116,440,085-117,923,705)](over 1400kb)(1)  del(7)(q33-q35)[hg19,(chr7:137,761,205-144,533,146)](over 6700kb) (1)  del(7)(q33-q34)[hg19,(chr7:137,761,178-140,624,728)](over 2900kb) (1)  del(7)(q36.1-q36.3)[hg19,(chr7:150,642,044-157,210,133)](over 6500kb);dup(18)(q23)[hg19,(chr18:77,439,801- 77,514,510)](over 200kb) (1)  del(9)(q33.3-q34.11)[hg19,(chr9:129,376,722-131,395,944)](over 2000kb) (1)  del(9)(q31.2-q31.3)[hg19,(chr9:108,320,411-113,563,278)](over 5200kb) (1)  del(9)(q21.11-q21.31)[hg19,(chr9:71000154-83236029)](12236kb)(1)  del(10)(q26.13-26.2)[hg19,(chr10:124,221,041-127,511,837)] (over 3200kb) (1)  del(12)(q24.31)[hg19,(chr12:122,755,981-124,246,301)](over 1400kb)(1)  del(12)(p13.33-p13.31) [hg19,(chr12:389,223-6,484,729)](over 6000kb) (1)  del(13)(q31.1-q32.1)[hg19,(79,314,118-96,544,277)](17230kb) (1)  del(13)(q34)(over 3000kb) (1)  del(15)(q11.2-q13.1)[hg19(23,707,494-28,525,454)](4818kb) (1)  dup(16)(p13.11)[hg19,(chr16:15,737,124-16,317,328)](over 500kb) (1)  del(16)p13.11[hg19,(chr16:15,737,124-16,317,328)(over 500kb) (1)  dup(17)(p11.2)(over 3000kb) (1)  dup(19)(p13.3)[hg19,(chr19:852,303-6,720,661)](over 5800kb) (1)  del(20)(p13)[hg19,(chr20:939,096-2,413,399)](over 1400kb)(1)  dup(22)(q11.23)[(hg19)chr22: 23,648,768-24,995,964](1347kb) (1)  dup(22)(q11.23)[hg19,(chr22:23,915,313-24,924,358)](over 1000kb) (1)  duplication of *ARID1B* gene (1) |

Supplement Table 5. Detailed genetic results of 367 patients were identified with P/LP variants.

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| --- | --- | --- | --- | --- | --- | --- |
| Patient | Genetic results | | | | |  |
| 6135 | *GH1* | c.242\_243delCA,p.Ser81\*; | Homozygote; | |
| 6515 | *GH1* | c.291+1G>A; | Heterozygote; | |
| 10010 | *GH1* | c.240delC,p.Ser81Glnfs\*19;del Exon 1-5; | Compound heterozygote; | |
| 6500 | *GHR* | c.136+1G>A ; | Homozygote; | |
| 5410 | *GHRHR* | c.1147G>T,p.Val383Leu ; | Homozygote; | |
| 5175 | *GLI2* | c.3463\_3464delGA,p.Asp1155Argfs\*39 ; | Heterozygote; | |
| 5589 | *GLI2* | c.3137delG,p.Gly1046Alafs\*84; | Heterozygote; | |
| 6606 | *GLI2* | c.3640C>T,p.Gln1214\* ; | Heterozygote; | |
| 6845 | *GHSR* | c.107\_109del,p.Gln36del ; | Heterozygote; | |
| 9601 | *IGF1R* | c.179A>C,p.His60Pro ; | Heterozygote; | |
| 13921 | *IGF1R* | deletion (whole gene); |
| 3973 | *SOX3* | c.424C>A,p.Pro142Thr ; | Heterozygote; | |
| 2321 | *ACAN* | c.661delT,p.Tyr221Metfs\*10; | Heterozygote; | |
| 4212 | *ACAN* | c.436delC，p.Leu146Trpfs\*5; | Heterozygote; | |
| 14569 | *ACAN* | c.1180C>T, p.Arg394\* ; | Heterozygote; | |
| 11292 | *ACAN* | c.4486delG,p.Val1496\* ; | Heterozygote; | |
| 11550 | *ACAN* | c.1429+1delG ; | Heterozygote; | |
| 11565 | *ACAN* | c.11T>G,p.Leu4\* ; | Heterozygote; | |
| 6533 | *COL1A1* | c.1171G>A,p.Asp391Asn; | Heterozygote; | |
| 10661 | *COL1A1* | c.1588G>A,p.Gly530Ser ; | Heterozygote; | |
| 14156 | *COL1A1* | c.3421C>T, p.Arg1141\* ; | Heterozygote; | |
| 11006 | *COL1A1* | c.63\_64ins19,p.Gly22Glnfs\*6 ; | Heterozygote; | |
| 6954 | *COL1A2* | c.838G>A,p.Gly280Ser; | Heterozygote; | |
| 3832 | *COL2A1* | c.1826G>T,p.Gly609Val ; | Heterozygote; | |
| 4042 | *COL2A1* | c.1016G>A,p.Gly339Asp ; | Heterozygote; | |
| 6967 | *COL2A1* | c.1942-2A>G ; | Heterozygote; | |
| 13832 | *COL2A1* | c.3662C>T, p.Ser1221Phe ; | Homozygote; | |
| 13361 | *COL2A1* | c.905C>T, p.Ala302Val ; | Heterozygote; | |
