**SUPPLEMENTARY MATERIAL**

[Supplementary Table 1. 70.97% (132/186) patients with facial dysmorphism were identified with P/LP variants, related to 53 genes. 2](#_Toc62568683)

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[Supplementary Table 7. 70.71% (99/140) patients with intellectual disability or developmental delay were identified with P/LP variants.48.48%(48/99) patients were identified with CNVs. 7](#_Toc62568691)

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[Supplementary Table 9. 143 of these patients presented with no other risk factors except for short stature (< −3 SDS), whereas 11.19% (16/143) of these patients harbored P/LP variants. 9](#_Toc62568693)

[Supplementary Table 10. Detailed genetic results of 364 patients were identified with P/LP variants. 10](#_Toc62568694)

# Supplementary Table 1. 70.97% (132/186) patients with facial dysmorphism were identified with P/LP variants, related to 53 genes.

|  |  |  |  |
| --- | --- | --- | --- |
| P/LP variants | No. of cases | | Identified variants (no. of patients affected) |
| Variants in genes | | 99 | *PTPN11*(17)*,FGFR3(10),NIPBL(6),IDS(5),KMT2A(4),COL2A1(3),FGFR2(3),SOS1(3),RAF1(2),KMT2D(2),OBSL1(2),CREBBP(2),GNPTAB*(2),*GH1(1),ACAN(1),COL1A1(1),COL3A1(1)BLM*(1)*,ERCC6*(1)*,CUL7*(1)*,CHD7*(1),*HDAC8*(1)*,SMC1A*(1),*SMC3*(1),*LMNA*(1),*ORC6*(1),*PCNT*(1),*KDM6A*(1),*ANKRD11*(1),*FGD1*(1)*FAM111A*(1)*MAP2K1*(1),*HRAS*(1),*KAT6B*(1),*BRAF*(1),*KRAS*(1),*RIT1*(1),*SHOC2*(1),*NBAS*(1),*TWIST1*(1),*EFNB1*(1),*TFAP2A*(1),*GJA1*(1),*PAX3*(1),*THRA*(1),*TPO*(1),*ARSB*(1) ,*GUSB*(1), *LMX1B*(1),*NOTCH2*(1),*OFD1*(1),*PLCB4(1),*  *POGZ*(1),*POC1A*(1) |
| Chromosomal abnormalities and CNVs | 33 | | Turner syndrome (3)  22q11.2 deletion syndrome (3)  Williams-Beuren syndrome (3)  Chromosome 18p deletion syndrome (3)  17p13.3 deletion syndrome (3)  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(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);del(15)(q26.3)[hg19,(chr15:99,191,768-101,792,253)](over 1600kb) (1)  del(2)(q32.3-q33.2)[hg19(chr2: 193,730,505-204,701,813)](10971kb) (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)  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(7)(q33-q35)[hg19,(chr7:137,761,205-144,533,146)](over 6700kb) (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(8)(q23.3-q24.11)[hg19,(chr8:116,420,724-119,124,058)](over 2700kb)(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(16)(p13.11)[hg19,(chr16:15,737,124-16,317,328)(over 500kb) (1)  dup(17)(p11.2)(over 3000kb) (1)  del(22)(q13.2)[hg19,(chr22:41,488,614-41,924,993)](over 400kb) (1)  dup(X)(q26.3-q28) [hg19,(chrX:135,067,586-149,841,616)] (over 14800kb) (1) |

# Supplementary Table 2. 54.17%(52/96) patients with DSD were identified with P/LP variants in our cohort.

|  |  |  |
| --- | --- | --- |
| P/LP variants | No. of cases | Identified variants (no. of patients affected) |
| Variants in genes | 35 | *PTPN11*(8),*GLI2*(3),*FGFR1*(2),*GH1*(1),*GHR*(1),*SOX3*(1),*ERCC6*(1),*CHD7*(1),  *NIPBL*(1),*ORC6*(1),*CREBBP*(1),*SOX11*(1),*KMT2A*(1),*GNAS*(1)  ,*MAP2K1*(1),*RAF1*(1),*SOS1*(1),*BBS2*(1),*SOX2*(1),*PEX26*(1),  *DUOX2*(1),*PAX8*(1), *AR*(1), *KMT2C*(1), *RYR1*(1), *SLC12A3*(1) |
| Chromosomal abnormalities and CNVs | 17 | Turner syndrome (4)  45X/46XY mosaicisms (2)  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)(p11.2)[hg19,(chr17:17,115,527-19,580,909)] (over 2400kb)(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)  del(7)(q33-q35)[hg19,(chr7:137,761,205-144,533,146)](over 6700kb)(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) |

# Supplementary 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) |

# Supplementary Table 4. 54.35% (50/92) patients with CHD were identified with P/LP variants, related to 15 genes, 9 CNVs and 1 chromosomal abnormalities.

|  |  |  |
| --- | --- | --- |
| P/LP  Variants | No. of cases | Identified variants (no. of patients affected) |
| Variants in  Genes | 32 | *PTPN11*(13)*,RAF1*(2)*,SHOC2(2),SOS1(2),ANKRD11(2) IDS(2),,FBN1(1),NIPBL(1),KMT2A(1),NF1(1),KRAS(1),RIT1(1),*  *PEX26(1),GNPTAB(1), GATA6(1)* |
| chromosomal abnormalities and CNV | 18 | 22q11.2 deletion syndrome (5)  Williams-Beuren syndrome (4)  Turner syndrome (2)  del(18)(p11.31-p11.21)[hg19,(chr18:2,916,992-12,884,236)](over 9900kb) (1)  del(17)(p13.3)[hg19,(chr17:1,247,834-1,680,868)](over 400kb) (1)  del(2)(q32.3-q33.2)[hg19(chr2:193,730,505-204,701,813)](10971kb) (1)  del(2)(q24.2-q31.1)[hg19(chr2:(157205510-170457666)](13252kb) (1)  dup(2)(p25.3)[hg19,(chr2:1,417,233-3,750,260)](over 2300kb) and del (11) (q24.1-q25) [hg19,(chr11:122,942,714-134,135,746)](over 1200kb)(1)  del(9)(q21.11-q21.31)[hg19,(chr9:71000154-83236029)](12236kb) (1)  del(13)(q31.1-q32.1)[hg19,(79,314,118-96,544,277)](17230kb) (1) |