| 13276 | *COL2A1* | c.823C>T, p.Arg275Cys ; | Heterozygote; | |
| 10032 | *COL3A1* | c.574G>A,p.Gly192Ser ; | Heterozygote; | |
| 13431 | *COL10A1* | c.1765T>C, p.Phe589Leu ; | Heterozygote; | |
| 13181 | *COL11A1* | c.1245+1G>A ; | Heterozygote; | |
| 2134 | *COMP* | c.976G>A,p.Asp326Asn; | Heterozygote; | |
| 7541 | *COMP* | c.1317C>G,p.Asp439Glu ; | Heterozygote; | |
| 14121 | *COMP* | c.1223A>G, p.Asp408Gly ; | Heterozygote; | |
| 8245 | *FBN1* | c.5284G>A,p.Gly1762Ser; | Heterozygote; | |
| 5621 | *FGFR1* | c.760C>T,p.Arg254Trp ; | Heterozygote; | |
| 7151 | *FGFR1* | c.1431-2A>C; | Heterozygote; | |
| 3478 | *FGFR2* | c.2324\_2332del,p.Pro775\_Glu777del ; | Heterozygote; | |
| 3514 | *FGFR2* | c.1026C>G,p.Cys342Trp ; | Heterozygote; | |
| 10422 | *FGFR2* | c.833G>T,p.Cys278Phe; | Heterozygote; | |
| 5721 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 6175 | *FGFR3* | c.1620C>A,p.Asn540Lys ; | Heterozygote; | |
| 7197 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 7419 | *FGFR3* | c.1138G>A,p.Gly380Arg; | Heterozygote; | |
| 7535 | *FGFR3* | c.1138G>A,p.Gly380Arg; | Heterozygote; | |
| 8305 | *FGFR3* | c.1138G>A,p.Gly380Arg; | Heterozygote; | |
| 9049 | *FGFR3* | c.1620C>G,p.Asn540Lys ; | Heterozygote; | |
| 8673 | *FGFR3* | c.833A>G,p.Tyr278Cys ; | Heterozygote; | |
| 10479 | *FGFR3* | c.791C>T,p.Thr264Met ; | Heterozygote; | |
| 12585 | *FGFR3* | c.1138G>A,p.Gly380Arg; | Heterozygote; | |
| 13193 | *FGFR3* | c.1138G>A, p.Gly380Arg; | Heterozygote; | |
| 12696 | *FGFR3* | c.1620C>A,p.Asn540Lys ; | Heterozygote; | |
| 13267 | *FGFR3* | c.1138G>A, p.Gly380Arg; | Heterozygote; | |
| 13843 | *FGFR3* | c.1620C>G, p.Asn540Lys ; | Heterozygote; | |
| 14365 | *FGFR3* | c.1620C>G, p.Asn540Lys ; | Heterozygote; | |
| 11935 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 12240 | *FGFR3* | c.1620C>G,p.Asn540Lys ; | Heterozygote; | |
| 12249 | *FGFR3* | c.1620C>A,p.Asn540Lys ; | Heterozygote; | |
| 2505 | *FGFR3* | c.1138G>A,p.Gly380Arg; | Heterozygote; | |
| 1899 | *FGFR3* | c.1620C>G,p.Asn540Lys ; | Heterozygote; | |
| 1969 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 2182 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 3031 | *FGFR3* | c.1620C>G,p.Asn540Lys ; | Heterozygote; | |
| 4230 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 4352 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 4554 | *FGFR3* | c.1619A>G,p.Asn540Ser ; | Heterozygote; | |
| 5144 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 5198 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 5407 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 5701 | *FGFR3* | c.1138G>A,p.Gly380Arg ; | Heterozygote; | |
| 3863 | *IHH* | c.797dupC,p.Arg267Thrfs\*15 ; | Heterozygote; | |
| 11354 | *FGFR3,NPR2* | *FGFR3*,c.1135T>C,p.Tyr379His ;  *NPR2*,c.2720C>T,p.Thr907Met; | Heterozygote;Heterozygote; | |
| 14081 | *NPR2* | c.329delG, p.Arg110Profs\*11 ; | Heterozygote; | |
| 5817 | *PTHLH* | c.101+1G>C ; | Heterozygote; | |
| WJ-525 | *BLM* | c.772\_773delCT , c.959+2T>A ; | Compound heterozygote; | |
| 10616 | *ERCC6* | c.643G>T,p.Glu215\*;  c.1607T>G,p.Leu536Trp; | Compound heterozygote; | |
| 3587 | *FANCE* | c.1111C>T,p.Arg371Trp ; | Homozygote; | |
| 6712 | *CUL7* | c.354delT,p.Gln119Argfs\*52;  c.40delC,p.Leu14Trpfs\*12; | Compound heterozygote; | |
| 2040 | *OBSL1* | c.458dupG,p.Leu154Profs\*100; | Homozygote; | |
| 7499 | *OBSL1* | c.458dupG,p.Leu154Profs\*100 ; | Homozygote; | |
| 7724 | *CHD7* | c.6193C>T,p.Arg2065Cys; | Heterozygote; | |
| 12343 | *CHD7* | c.5211-2A>G; | Heterozygote; | |
| 10868 | *HDAC8* | c.628+1G>C; | Heterozygote; | |
| WJ-861 | *NIPBL* | c.6854\_6855delAG,p.Gln2285Argfs\*3; | Heterozygote; | |
| 4734 | *NIPBL* | c.3344G>A,p.Trp1115\*; | Heterozygote; | |
| 7178 | *NIPBL* | c.4422G>T,p.Arg1474Ser; | Heterozygote; | |
| 7833 | *NIPBL* | c.7264-6T>G ; | Heterozygote; | |
| 9838 | *NIPBL* | c.330\_331delAA,p.Ser111Hisfs\*16; | Heterozygote; | |
| 11213 | *NIPBL* | c.-79-2A>G; | Heterozygote; | |
| 10475 | *SMC1A* | c.1088G>T,p.Arg363Ile; | Heterozygote; | |
| 12723 | *SMC3* | c.95G>A,p.Gly32Asp; | Heterozygote; | |
| 10635 | *LMNA* | c.433G>A , p.Glu145Lys ; | Heterozygote; | |
| 2066 | *ORC6* | c.67A>G,p.Lys23Glu; | Homozygote; | |
| 2258 | *PCNT* | c.3103C>T,p.Arg1035\*;c.502C>T,p.Gln168\*; | Compound heterozygote; | |
| 6556 | *KDM6A* | c.1834C>T,p.Arg612\* ; | Heterozygote; | |
| 7933 | *KDM6A* | c.404G>A,p.Gly135Asp; | Heterozygote; | |
| 6983 | *KMT2D* | c.16018C>T,p.Arg5340\*; | Heterozygote; | |
| 11639 | *KMT2D* | c.15163\_15168del,p.Asp5055\_Leu5056del ; | Heterozygote; | |
| 14242 | *KMT2D* | c.11839C>T,p.Gln3947\* ; | Heterozygote; | |
| 6230 | *ANKRD11* | c.6982dupC,p.Arg2328Profs\*204 ; | Heterozygote; | |
| 8816 | *ANKRD11* | c.3140\_3143del,p.Gln1047Argfs\*270 ; | Heterozygote; | |
| 12210 | *ANKRD11* | c.7569+1G>C ; | Heterozygote; | |
| 11966 | *ANKRD11* | c.7237C>T,p.Gln2413\* ; | Heterozygote; | |
| 4487 | *CREBBP* | c.1775G>A,p.Trp592\* ; | Heterozygote; | |
| 9125 | *CREBBP* | c.2881-13G>A ; | Heterozygote; | |
| 4830 | *SHOX* | c.526G>T,p.Glu176\* ; | Heterozygote; | |
| 11457 | *SHOX* | c.283dupT,p.Tyr95Leufs\*2 ; | Heterozygote; | |
| 13343 | *SHOX* | deletion (whole gene); |
| 8895 | *SHOX* | deletion (whole gene); |
| 5558 | *SOX11* | c.337T>C,p.Tyr113His ; | Heterozygote; | |
| 4213 | *KMT2A* | c.10752dupA,p.Gly3585Argfs\*8 ; | Heterozygote; | |
| 5341 | *KMT2A* | c.11716C>T,p.Arg3906Cys ; | Heterozygote; | |
| 6436 | *KMT2A* | c.3241C>T,p.Arg1081\* ; | Heterozygote; | |
| 6557 | *KMT2A* | c.5871T>A,p.Tyr1957\* ; | Heterozygote; | |
| 8569 | *KMT2A* | c.7371delT,p.Gly2458Valfs\*6; | Heterozygote; | |
| 8304 | *KMT2A* | c.3241C>T,p.Arg1081\* ; | Heterozygote; | |
| 10589 | *KMT2A* | c.3460C>T,p.Arg1154Trp ; | Heterozygote; | |
| 10925 | *KMT2A* | c.3461\_3473del,p.Arg1154Leufs\*17 ; | Heterozygote; | |
| 11344 | *KMT2A* | c.4086+2T>G ; | Heterozygote; | |
| 11962 | *KMT2A* | c.7849delA,p.Arg2617Glyfs\*24 ; | Heterozygote; | |
| 3793 | *FGD1* | c.1555C>T,p.Arg519Cys ; | Heterozygote; | |
| 2822 | *GNAS* | c.212+3\_212+6delAAGT ; | Heterozygote; | |
| 4103 | *GNAS* | c.314C>T,p.Thr105Ile ; | Heterozygote; | |
| 4881 | *GNAS* | c.1006C>T,p.Arg336Trp ; | Heterozygote; | |
| 6562 | *GNAS* | c.308T>C,p.Ile103Thr; | Heterozygote; | |
| 7246 | *GNAS* | c.565\_568del,p.Asp189Metfs\*14 ; | Heterozygote; | |
| 12628 | *GNAS* | c.308T>C,p.Ile103Thr ; | Heterozygote; | |
| 9056 | *FAM111A* | c.952dupA,p.Met318Asnfs\*11 ; | Heterozygote; | |
| 7780 | *FAM111A* | c.1706G>A,p.Arg569His; | Heterozygote; | |
| 6522 | *MAP2K1* | c.161T>C,p.Leu54Pro ; | Heterozygote; | |
| 6891 | *MAP2K1* | c.389A>G,p.Tyr130Cys ; | Heterozygote; | |
| 7015 | *HRAS* | c.34G>A,p.Gly12Ser ; | Heterozygote; | |