# Supplementary Table 5. 65.53% (154/235) patients with skeletal dysplasia were identified with P/LP variants. Pathogenic variants were identified in 61 genes and 6 CNVs.

|  |  |  |
| --- | --- | --- |
| P/LP variants | No. of cases | Identified variants (no. of patients affected) |
| Variants in genes | 148 | *FGFR3*(29),*PHEX*(11),*PTPN11*(11),*IDS(9),COL2A1*(5),*SHOX*(4),*GNAS*(4),*RUNX2* (4), *EXT1* (4), *NF1*(2), *COMP* (3),*ACAN* (3), *COL1A1*(3),*FGFR2* (2), *FAM111A* (1),*CLCN7*(2), *ARSB* (2), *GNPTAB* (2),*TRPS1*(2)*TRPV4*(2),*OFD1*(2), *NPR2*(2),*WISP3*(1), *GLI2*(1),*COL1A2*(1),*COL3A1*(1),*COL10A1*(1), *COL11A1*(1),*FBN1*(1),*PTHLH*(1),*ERCC6*(1),*CUL7*(1),*OBSL1*(1),*HDAC8*(1),*LMNA*(1),*ANKRD11*(1),*HRAS*(1),*KRAS*(1),*RAF1*(1),*RIT1*(1),*SHOC2*(1),*SOS1*(1),*NBAS*(1),*SBDS*(1),*GALNS*(1),*TWIST1*(1),*EFNB1*(1),*GALNS*(1),*TCIRG1*(1),*TPO*(1), *GUSB*(1), *SGSH*(1),*CYP27B1*(1),*FLNA*(1), *FLNB(1)*, *LMX1B*(1), *NOTCH2*(1), *TRAPPC2*(1), *AMER1*(1)*MFN2*(1), *PLCB4*(1),*RYR1*(1) |
| CNVs | 6 | 22q11.2 deletion syndrome (1)  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)  del(2)(q37.3[hg19,(chr2:239,152,679-242,708,231)](over 3500kb) (1)  dup(2)(p11.2-p13.2)[hg19,(chr2:71,693,832-86,565,206)] (over 15000kb) (1)  del(8)(q23.3-q24.11)[hg19,(chr8:116,420,724-119,124,058)](over 2700kb) (1)  dup(X)(q26.3-q28) [hg19,(chrX:135,067,586-149,841,616)] (over 14800kb) (1) |

# Supplementary Table 6. 70.71% (99/140) patients with intellectual disability or developmental delay were identified with P/LP variants, related to 35 genes.

|  |  |  |
| --- | --- | --- |
| P/LP variants | Disorder/Phenotype | Identified variants (no. of patients affected) |
| Variants in genes | Wiedemann-Steiner Syndrome | *KMT2A*(5) |
| Cornelia de Lange syndrome | *NIPBL*(4) ,*HDAC8*(1), *SMC1A*(1), *SMC3*(1) |
| Kabuki syndrome | *KDM6A*(2), *KMT2D*(1) |
| KBG syndrome | *ANKRD11*(2) |
| Rubinstein–Taybi syndrome | *CREBBP*(2) |
| Cardio-facio-cutaneous syndrome | *MAP2K1*(2), *BRAF*(1) |
| Noonan syndrome | *PTPN11*(1), *RAF1*(2) |
| Costello syndrome | *HRAS* (1) |
| Say-Barber-Biesecker-Young-Simpson syndrome | *KAT6B*(2) |
| FG syndrome | *MED12*(2) |
| White-Sutton syndrome | *POGZ*(2) |
| Rett syndrome | *MECP2*(2) |
| Mucopolysaccharidosis | *IDS(1), SGSH*(1) |
| Hereditary distal renal tubular acidosis | *ATP6V1B1*(1) |
| Joubert Syndrome | *OFD1*(1) |
| Gitelman syndrome | *SLC12A3*(1) |
| Metaphyseal chondro-dysplasia, Schmid type | *COL10A1*(1) |
| Achondroplasia | *FGFR3*(1) |
| Cockayne syndrome | *ERCC6*(1) |
| MOPD II | *PCNT*(1) |
| Kenny-Caffey syndrome | *FAM111A*(1) |
| Bardet-Biedl syndrome | *BBS2*(1) |
| Saethre-Chotzen syndrome | *TWIST1*(1) |
| Claes-Jensen syndrome | *KDM5C*(1) |
| Osteopetrosis | *CLCN7*(1) |
| GRIN1-Related Neurodevelopmental Disorder | *GRIN1*(1) |
|  | MFN2 Hereditary Motor and Sensory Neuropathy | *MFN2*(1) |
|  | Auriculocondylar syndrome | *PLCB4*(1) |

# Supplementary Table 7. 70.71% (99/140) patients with intellectual disability or developmental delay were identified with P/LP variants.48.48%(48/99) 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,(chr13:79,314,118-96,544,277)](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) |

**Supplementary Table 8. 56.25%(9/16) patients with microcephaly were identified with P/LP variants, related to 6 genes and 4 CNVs.**

|  |  |  |
| --- | --- | --- |
| P/LP variants | Disorder/  Phenotype | Identified variants (no. of patients affected) |
| Variants in genes | Bloom syndrome | *BLM*(1) |
| Cockayne syndrome | *ERCC6*(1) |
| Cornelia de Lange syndrome | *NIPBL*(1), *SMC1A*(1) |
| MOPD II | *PCNT*(1) |
| Hereditary distal renal tubular acidosis | *ATP6V1B1*(1) |
| Microcephaly, facial dysmorphism,  development delay | del(2)(q24.2-q31.1)[hg19(chr2: (157205510-170457666)](13252kb)(1) |
| Microcephaly, facial dysmorphism,  development delay | 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) |
| Microcephaly,  development delay | del(10)(q26.13-26.2)[hg19,(chr10:124,221,041-127,511,837)] (over 3200kb)(1) |