| 2541 | *KAT6B* | c.2636T>A,p.Leu879\* ; | Heterozygote; | |
| 9948 | *KAT6B* | c.3405dupT,p.Lys1136\* ; | Heterozygote; | |
| 3221 | *NF1* | c.281T>C,p.Leu94Pro ; | Heterozygote; | |
| 3985 | *NF1* | c.3445A>G,p.Met1149Val ; | Heterozygote; | |
| 4100 | *NF1* | c.1541\_1542delAG,p.Gln514Argfs\*43 ; | Heterozygote; | |
| 4837 | *NF1* | c.3256C>T,p.Gln1086\* ; | Heterozygote; | |
| 5671 | *NF1* | c.4180A>C,p.Asn1394His ; | Heterozygote; | |
| 5683 | *NF1* | c.4469T>C,p.Leu1490Pro ; | Heterozygote; | |
| 7824 | *NF1* | c.1586T>C,p.Leu529Pro ; | Heterozygote; | |
| 8633 | *NF1* | c.5605G>C,p.Gly1869Arg ; | Heterozygote; | |
| 9077 | *NF1* | c.6792C>A,p.Tyr2264\* ; | Heterozygote; | |
| 9127 | *NF1* | c.5749+1G>A ; | Heterozygote; | |
| 13983 | *NF1* | c.6789\_6792del, p.Tyr2264Thrfs\*5 ; | Heterozygote; | |
| 13815 | *NF1* | c.3610C>T, p.Arg1204Trp ; | Heterozygote; | |
| 11564 | *NF1* | c.4267A>G,p.Lys1423Glu ; | Heterozygote; | |
| 12945 | *BRAF* | c.739T>C,p.Phe247Leu ; | Heterozygote; | |
| 9097 | *KRAS* | c.458A>T,p.Asp153Val ; | Heterozygote; | |
| 3745 | *PTPN11* | c.1510A>G,p.Met504Val ; | Heterozygote; | |
| 4350 | *PTPN11* | c.1510A>G,p.Met504Val ; | Heterozygote; | |
| 5657 | *PTPN11* | c.922A>G,p.Asn308Asp ; | Heterozygote; | |
| 6758 | *PTPN11* | c.922A>G,p.Asn308Asp ; | Heterozygote; | |
| 8953 | *PTPN11* | c.923A＞G,p.Asn308Ser; | Heterozygote; | |
| 8394 | *PTPN11* | c.218C>T,p.Thr73Ile ; | Heterozygote; | |
| 8491 | *PTPN11* | c.1492C>T,p.Arg498Trp ; | Heterozygote; | |
| 8591 | *PTPN11* | c.188A>G,p.Tyr63Cys ; | Heterozygote; | |
| 8823 | *PTPN11* | c.922A>G,p.Asn308Asp ; | Heterozygote; | |
| 8824 | *PTPN11* | c.922A>G,p.Asn308Asp ; | Heterozygote; | |
| 5465 | *PTPN11* | c.182A>G,p.Asp61Gly ; | Heterozygote; | |
| 9217 | *PTPN11* | c.181G>A,p.Asp61Asn ; | Heterozygote; | |
| 9371 | *PTPN11* | c.417G>C,p.Glu139Asp ; | Heterozygote; | |
| 10342 | *PTPN11* | c.181G>A,p.Asp61Asn ; | Heterozygote; | |
| 10482 | *PTPN11* | c.1472C>A,p.Pro491His ; | Heterozygote; | |
| 10534 | *PTPN11* | c.922A>G,p.Asn308Asp ; | Heterozygote; | |
| 14222 | *PTPN11* | c.215C>G, p.Ala72Gly ; | Heterozygote; | |
| 13282 | *PTPN11* | c.184T>G, p.Tyr62Asp ; | Heterozygote; | |
| 13477 | *PTPN11* | c.922A>G, p.Asn308Asp ; | Heterozygote; | |
| 11149 | *PTPN11* | c.1502G>A,p.Arg501Lys ; | Heterozygote; | |
| 11855 | *PTPN11* | c.922A>G,p.Asn308Asp ; | Heterozygote; | |
| 11825 | *PTPN11* | c.922A>G,p.Asn308Asp ; | Heterozygote; | |
| 10892 | *PTPN11* | c.922A>G,p.Asn308Asp ; | Heterozygote; | |
| ＊ | *PTPN11* | c.844A＞G,p.Ile282Val; | Heterozygote; | |
| 8637 | *RAF1* | c.786T>G,p.Asn262Lys ; | Heterozygote; | |
| 8708 | *RAF1* | c.1837C>G,p.Leu613Val ; | Heterozygote; | |
| 11157 | *RIT1* | c.67A>C,p.Lys23Gln ; | Heterozygote; | |
| WJ-862 | *SHOC2* | c.4A>G , p.Ser2Gly ; | Heterozygote; | |
| 12429 | *SHOC2* | c.4A>G , p.Ser2Gly ; | Heterozygote; | |
| 4417 | *SOS1* | c.587C>T,p.Ser196Leu ; | Heterozygote; | |
| 6896 | *SOS1* | c.1656G>T,p.Arg552Ser ; | Heterozygote; | |
| 6944 | *SOS1* | c.1655G>A,p.Arg552Lys ; | Heterozygote; | |
| 5001 | *CLCN7* | c.896C>T,p.Ala299Val; | Heterozygote; | |
| 11795 | *CLCN7* | c.857G>A , p.Arg286Gln ; | Heterozygote; | |
| 13779 | *TCIRG1* | c.117+5G>A ;c.2236+6T>G; | Compound heterozygote; | |
| 2975 | *EXT1* | c.2098C>T,p.Gln700\*; | Heterozygote; | |
| 4111 | *EXT1* | c.1431dupC,p.Ser478Leufs\*43 ; | Heterozygote; | |
| 9162 | *EXT1* | c.1154T>A,p.Leu385\* ; | Heterozygote; | |
| 10426 | *EXT1* | c.1836G>A,p.Trp612\* ; | Heterozygote; | |
| 10346 | *DUOX2* | c.1462G>A,p.Gly488Arg ; | Heterozygote; | |
| 2221 | *DUOX2* | c.3329G>A,p.Arg1110Gln;  c.1310G>C,p.Gly437Ala; | Compound heterozygote; | |
| 8911 | *PAX3* | c.811C>T,p.Arg271Cys ; | Heterozygote; | |
| 8894 | *PAX8* | c.92G>A,p.Arg31His ; | Heterozygote; | |
| 8903 | *THRA* | c.802G>A,p.Asp268Asn ; | Heterozygote; | |
| 13050 | *TPO* | c.977C>T,p.Ala326Val;c.670\_672del,p.Asp224del; | Compound heterozygote; | |
| 5866 | *TSHR* | c.394G>C,p.Gly132Arg; c.1556G>A,p.Arg519His; | Compound heterozygote; | |
| 9328 | *ARSB* | c.1197C>G,p.Phe399Leu;c.943C>T,p.Arg315\*; | Compound heterozygote; | |
| 9887 | *ARSB* | c.200T>G,p.Ile67Ser; c.342C>G,p.Ile114Met; |
| 6479 | *G6PC* | c.648G>T,p.Leu216Le; c.248G>A,p.Arg83His; | Compound heterozygote; | |
| 9581 | *GNPTAB* | c.1090C>T,p.Arg364\* ; | Homozygote; | |
| 12021 | *GNPTAB* | c.1284+1G>T ;c.1307C>T,p.Pro436Leu; | Compound heterozygote; | |
| 9669 | *GUSB* | c.1244+3G>C;c.1324G>A,p.Ala442Thr; | Compound heterozygote; | |
| 3549 | *IDS* | c.1006+2T>G ; | Heterozygote; | |
| 3992 | *IDS* | c.1037C>T,p.Ala346Val ; | Heterozygote; | |
| 6607 | *IDS* | c.240+1G>A ; | Heterozygote; | |
| 8940 | *IDS* | c.820dupG,p.Glu274Glyfs\*68 ; | Heterozygote; | |
| 8976 | *IDS* | c.182C>T,p.Ser61Phe ; | Heterozygote; | |
| 9761 | *IDS* | c.943\_944dupTT,p.Leu315Phefs\*2 ; | Heterozygote; | |
| 13423 | *IDS* | c.507+1G>A ; | Heterozygote; | |
| 11189 | *IDS* | c.182C>T,p.Ser61Phe ; | Heterozygote; | |
| 11979 | *IDS* | del Exon 9; |  | |
| 6759 | *PHKA2* | c.1459+1G>A ; | Heterozygote; | |
| 13555 | *SGSH* | c.962C>A, p.Thr321Asn ; | Homozygote; | |
| 11072 | *AR* | c.2113C>T,p.Leu705Phe ; | Heterozygote; | |
| 14304 | *ATP6V1B1* | c.370C>T, p.Arg124Trp ;c.1397C>A, p.Ser466\*; | Heterozygote; | |
| 4743 | *CASR* | c.3082C>T,p.Gln1028\* ; | Heterozygote; | |
| 2671 | *CASR* | c.2405A>T,p.Asn802Ile ; | Heterozygote; | |
| 9153 | *CASR* | c.897G>A,p.Trp299\* ; | Heterozygote; | |
| 10775 | *CYP27B1* | c.1165C>T,p.Arg389Cys; c.589+1G>A; | Compound heterozygote; | |
| 12496 | *FLNA* | c.3527G>A,p.Gly1176Glu ; | Heterozygote; | |
| 13184 | *FLNB* | c.4241delC, p.Pro1414Leufs\*2 ; | Heterozygote; | |
| 9990 | *GATA6* | c.1366C>T,p.Arg456Cys ; | Heterozygote; | |
| 10666 | *GJB2* | c.235delC,p.Leu79Cysfs\*3; c.109G>A,p.Val37Ile; | Compound heterozygote; | |
| 11897 | *GJB2* | c.235delC,p.Leu79Cysfs\*3 ; | Homozygote; | |
| 9444 | *GRIN1* | c.1852G>A,p.Gly618Ser ; | Heterozygote; | |
| 13165 | *KMT2C* | c.3841+1G>A ; | Heterozygote; | |
| 6872 | *LMX1B* | c.248G>T,p.Cys83Phe ; | Heterozygote; | |
| 3573 | *MED12* | c.887G>C,p.Arg296Pro; | Heterozygote; | |
| 8071 | *MED12* | c.2881C>T,p.Arg961Trp ; | Heterozygote; | |
| 8821 | *MFN2* | c.384C>A,p.His128Gln ; | Heterozygote; | |
| 8229 | *NOTCH2* | c.6449\_6450delCT,p.Pro2150Argfs\*2 ; | Heterozygote; | |
| 7311 | *OFD1* | c.2delT ; | Heterozygote; | |
| 2867 | *OFD1* | c.2590C>T,p.Gln864\* ; | Heterozygote; | |
| 2791 | *PHEX* | c.1960\_1965+2dupTTTAGGGT ; | Heterozygote; | |
| 6386 | *PHEX* | del Exon 1-5; | Heterozygote; | |
| 8828 | *PHEX* | del Exon 13; | Heterozygote; | |
| 8926 | *PHEX* | c.2147+1\_2147+2delinsAGGGGC ; | Heterozygote; | |