**Supplementary Table 9. 143 of these patients presented with no other risk factors except for short stature (< −3 SDS), whereas 11.19% (16/143) of these patients harbored P/LP variants.**

|  |  |  |
| --- | --- | --- |
| P/LP variants | No. of cases | Identified variants (no. of patients affected) |
| Variants in genes | 12 | *KMT2A* (3), *FGFR3*(Hypochondroplasia) (2)*, IHH* (1)*, GNAS* (1)*, ACAN* (2)*, GHRHR* (1), *IGF1R* (1), *GHSR* (1) |
| Chromosomal abnormalities and CNVs | 4 | del(6)(q25.1-q25.2)[hg19,(chr6:149,539,060152,958,497)  ](over 3400kb) (1)  dup(16)(p11.2)(over 300kb) (1)  large deletion of Xp and large duplication of Xq(1)  del(X)(p22.33-p22.32)[(hg19(chrX: 60,701- 4,664,247)] (over 4600kb)(include *SHOX* gene) (1) |

# Supplementary Table 10. Detailed genetic results of 364 patients were identified with P/LP variants.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Patient | Genetic results | | | | | |  |
| 6135 | *GH1* | NM\_000515.4:c.242\_243del | p.(Ser81\*) | | | Homozygote; | |
| 6515 | *GH1* | NM\_000515.4:c.291+1G>A | p.? | | | Heterozygote | |
| 10010 | *GH1* | NM\_000515.4:[c.240del]  /[Exon 1-5 del] | [p.(Ser81Glnfs\*19)]  /[p.?] | | | Compound heterozygote | |
| 6500 | *GHR* | NM\_000163.4: c.136+1G>A | p.? | | | Homozygote; | |
| 5410 | *GHRHR* | NM\_000823.3:c.1147G>T | p.(Val383Leu) | | | Homozygote; | |
| 5175 | *GLI2* | NM\_005270.4:c.3463\_3464del | p.(Asp1155Argfs\*39) | | | Heterozygote | |
| 5589 | *GLI2* | NM\_005270.4:c.3137del | p.(Gly1046Alafs\*84) | | | Heterozygote | |
| 6606 | *GLI2* | NM\_005270.4: c.3640C>T | p.(Gln1214\*) | | | Heterozygote | |
| 6845 | *GHSR* | NM\_198407.2:c.107\_109del, | p.(Gln36del) | | | Heterozygote | |
| 9601 | *IGF1R* | NM\_000875.4:c.179A>C | p.(His60Pro) | | | Heterozygote | |
| 13921 | *IGF1R* | deletion (whole gene) |  |  | |
| 3973 | *SOX3* | NM\_005634.2: c.424C>A | p.(Pro142Thr) | | | Heterozygote | |
| 2321 | *ACAN* | NM\_013227.3:c.661del | p.(Tyr221Metfs\*10) | | | Heterozygote | |
| 4212 | *ACAN* | NM\_013227.3:c.436delC | p.(Leu146Trpfs\*5) | | | Heterozygote | |
| 14569 | *ACAN* | NM\_013227.3:c.1180C>T | p.(Arg394\*) | | | Heterozygote | |
| 11292 | *ACAN* | NM\_013227.3: c.4486del | p.(Val1496\*) | | | Heterozygote | |
| 11550 | *ACAN* | NM\_013227.3:c.1429+1del | p.? | | | Heterozygote | |
| 11565 | *ACAN* | NM\_013227.3:c.11T>G | p.(Leu4\*) | | | Heterozygote | |
| 6533 | *COL1A1* | NM\_000088.3:c.1171G>A | p.(Asp391Asn) | | | Heterozygote | |
| 10661 | *COL1A1* | NM\_000088.3 c.1588G>A | p.(Gly530Ser) | | | Heterozygote | |
| 14156 | *COL1A1* | NM\_000088.3:c.3421C>T | p.(Arg1141\* ) | | | Heterozygote | |
| 11006 | *COL1A1* | NM\_000088.3:c.63\_64ins19 | p.(Gly22Glnfs\*6 ) | | | Heterozygote | |
| 6954 | *COL1A2* | NM\_000089.3: [c.749G>T]  /[c.838G>A] | [p.(Gly250Val)]  /[p.(Gly280Ser)] | | | Heterozygote | |
| 3832 | *COL2A1* | NM\_001844.4:c.1826G>T | p.(Gly609Val) | | | Heterozygote | |
| 4042 | *COL2A1* | NM\_001844.4:c.1016G>A | p.(Gly339Asp) | | | Heterozygote | |
| 6967 | *COL2A1* | NM\_001844.4:c.1942-2A>G | p.? | | | Heterozygote | |
| 13832 | *COL2A1* | NM\_001844.4:c.3662C>T | p.(Ser1221Phe) | | | Homozygote; | |
| 13361 | *COL2A1* | NM\_001844.4:c.905C>T | p.(Ala302Val) | | | Heterozygote | |
| 13276 | *COL2A1* | NM\_001844.4:c.823C>T | p.(Arg275Cys) | | | Heterozygote | |
| 10032 | *COL3A1* | NM\_000090.3:c.574G>A | p.(Gly192Ser) | | | Heterozygote | |
| 13431 | *COL10A1* | NM\_000493.3:c.1765T>C | p.(Phe589Leu) | | | Heterozygote | |
| 13181 | *COL11A1* | NM\_001854.3:c.1245+1G>A | p.? | | | Heterozygote | |
| 2134 | *COMP* | NM\_000095.2:c.976G>A | p.(Asp326Asn) | | | Heterozygote | |
| 7541 | *COMP* | NM\_000095.2:c.1317C>G | p.(Asp439Glu) | | | Heterozygote | |
| 14121 | *COMP* | NM\_000095.2:c.1223A>G | p.(Asp408Gly) | | | Heterozygote | |
| 8245 | *FBN1* | NM\_000138.4:c.5284G>A | p.(Gly1762Ser) | | | Heterozygote | |
| 5621 | *FGFR1* | NM\_023110.2:c.760C>T | p.(Arg254Trp) | | | Heterozygote | |
| 7151 | *FGFR1* | NM\_023110.2:c.1431-2A>C | p.? | | | Heterozygote | |
| 3514 | *FGFR2* | NM\_000141.4:c.1026C>G | p.(Cys342Trp) | | | Heterozygote | |
| 10422 | *FGFR2* | NM\_000141.4:c.833G>T | p.(Cys278Phe) | | | Heterozygote | |
| 5721 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 6175 | *FGFR3* | NM\_000142.4:c.1620C>A | p.(Asn540Lys) | | | Heterozygote | |
| 7197 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 7419 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 7535 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 8305 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 9049 | *FGFR3* | NM\_000142.4:c.1620C>G | p.(Asn540Lys) | | | Heterozygote | |
| 8673 | *FGFR3* | NM\_000142.4: c.833A>G | p.(Tyr278Cys) | | | Heterozygote | |
| 10479 | *FGFR3* | NM\_000142.4: c.791C>T | p.(Thr264Met) | | | Heterozygote | |
| 12585 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 13193 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 12696 | *FGFR3* | NM\_000142.4:c.1620C>A | p.(Asn540Lys) | | | Heterozygote | |
| 13267 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 13843 | *FGFR3* | NM\_000142.4:c.1620C>G | p.(Asn540Lys) | | | Heterozygote | |
| 14365 | *FGFR3* | NM\_000142.4:c.1620C>G | p.(Asn540Lys) | | | Heterozygote | |
| 11935 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 12240 | *FGFR3* | NM\_000142.4:c.1620C>G | p.(Asn540Lys) | | | Heterozygote | |
| 12249 | *FGFR3* | NM\_000142.4:c.1620C>A | p.(Asn540Lys) | | | Heterozygote | |
| 2505 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 1899 | *FGFR3* | NM\_000142.4:c.1620C>G | p.(Asn540Lys) | | | Heterozygote | |
| 1969 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 2182 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 3031 | *FGFR3* | NM\_000142.4:c.1620C>G | p.(Asn540Lys) | | | Heterozygote | |
| 4230 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 4352 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 4554 | *FGFR3* | NM\_000142.4:c.1619A>G | p.(Asn540Ser) | | | Heterozygote | |
| 5144 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 5198 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 5407 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 5701 | *FGFR3* | NM\_000142.4:c.1138G>A | p.(Gly380Arg) | | | Heterozygote | |
| 3863 | *IHH* | NM\_002181.3:c.797dup | p.(Arg267Thrfs\*15 ) | | | Heterozygote | |
| 11354 | *FGFR3,NPR2* | NM\_000142.4:c.1135T>C;  NM\_003995.3:c.2720C>T | FGFR3:p.(Tyr379His);  NPR2:p.(Thr907Met) | | | Heterozygote;Heterozygote | |
| 14081 | *NPR2* | NM\_003995.3:c.329del | p.(Arg110Profs\*11) | | | Heterozygote | |
| 5817 | *PTHLH* | NM\_198965.1:c.101+1G>C | p.? | | | Heterozygote | |
| WJ-525 | *BLM* | NM\_000057.3:[c.959+2T>A]  /[c.772\_773del] | [p.?]/[p.(Leu258Glufs\*7)] | | | Compound heterozygote | |
| 10616 | *ERCC6* | NM\_000124.3:[c.643G>T]  /[c.1607T>G] | [p.(Glu215\*)]  /[p.(Leu536Trp)] | | | Compound heterozygote | |
| 3587 | *FANCE* | NM\_021922.2:c.1111C>T | p.(Arg371Trp) | | | Homozygote; | |
| 6712 | *CUL7* | NM\_014780.4:[c.354delT]  /[c.40delC] | [p.(Gln119Argfs\*52)]  /[p.(Leu14Trpfs\*12)] | | | Compound heterozygote | |
| 2040 | *OBSL1* | NM\_015311.2:c.458dup | p.(Leu154Profs\*100) | | | Homozygote; | |
| 7499 | *OBSL1* | NM\_015311.2:c.458dup | p.(Leu154Profs\*100) | | | Homozygote; | |
| 4417 | *CHD7* | NM\_017780.3: c.6193C>T | p.(Arg2065Cys) | | | Heterozygote | |
| 7724 | *CHD7* | NM\_017780.3: c.5211-2A>G | p.? | | | Heterozygote | |
| 12343 | *CHD7* | NM\_017780.3:c.4964A>G | p.(Lys1655Arg) | | | Heterozygote | |
| 10868 | *HDAC8* | NM\_018486.2: c.628+1G>C | p.? | | | Heterozygote | |
| WJ-861 | *NIPBL* | NM\_133433.3:c.6854\_6855del | p.(Gln2285Argfs\*3) | | | Heterozygote | |
| 4734 | *NIPBL* | NM\_133433.3:c.3344G>A | p.(Trp1115\*) | | | Heterozygote | |
| 7178 | *NIPBL* | NM\_133433.3:c.4422G>T | p.(Arg1474Ser) | | | Heterozygote | |
| 7833 | *NIPBL* | NM\_133433.3:c.7264-6T>G | p.? | | | Heterozygote | |
| 9838 | *NIPBL* | NM\_133433.3:c.330\_331delAA | p.(Ser111Hisfs\*16) | | | Heterozygote | |
| 11213 | *NIPBL* | NM\_133433.3:c.-79-2A>G | p.? | | | Heterozygote | |
| 10475 | *SMC1A* | NM\_006306.3:c.1088G>T | p.(Arg363Ile) | | | Heterozygote | |