| 6824 | *PHEX* | c.1971C>A,p.Tyr657\* ; | Heterozygote; | |
| 7398 | *PHEX* | c.776T>C,p.Leu259Pro ; | Heterozygote; | |
| 7404 | *PHEX* | c.1601C>T,p.Pro534Leu ; | Heterozygote; | |
| 7761 | *PHEX* | c.733-1G>A ; | Heterozygote; | |
| 8492 | *PHEX* | c.1971C>G,p.Tyr657\* ; | Heterozygote; | |
| 12638 | *PHEX* | c.350-1G>T ; | Heterozygote; | |
| 11777 | *PHEX* | c.1735G>A , p.Gly579Arg ; | Heterozygote; | |
| 5260 | *PLCB4* | c.2980delA,p.Met994\* ; | Heterozygote; | |
| 8524 | *POGZ* | c.3847C>T,p.Gln1283\* ; | Heterozygote; | |
| 8987 | *POGZ* | c.2310C>G,p.Tyr770\*; | Heterozygote; | |
| 7387 | *MECP2* | del Exon 2-3; | Heterozygote; | |
| 10505 | *MECP2* | c.905C>T,p.Pro302Leu ; | Heterozygote; | |
| 5867 | *RUNX2* | c.912delC,p.Ser305Profs\*3; | Heterozygote; | |
| 7950 | *RUNX2* | c.606delC,p.Val203Serfs\*8 ; | Heterozygote; | |
| 8044 | *RUNX2* | c.674G>A,p.Arg225Gln ; | Heterozygote; | |
| 13439 | *RUNX2* | c.673C>T, p.Arg225Trp ; | Heterozygote; | |
| 9882 | *RYR1* | c.7523G>A,p.Arg2508His ; | Heterozygote; | |
| 13700 | *SLC12A1* | c.2711delA, p.Lys904Argfs\*19;c.3096+1G>A; | Compound heterozygote; | |
| 5722 | *SLC12A3* | c.965-1\_976delinsACCGAAAATTTT ;(2) c.1456G>A,p.Asp486Asn ; | Compound heterozygote; | |
| 11249 | *SLC12A3* | c.836T>G,p.Met279Arg;c.602-16G>A; | Compound heterozygote; | |
| 10972 | *SLC12A3* | c.2877\_2878delAG,p.Arg959Serfs\*11;  c.179C>T,p.Thr60Met; | Compound heterozygote; | |
| 12583 | *SLC12A3* | c.179C>T,p.Thr60Met;  c.533C>T,p.Ser178Leu; | Compound heterozygote; | |
| 12760 | *SLC12A3* | c.911C>T,p.Thr304Met;  c.1445G>A,p.Cys482Tyr; | Compound heterozygote; | |
| 13591 | *SLC12A3* | c.1679C>A, p.Pro560His ; | Homozygote; | |
| 3254 | *TRPS1* | c.2657C>A,p.Ser886\* ; | Heterozygote; | |
| 5836 | *TRPS1* | c.2762G>A,p.Arg921Gln ; | Heterozygote; | |
| 6428 | *TRPV4* | c.1781G>A,p.Arg594His ; | Heterozygote; | |
| 12639 | *TRPV4* | c.1780C>A,p.Arg594Ser ; | Heterozygote; | |
| 5862 | *WISP3* | c.667T>C,p.Cys223Arg; | Homozygote; | |
| 2378 | *BBS2* | c.1148\_1149dupTC, p.His384Serfs\*34 ; | Homozygote; | |
| 12377 | *GALNS* | c.775C>T,p.Arg259Trp,  c.245C>T,p.Ser82Leu; | Compound heterozygote; | |
| 2568 | *GALNS* | c.106\_111del,p.Leu36\_Leu37del ;c  .812T>C,p.Leu271Pro; | Compound heterozygote; | |
| 2574 | *NBAS* | c.500\_501delTT,p.Phe167Cysfs\*7;  c.5752A>C,p.Thr1918Pro; | Compound heterozygote; | |
| 2896 | *TFAP2A* | c.202C>T,p.Gln68\* ; | Heterozygote; | |
| 3969 | *NPHP4* | c.3196C>T,p.Gln1066\* ; | Homozygote; | |
| 4582 | *GPD1* | c.220-2A>G;c.820G>A,p.Ala274Thr; | Compound heterozygote; | |
| 4774 | *PEX26* | c.354delC,p.Val120Serfs\*61;  c.34delC,p.Leu12Serfs\*70; | Compound heterozygote; | |
| 6701 | *EFNB1* | c.196C>T,p.Arg66\* ; | Heterozygote; | |
| 7290 | *RPS7* | c.75+2T>C ; | Heterozygote; | |
| 8061 | *AMER1* | c.301G>T,p.Glu101\* ; | Heterozygote; | |
| 8086 | *KDM5C* | c.4402G>T,p.Glu1468\* ; | Heterozygote; | |
| 9021 | *POC1A* | c.981+1G>A ; | Homozygote; | |
| 6746 | *GJA1* | c.715C>T,p.Arg239Trp ; | Heterozygote; | |
| 12978 | *SBDS* | c.258+2T>C ; | Homozygote; | |
| 14317 | *SOX2* | c.259A>G, p.Lys87Glu ; | Heterozygote; | |
| 12004 | *TRAPPC2* | c.271\_275del,p.Gln91Argfs\*9 ; | Heterozygote; | |
| 12446 | *TWIST1* | c.309C>G,p.Tyr103\* ; |  | |
| 7500 | del(1)(q24.2-25.1)[hg19,(chr1:169,433,149-173,827,682)] (over 4300kb)and  *SOX11*,c.425C>G,p.Ala142Gly;Heterozygote | | | | | |