| 12723 | *SMC3* | NM\_005445.3:c.95G>A | p.(Gly32Asp) | | | Heterozygote | |
| 10635 | *LMNA* | NM\_170707.3:c.433G>A | p.(Glu145Lys) | | | Heterozygote | |
| 2066 | *ORC6* | NM\_014321.3:c.67A>G | p.(Lys23Glu) | | | Homozygote; | |
| 2258 | *PCNT* | NM\_006031.5:[c.3103C>T]  /[c.502C>T] | [p.(Arg1035\*)]  /[p.(Gln168\*)] | | | Compound heterozygote | |
| 6556 | *KDM6A* | NM\_021140.3:c.1834C>T | p.(Arg612\* ) | | | Heterozygote | |
| 7933 | *KDM6A* | NM\_021140.3:c.404G>A | p.(Gly135Asp) | | | Heterozygote | |
| 6983 | *KMT2D* | NM\_003482.3:c.16018C>T | p.(Arg5340\* ) | | | Heterozygote | |
| 11639 | *KMT2D* | NM\_003482.3:c.15163\_15168del | p.(Asp5055\_Leu5056del) | | | Heterozygote | |
| 14242 | *KMT2D* | NM\_003482.3:c.11839C>T | p.(Gln3947\* ) | | | Heterozygote | |
| 6230 | *ANKRD11* | NM\_013275.5:c.6982dup | p.(Arg2328Profs\*204 ) | | | Heterozygote | |
| 8816 | *ANKRD11* | NM\_013275.5:c.3140\_3143del | p.(Gln1047Argfs\*270 ) | | | Heterozygote | |
| 12210 | *ANKRD11* | NM\_013275.5:c.7569+1G>C | p.? | | | Heterozygote | |
| 11966 | *ANKRD11* | NM\_013275.5:c.7237C>T | p.(Gln2413\* ) | | | Heterozygote | |
| 4487 | *CREBBP* | NM\_004380.2:c.1775G>A | p.(Trp592\*) | | | Heterozygote | |
| 9125 | *CREBBP* | NM\_004380.2:c.2881-13G>A | p.? | | | Heterozygote | |
| 4830 | *SHOX* | NM\_000451.3:c.526G>T | p.(Glu176\*) | | | Heterozygote | |
| 11457 | *SHOX* | NM\_000451.3:c.283dupT | p.(Tyr95Leufs\*2) | | | Heterozygote | |
| 13343 | *SHOX* | deletion (whole gene) |  |  | |
| 8895 | *SHOX* | deletion (whole gene) |  |  | |
| 5558 | *SOX11* | NM\_003108.3:c.337T>C | p.(Tyr113His) | | | Heterozygote | |
| 4213 | *KMT2A* | NM\_001197104.1:c.10752dup | p.(Gly3585Argfs\*8) | | | Heterozygote | |
| 5341 | *KMT2A* | NM\_001197104.1:c.11716C>T | p.(Arg3906Cys) | | | Heterozygote | |
| 6436 | *KMT2A* | NM\_001197104.1:c.3241C>T | p.(Arg1081\*) | | | Heterozygote | |
| 6557 | *KMT2A* | NM\_001197104.1:c.5871T>A | p.(Tyr1957\*) | | | Heterozygote | |
| 8569 | *KMT2A* | NM\_001197104.1:c.7371del | p.(Gly2458Valfs\*6) | | | Heterozygote | |
| 8304 | *KMT2A* | NM\_001197104.1:c.3241C>T | p.(Arg1081\*) | | | Heterozygote | |
| 10589 | *KMT2A* | NM\_005933.3:c.3460C>T | p.(Arg1154Trp) | | | Heterozygote | |
| 10925 | *KMT2A* | NM\_001197104.1:c.3461\_3473del | p.(Arg1154Leufs\*17) | | | Heterozygote | |
| 11344 | *KMT2A* | NM\_001197104.1:c.4086+2T>G | p.? | | | Heterozygote | |
| 11962 | *KMT2A* | NM\_001197104.1:c.7849del | p.(Arg2617Glyfs\*24) | | | Heterozygote | |
| 2822 | *GNAS* | NM\_000516.4:c.212+3\_212+  6delAAGT | p.? | | | Heterozygote | |
| 4103 | *GNAS* | NM\_000516.4:c.314C>T | p.(Thr105Ile) | | | Heterozygote | |
| 4881 | *GNAS* | NM\_000516.4: c.1006C>T | p.(Arg336Trp) | | | Heterozygote | |
| 6562 | *GNAS* | NM\_000516.4:c.308T>C | p.(Ile103Thr) | | | Heterozygote | |
| 7246 | *GNAS* | NM\_000516.4:c.565\_568del | p.(Asp189Metfs\*14) | | | Heterozygote | |
| 12628 | *GNAS* | NM\_000516.5:c.308T>C | p.(Ile103Thr) | | | Heterozygote | |
| 7780 | *FAM111A* | NM\_022074.3:c.1706G>A | p.(Arg569His) | | | Heterozygote | |
| 6522 | *MAP2K1* | NM\_002755.3:c.161T>C | p.(Leu54Pro) | | | Heterozygote | |
| 6891 | *MAP2K1* | NM\_002755.3:c.389A>G | p.(Tyr130Cys) | | | Heterozygote | |
| 7015 | *HRAS* | NM\_005343.2:c.34G>A | p.(Gly12Ser) | | | Heterozygote | |
| 2541 | *KAT6B* | NM\_012330.3:c.2636T>A | p.(Leu879\*） | | | Heterozygote | |
| 9948 | *KAT6B* | NM\_012330.3:c.3405dup | p.(Lys1136\*) | | | Heterozygote | |
| 3221 | *NF1* | NM\_000267.3:c.281T>C | p.(Leu94Pro) | | | Heterozygote | |
| 3985 | *NF1* | NM\_000267.3:c.3445A>G | p.(Met1149Val) | | | Heterozygote | |
| 4100 | *NF1* | NM\_000267.3:c.1541\_1542del | p.(Gln514Argfs\*43) | | | Heterozygote | |
| 4837 | *NF1* | NM\_000267.3: c.3256C>T | p.(Gln1086\*) | | | Heterozygote | |
| 5671 | *NF1* | NM\_000267.3: c.4180A>C | p.(Asn1394His) | | | Heterozygote | |
| 5683 | *NF1* | NM\_000267.3c.4469T>C | p.(Leu1490Pro) | | | Heterozygote | |
| 7824 | *NF1* | NM\_000267.3:c.1586T>C | p.(Leu529Pro) | | | Heterozygote | |
| 8633 | *NF1* | NM\_000267.3:c.5605G>C | p.(Gly1869Arg) | | | Heterozygote | |
| 9077 | *NF1* | NM\_000267.3:c.6792C>A | p.(Tyr2264\*) | | | Heterozygote | |
| 9127 | *NF1* | NM\_000267.3:c.5749+1G>A | p.? | | | Heterozygote | |
| 13983 | *NF1* | NM\_000267.3:c.6789\_6792del | p.(Tyr2264Thrfs\*5) | | | Heterozygote | |
| 13815 | *NF1* | NM\_000267.3:c.3610C>T | p.(Arg1204Trp) | | | Heterozygote | |
| 11564 | *NF1* | NM\_000267.3:c.4267A>G | p.(Lys1423Glu) | | | Heterozygote | |
| 12945 | *BRAF* | NM\_004333.5:c.739T>C | p.(Phe247Leu) | | | Heterozygote | |
| 9097 | *KRAS* | NM\_004985.4:c.458A>T | p.(Asp153Val) | | | Heterozygote | |
| 3745 | *PTPN11* | NM\_002834.3:c.1510A>G | p.(Met504Val) | | | Heterozygote | |
| 4350 | *PTPN11* | NM\_002834.3:c.1510A>G | p.(Met504Val) | | | Heterozygote | |
| 5657 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| 6758 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| 8953 | *PTPN11* | NM\_002834.3:c.923A＞G | p.(Asn308Ser) | | | Heterozygote | |
| 8394 | *PTPN11* | NM\_002834.3:c.218C>T | p.(Thr73Ile) | | | Heterozygote | |
| 8491 | *PTPN11* | NM\_002834.3:c.1492C>T | p.(Arg498Trp) | | | Heterozygote | |
| 8591 | *PTPN11* | NM\_002834.3:c.188A>G | p.(Tyr63Cys) | | | Heterozygote | |
| 8823 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| 8824 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| 5465 | *PTPN11* | NM\_002834.3:c.182A>G | p.(Asp61Gly） | | | Heterozygote | |
| 9217 | *PTPN11* | NM\_002834.4:c.181G>A | p.(Asp61Asn) | | | Heterozygote | |
| 9371 | *PTPN11* | NM\_002834.4:c.417G>C | p.(Glu139Asp) | | | Heterozygote | |
| 10342 | *PTPN11* | NM\_002834.4:c.181G>A | p.(Asp61Asn) | | | Heterozygote | |
| 10482 | *PTPN11* | NM\_002834.4:c.1472C>A | p.(Pro491His) | | | Heterozygote | |
| 10534 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| 14222 | *PTPN11* | NM\_002834.3:c.215C>G | p.(Ala72Gly) | | | Heterozygote | |
| 13282 | *PTPN11* | NM\_002834.4:c.184T>G | p.(Tyr62Asp) | | | Heterozygote | |
| 13477 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| 11149 | *PTPN11* | NM\_002834.4:c.1502G>A | p.(Arg501Lys) | | | Heterozygote | |
| 11855 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| 11825 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| 10892 | *PTPN11* | NM\_002834.3:c.922A>G | p.(Asn308Asp) | | | Heterozygote | |
| ＊ | *PTPN11* | NM\_002834.3:c.844A＞G | p.(Ile282Val) | | | Heterozygote | |
| 8637 | *RAF1* | NM\_002880.3:c.786T>G | p.(Asn262Lys) | | | Heterozygote | |
| 8708 | *RAF1* | NM\_002880.3:c.1837C>G | p.(Leu613Val) | | | Heterozygote | |
| 11157 | *RIT1* | NM\_006912.5:c.67A>C | p.(Lys23Gln) | | | Heterozygote | |
| WJ-862 | *SHOC2* | NM\_007373.3:c.4A>G | p.(Ser2Gly) | | | Heterozygote | |
| 12429 | *SHOC2* | NM\_007373.3:c.4A>G | p.(Ser2Gly) | | | Heterozygote | |
| 6896 | *SOS1* | NM\_005633.3:c.1656G>T | p.(Arg552Ser) | | | Heterozygote | |
| 6944 | *SOS1* | NM\_005633.3:c.1655G>A | p.(Arg552Lys) | | | Heterozygote | |
| 5001 | *CLCN7* | NM\_001287.5:c.896C>T | p.(Ala299Val) | | | Heterozygote | |
| 11795 | *CLCN7* | NM\_001287.5:c.857G>A | p.(Arg286Gln) | | | Heterozygote | |
| 13779 | *TCIRG1* | NM\_006019.3:[c.117+5G>A]  /[c.2236+6T>G] | [p.?]/[p.?] | | | Compound heterozygote | |
| 2975 | *EXT1* | NM\_000127.2:c.2098C>T | p.(Gln700\*) | | | Heterozygote | |
| 4111 | *EXT1* | NM\_000127.2:c.1431dupC | p.(Ser478Leufs\*43) | | | Heterozygote | |
| 9162 | *EXT1* | NM\_000127.2:c.1154T>A | p.(Leu385\*) | | | Heterozygote | |
| 10426 | *EXT1* | NM\_000127.2:c.1836G>A | p.(Trp612\*) | | | Heterozygote | |
| 10346 | *DUOX2* | NM\_014080.4:c.1462G>A | p.(Gly488Arg) | | | Homozygote; | |
| 2221 | *DUOX2* | NM\_014080.4:[c.3329G>A]  /[c.1310G>C] | [p.(Arg1110Gln)]  /[p.(Gly437Ala)] | | | Compound heterozygote | |
| 8911 | *PAX3* | NM\_181457.3:c.811C>T | p.(Arg271Cys) | | | Heterozygote | |
| 8894 | *PAX8* | NM\_003466.3:c.92G>A | p.(Arg31His) | | | Heterozygote | |
| 8903 | *THRA* | NM\_199334.3:c.802G>A | p.(Asp268Asn) | | | Heterozygote | |
| 13050 | *TPO* | NM\_000547.5:[c.977C>T]  /[c.670\_672del] | [p.(Ala326Val)]  /[p.(Asp224del)] | | | Compound heterozygote | |