| 13099 | del(22)(q11.21)[hg19(chr22:18,900,287-21,245,501)] (over 2300kb) | | | | | |
| 12594 | del(22)(q11.21)[hg19(chr22:18,900,287-21,245,501)] (over 2300kb) | | | | | |
| 10499 | del(22)(q11.21)[hg19(chr22:18,900,287-21,245,501)] (over 2300kb) | | | | | |
| 9260 | del(22)(q11.21)[hg19(chr22:18,900,287-21,245,501)] (over 2300kb) | | | | | |
| 8141 | del(22)(q11.21)[hg19(chr22:18,900,287-21,245,501)] (over 2300kb) | | | | | |
| 7611 | del(22)(q11.21)[hg19(chr22:18,900,287-21,245,501)] (over 2300kb) | | | | | |
| 3691 | del(22)(q11.21)[hg19(chr22: 18,910,683- 21,461,788)] ( 2551kb) | | | | | |
| 5428 | del(7)(q11.23) (over 1000kb) | | | | | |
| 5927 | del(7)(q11.23) (over 1000kb) | | | | | |
| 12367 | del(7)(q11.23)[hg19,(chr7:73,442,119-74,175,022)] (over 700kb) | | | | | |
| 13693 | del(7)(q11.23)[hg19,(chr7:73,442,119-74,175,022)] (over 700kb) | | | | | |
| 9578 | del(7)(q11.23)[hg19,(chr7:73,442,119-74,175,022)] (over 700kb) | | | | | |
| 12497 | del(7)(q11.23)[hg19,(chr7:73,442,119-74,175,022)] (over 700kb) | | | | | |
| 12480 | del(18)(p11.31-p11.21)[hg19,(chr18:2,916,992-12,884,236)] (over10000kb) | | | | | |
| 10850 | del(18)(p11.31-p11.21)[hg19,(chr18:2,916,992-12,884,236)] (over 9900kb) | | | | | |
| 8339 | del(18)(p11.31-p11.21)[hg19,(chr18:2,916,992-12,377,275)](over 9400kb) | | | | | |
| 8526 | del(17)(p13.3) [hg19,(chr17:1,247,834-1,680,868)] (over 400kb) | | | | | |
| 8551 | del(17)(p13.3)[hg19,(chr17:1,247,834-1,680,868)] (over 400kb) | | | | | |
| 14170 | del(17)(p13.3)[hg19,(chr17:411,908-1,948,259)] (over 1500kb) | | | | | |
| 13048 | del(17)(p11.2)[hg19,(chr17:17,115,527-19,580,909)] (over 2400kb) | | | | | |
| 3930 | del(17)(p12-p11.2)[hg19(chr17:15,801,183-20,274,157)](4473kb) | | | | | |
| 3660 | del(1)(q24.3-q25.3)[hg19(chr1:171,130,803-185,746,128)(14615kb) | | | | | |
| 6284 | del(1)(q24.2-q25.1)[hg19,(chr1:170,501,263-173,886,516)] (over 3300kb) | | | | | |
| 6409 | del(1)(q24.2-q25.1)[hg19,(chr1:168,250,278-173,886,516)](over 5000kb) | | | | | |
| 12612 | del(4)(p16.3)[hg19,(chr1:168,250,278-173,886,516)](over 5000kb);dup(21)(q22.3)[hg19,(chr21:46,305,869-47,865,682)](over 1500kb） | | | | | |
| 7765 | del(1)(p36.33-p36.31)[hg19,(chr1:1,950,768-6,551,761)] (over 4500kb) | | | | | |
| 9726 | dup(1)(q42.2-q44)[hg19,(chr1:230,838,270-247,612,406)](over 16000kb） | | | | | |
| 7951 | del(1)(p36.33-p36.23 )[hg19,(chr1:955,503-7,829,766)](over 6800kb) | | | | | |
| 12665 | dup(1)(q42.13-q44)[hg19,(chr1:229,566,993-247,612,406)](over 18000kb);del(15)(q26.3)[hg19,(chr15:99,191,768-101,792,253)](over 1600kb) | | | | | |
| 4041 | del(2)(q32.3-q33.2)[hg19(chr2: 193,730,505-204,701,813)](10971kb) | | | | | |
| 4557 | del(2)(q24.2-q31.1)[hg19(chr2: (157205510-170457666)](13252kb) | | | | | |
| 6378 | dup(2)(p25.3)[hg19,(chr2:1,417,233-3,750,260)](over 2300kb);del(11)(q24.1-q25)[hg19,(chr11:122,942,714-134,135,746)](over 1200kb) | | | | | |
| 8905 | del(2)(q37.3)[hg19,(chr2:239,152,679-242,708,231)](over 3500kb) | | | | | |
| 8884 | dup(2)(p24.1-16.3)[hg19,(chr2:20,110,029-51,259,674)](over 31000kb) | | | | | |
| 9400 | del(2)(q37.3)[hg19,(chr2:239,969,864-242,708,231)](over 2700kb);dup(9)(q34.3)[hg19,(chr9:140,149,752-140,730,578)](over 500kb) | | | | | |
| 6803 | dup(2)(p11.2-p13.2)[hg19,(chr2:71,693,832-86,565,206)] (over 15000kb) | | | | | |