| 5866 | *TSHR* | NM\_000369.2:[c.394G>C]  /[c.1556G>A] | [p.(Gly132Arg)]  /[p.(Arg519His)] | | | Compound heterozygote | |
| 9328 | *ARSB* | NM\_000046.4:[c.1197C>G]  /[c.943C>T] | [p.(Phe399Leu)]  /[p.(Arg315\*)] | | | Compound heterozygote | |
| 9887 | *ARSB* | NM\_000046.4:[c.200T>G]  /[c.342C>G]/[c.475C>T] | [p.(Ile67Ser)]  /[p.(Ile114Met)]  /[p.(Arg159Cys)] | | | Compound heterozygote | |
| 6479 | *G6PC* | NM\_000151.3:[c.648G>T]  /[c.248G>A] | [p.(Leu216Leu)]  /[p.(Arg83His)] | | | Compound heterozygote | |
| 9581 | *GNPTAB* | NM\_024312.4:c.1090C>T | p.(Arg364\*) | | | Homozygote; | |
| 12021 | *GNPTAB* | NM\_024312.4:c.[1284+1G>T]  /[1307C>T] | [p.?]/[p.(Pro436Leu)] | | | Compound heterozygote | |
| 9669 | *GUSB* | NM\_000181.3:c.[1244+3G>C]  /[1324G>A] | [p.?]/[p.(Ala442Thr)] | | | Compound heterozygote | |
| 3549 | *IDS* | NM\_000202.6:c.1006+2T>G | p.? | | | Heterozygote | |
| 3992 | *IDS* | NM\_000202.6:c.1037C>T | p.(Ala346Val) | | | Heterozygote | |
| 6607 | *IDS* | NM\_000202.6:c.240+1G>A | p.? | | | Heterozygote | |
| 8940 | *IDS* | NM\_000202.6:c.820dup | p.(Glu274Glyfs\*68) | | | Heterozygote | |
| 8976 | *IDS* | NM\_000202.6:c.182C>T | p.(Ser61Phe) | | | Heterozygote | |
| 9761 | *IDS* | NM\_000202.6:c.943\_944dup | p.(Leu315Phefs\*2) | | | Heterozygote | |
| 13423 | *IDS* | NM\_000202.6:c.507+1G>A | p.? | | | Heterozygote | |
| 11189 | *IDS* | NM\_000202.6:c.182C>T | p.(Ser61Phe) | | | Heterozygote | |
| 11979 | *IDS* | NM\_000202.6:[Exon 9 del] | p.? | | |  | |
| 6759 | *PHKA2* | NM\_000292.2:c.1459+1G>A | p.? | | | Heterozygote | |
| 13555 | *SGSH* | NM\_000199.4:c.962C>A | p.(Thr321Asn) | | | Homozygote; | |
| 11072 | *AR* | NM\_000044.4:c.2113C>T | p.(Leu705Phe) | | | Heterozygote | |
| 14304 | *ATP6V1B1* | NM\_001692.4:[c.370C>T]  /[c.1397C>A] | [p.(Arg124Trp)]  /[p.(Ser466\*)] | | | Heterozygote | |
| 4743 | *CASR* | NM\_000388.3:c.3082C>T | p.(Gln1028\*) | | | Heterozygote | |
| 2671 | *CASR* | NM\_000388.3:c.2405A>T | p.(Asn802Ile) | | | Heterozygote | |
| 9153 | *CASR* | NM\_000388.3:c.897G>A | p.(Trp299\*) | | | Heterozygote | |
| 10775 | *CYP27B1* | NM\_000785.3:[c.1165C>T]  /[c.589+1G>A] | [p.(Arg389Cys)]  /[p.?] | | | Compound heterozygote | |
| 12496 | *FLNA* | NM\_001456.3:c.3527G>A | p.(Gly1176Glu) | | | Heterozygote | |
| 13184 | *FLNB* | NM\_001457.3:c.4241del | p.(Pro1414Leufs\*2) | | | Heterozygote | |
| 9990 | *GATA6* | NM\_005257.5:c.1366C>T | p.(Arg456Cys) | | | Heterozygote | |
| 10666 | *GJB2* | NM\_004004.5:[c.235del]  /[c.109G>A] | [p.(Leu79Cysfs\*3)]  /[p.(Val37Ile)] | | | Compound heterozygote | |
| 11897 | *GJB2* | NM\_004004.5:c.235del | p.(Leu79Cysfs\*3) | | | Homozygote; | |
| 9444 | *GRIN1* | NM\_007327.3:c.1852G>A | p.(Gly618Ser) | | | Heterozygote | |
| 13165 | *KMT2C* | NM\_170606.3:c.3841+1G>A | p.? | | | Heterozygote | |
| 6872 | *LMX1B* | NM\_002316.3:c.248G>T | p.(Cys83Phe) | | | Heterozygote | |
| 3573 | *MED12* | NM\_005120.2:c.887G>C | p.(Arg296Pro) | | | Heterozygote | |
| 8071 | *MED12* | NM\_005120.2:c.2881C>T | p.(Arg961Trp) | | | Heterozygote | |
| 8821 | *MFN2* | NM\_014874.3:c.384C>A | p.(His128Gln) | | | Heterozygote | |
| 8229 | *NOTCH2* | NM\_024408.3:c.6449\_6450del | p.(Pro2150Argfs\*2) | | | Heterozygote | |
| 7311 | *OFD1* | NM\_003611.2:c.2del | p.? | | | Heterozygote | |
| 2867 | *OFD1* | NM\_003611.2:c.2590C>T | p.(Gln864\*) | | | Heterozygote | |
| 2791 | *PHEX* | NM\_000444.5:c.1960\_1965  +2dupTTTAGGGT | p.? | | | Heterozygote | |
| 6386 | *PHEX* | NM\_000444.5:[Exon 1-5 del] | p.? | | | Heterozygote | |
| 8828 | *PHEX* | NM\_000444.5:[Exon 13 del] | p.? | | | Heterozygote | |
| 8926 | *PHEX* | NM\_000444.5:c.2147+1\_2147  +2delinsAGGGGC | p.? | | | Heterozygote | |
| 6824 | *PHEX* | NM\_000444.5:c.1971C>A | p.(Tyr657\*) | | | Heterozygote | |
| 7398 | *PHEX* | NM\_000444.5:c.776T>C | p.(Leu259Pro) | | | Heterozygote | |
| 7404 | *PHEX* | NM\_000444.5:c.1601C>T | p.(Pro534Leu) | | | Heterozygote | |
| 7761 | *PHEX* | NM\_000444.5:c.733-1G>A | p.? | | | Heterozygote | |
| 8492 | *PHEX* | NM\_000444.5:c.1971C>G | p.(Tyr657\*) | | | Heterozygote | |
| 12638 | *PHEX* | NM\_000444.5:c.350-1G>T | p.? | | | Heterozygote | |
| 11777 | *PHEX* | NM\_000444.5:c.1735G>A | p.(Gly579Arg) | | | Heterozygote | |
| 5260 | *PLCB4* | NM\_000933.3:c.2980delA | p.(Met994\*) | | | Heterozygote | |
| 8524 | *POGZ* | NM\_015100.3:c.3847C>T | p.(Gln1283\*) | | | Heterozygote | |
| 8987 | *POGZ* | NM\_015100.3:c.2310C>G | p.(Tyr770\*) | | | Heterozygote | |
| 7387 | *MECP2* | NM\_004992.3:[Exon 2-3 del] | p.? | | | Heterozygote | |
| 10505 | *MECP2* | NM\_004992.3:c.905C>T | p.(Pro302Leu) | | | Heterozygote | |
| 5867 | *RUNX2* | NM\_001024630.3:c.912delC | p.(Ser305Profs\*3) | | | Heterozygote | |
| 7950 | *RUNX2* | NM\_001024630.3:c.606delC | p.(Val203Serfs\*8) | | | Heterozygote | |
| 8044 | *RUNX2* | NM\_001024630.3:c.674G>A | p.(Arg225Gln) | | | Heterozygote | |
| 13439 | *RUNX2* | NM\_001024630.3:c.673C>T | p.(Arg225Trp) | | | Heterozygote | |
| 9882 | *RYR1* | NM\_000540.2:c.7523G>A | p.(Arg2508His) | | | Heterozygote | |
| 13700 | *SLC12A1* | NM\_000338.2:[c.2711delA]  /[c.3096+1G>A] | [p.(Lys904Argfs\*19)]  /[p.?] | | | Compound heterozygote | |
| 5722 | *SLC12A3* | NM\_000339.2:[c.965-1\_976delinsACCGAAAATTTT]  /[c.1456G>A] | [p.?]/[p.(Asp486Asn)] | | | Compound heterozygote | |
| 11249 | *SLC12A3* | NM\_000339.2:[c.836T>G]  /[c.602-16G>A] | [p.(Met279Arg)]/[p.?] | | | Compound heterozygote | |
| 10972 | *SLC12A3* | NM\_000339.2:[c.2877\_2878delAG]  /[c.179C>T] | [p.(Arg959Serfs\*11)]  /[p.(Thr60Met)] | | | Compound heterozygote | |
| 12583 | *SLC12A3* | NM\_000339.2:[c.179C>T]  /[c.533C>T] | [p.(Thr60Met)]  /[p.(Ser178Leu)] | | | Compound heterozygote | |
| 12760 | *SLC12A3* | NM\_000339.2:[c.911C>T]  /[c.1445G>A] | [p.(Thr304Met)]  /[(p.Cys482Tyr)] | | | Compound heterozygote | |
| 13591 | *SLC12A3* | NM\_000339.2:c.1679C>A | p.(Pro560His) | | | Homozygote; | |
| 3254 | *TRPS1* | NM\_014112.4:c.2657C>A | p.(Ser886\*) | | | Heterozygote | |
| 5836 | *TRPS1* | NM\_014112.4:c.2762G>A | p.(Arg921Gln) | | | Heterozygote | |
| 6428 | *TRPV4* | NM\_021625.4:c.1781G>A | p.(Arg594His) | | | Heterozygote | |
| 12639 | *TRPV4* | NM\_021625.4:c.1780C>A | p.(Arg594Ser) | | | Heterozygote | |
| 5862 | *WISP3* | NM\_003880.3:c.667T>C | p.(Cys223Arg) | | | Homozygote; | |
| 2378 | *BBS2* | NM\_031885.3:c.1148\_1149dup | p.(His384Serfs\*34) | | | Homozygote; | |
| 12377 | *GALNS* | NM\_000512.4:[c.775C>T]  /[c.245C>T] | [p.(Arg259Trp)]  /[p.(Ser82Leu)] | | | Compound heterozygote | |
| 2568 | *GALNS* | NM\_000512.4:[c.106\_111del]  /[c.812T>C] | [p.(Leu36\_Leu37del)]  /[p.(Leu271Pro)] | | | Compound heterozygote | |
| 2574 | *NBAS* | NM\_015909.3:[c.500\_501del]  /[c.5752A>C] | [p.(Phe167Cysfs\*7)]  /[p.(Thr1918Pro)] | | | Compound heterozygote | |
| 2896 | *TFAP2A* | NM\_003220.2:c.202C>T | p.(Gln68\*) | | | Heterozygote | |
| 3969 | *NPHP4* | NM\_015102.4:c.3196C>T | p.(Gln1066\*) | | | Homozygote; | |
| 4582 | *GPD1* | NM\_005276.3:[c.220-2A>G]  /[c.820G>A] | [p.?]/[p.(Ala274Thr)] | | | Compound heterozygote | |
| 4774 | *PEX26* | NM\_017929.5:[c.354delC]  /[c.34del] | [p.(Val120Serfs\*61)]  /[p.(Leu12Serfs\*70)] | | | Compound heterozygote | |
| 6701 | *EFNB1* | NM\_004429.4:c.196C>T | p.(Arg66\*) | | | Heterozygote | |
| 7290 | *RPS7* | NM\_021140.3:c.75+2T>C | p.? | | | Heterozygote | |
| 8061 | *AMER1* | NM\_152424.3:c.301G>T | p.(Glu101\*) | | | Heterozygote | |
| 8086 | *KDM5C* | NM\_004187.3:c.4402G>T | p.(Glu1468\*) | | | Heterozygote | |
| 9021 | *POC1A* | NM\_015426.4:c.981+1G>A | p.? | | | Homozygote; | |
| 6746 | *GJA1* | NM\_000165.4:c.715C>T | p.(Arg239Trp) | | | Heterozygote | |
| 12978 | *SBDS* | NM\_016038.3:c.258+2T>C | p.? | | | Homozygote; | |
| 14317 | *SOX2* | NM\_003106.3:c.259A>G | p.(Lys87Glu) | | | Heterozygote | |
| 12004 | *TRAPPC2* | NM\_001011658.3:c.271\_275del | p.(Gln91Argfs\*9) | | | Heterozygote | |
| 12446 | *TWIST1* | NM\_000474.3:c.309C>G | p.(Tyr103\*) | | | Heterozygote | |
| 7500 | del(1)(q24.2-25.1)[hg19,(chr1:169,433,149-173,827,682)] (over 4300kb)and  *SOX11* NM\_003108.3: 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) and 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) | | | | | | |