| 11516 | del(2)(p21-p16.3) [hg19,(chr2:47,596,287-49,381,666)](over 1800kb) | | | | | |
| 7033 | del(5)(p15.33)[hg19,(chr5:218,338-1,816,167)](over 1600kb) | | | | | |
| 8284 | del(5)(p15.33-p15.1)[hg19,(chr5:218,338-16,617,094)](over 16000kb) | | | | | |
| 8142 | dup(6)(p25.3-p22.3)[hg19,(chr6:391,739-18,155,400)](over 17000kb) | | | | | |
| 7685 | del(6)(q22.1)[hg19,(chr6:116,440,085-117,923,705)](over 1400kb) | | | | | |
| 6640 | del(6)(p21.1)[hg19,(chr6:41,126,244-43,021,683)](over 1900kb） | | | | | |
| 11167 | del(6)(q25.1-q25.2)[hg19,(chr6:149,539,060-152,958,497)](over 3400kb) | | | | | |
| 10239 | del(7)(q33-q35)[hg19,(chr7:137,761,205-144,533,146)](over 6700kb) | | | | | |
| 6897 | del(7)(q33-q34)[hg19,(chr7:137,761,178-140,624,728)](over 2900kb) | | | | | |
| 12721 | del(7)(q36.1-q36.3)[hg19,(chr7:150,642,044-157,210,133)](over 6500kb);dup(18)(q23)[hg19,(chr18:77,439,801- 77,514,510)](over 200kb) | | | | | |
| 12260 | del(8)(q23.3-q24.11)[hg19,(chr8:116,420,724-119,124,058)](over 2700kb) | | | | | |
| 8164 | del(9)(q33.3-q34.11)[hg19,(chr9:129,376,722-131,395,944)](over 2000kb) | | | | | |
| 8720 | del(9)(q31.2-q31.3)[hg19,(chr9:108,320,411-113,563,278)](over 5200kb) | | | | | |
| 2882 | del(9)(q21.11-q21.31)[hg19,(chr9:71000154-83236029)](12236kb); | | | | | |
| 9057 | del(10)(q26.13-26.2)[hg19,(chr10:124,221,041-127,511,837)] (over 3200kb) | | | | | |
| 10424 | del(12)(q24.31)[hg19,(chr12:122,755,981-124,246,301)](over 1400kb） | | | | | |
| 13003 | del(12)(p13.33-p13.31) [hg19,(chr12:389,223-6,484,729)](over 6000kb) | | | | | |
| 7767 | del(13)(q31.1-q32.1)[hg19,(79,314,118-96,544,277)](17230kb) | | | | | |
| 8046 | del(13)(q34)(over 3000kb) | | | | | |
| 3626 | del(15)(q11.2-q13.1)[hg19(23,707,494-28,525,454)](4818kb) | | | | | |
| 7177 | del(15)(q26.3)[hg19,(chr15:99,191,768-101,792,137)] (over 2600kb) | | | | | |
| 8120 | dup(16)(p13.11)[hg19,(chr16:15,737,124-16,317,328)](over 500kb) | | | | | |
| 9951 | del(16)(p13.11)[hg19,(chr16:15,737,124-16,317,328)(over 500kb) | | | | | |
| 5766 | dup(16)(p11.2)(over 300kb) | | | | | |
| 5548 | dup(17)(p11.2)(over 3000kb) | | | | | |
| 13727 | dup(19)(p13.3)[hg19,(chr19:852,303-6,720,661)](over 5800kb) | | | | | |
| 5909 | del(20)(p13)[hg19,(chr20:939,096-2,413,399)](over 1400kb) | | | | | |
| 6338 | del(22)(q13.2)[hg19,(chr22:41,488,614-41,924,993)](over 400kb) | | | | | |
| 9055 | del(22)(q12.3-q13.1)[hg19,(chr22:36,649,117-38,380,539)](over 1700kb) | | | | | |
| 3977 | dup(22)(q11.23)[(hg19)chr22: 23,648,768-24,995,964](1347kb） | | | | | |
| 13661 | dup(22)(q11.23)[hg19,(chr22:23,915,313-24,924,358)](over 1000kb） | | | | | |
| 10822 | duplication of ARID1B | | | | |  |
| 13508 | dup(X)(q26.3-q28) [hg19,(chrX:135,067,586-149,841,616)] (over 14800kb) | | | | | |
| 13816 | large deletion of Xp,large duplication of Xq | | | | | |
| 6036 | 45X/46XY mosaicisms | | | | |  |
| 13120 | 47XXY | | |  | |  |
| 12368 | 45X/46XY mosaicisms | | | | |  |
| 10618 | 45X | | |  | |  |
| 10848 | large deletion of Xp,large duplication of Xq | | | | | |
| 13796 | 45X | | |  | |  |
| 4037 | 45X | | |  | |  |
| 8220 | 45X | | |  | |  |
| 9210 | 45X | | |  | |  |
| 9598 | 45X | | |  | |  |
| 4187 | del(X)(p22.33-p22.32)[(hg19chrX: 60,701- 4,664,247)] (over 4600kb)(include *SHOX gene*) | | | | | |
| 4057 | del(X)(p22.33-p22.31)[(hg19chrX: 60,701- 6445238)] (6385kb)(include SHOX gene) | | | | | |