Supplement Table S1. Insertions/deletions (InDels) and single nucleotide variants (SNVs) identified by whole-exome sequencing after filtering dbSNP, single-nucleotide polymorphism database; MAF, minor allele frequency; EVS, exome variant server; Eur.Am., European American; Afr.Am., African American; ExAC, exome aggregation consortium; AFR, African; AMR, American; EAS, East Asian; SAS, South Asian; NFE, Non-Finnish European; FIN, Finnish; OTH, Other; SIFT, Sorting Intolerant from Tolerant; UCSC, UCSC Genome Browser.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Chrom** | **Position** | **Gene** | **refseq** | **Protein Variation** | **Consequence** | **Genotype** | **HGVSc** |
| chr1 | 1247578 | *PUSL1* | NM\_153339.1 | . | downstream\_gene\_variant | -/G | . |
| chr1 | 1247578 | *CPSF3L* | NM\_001256456.1 | . | intron\_variant | -/G | c.1625+27dupC |
| chr1 | 1247578 | *ACAP3* | NM\_030649.2 | . | upstream\_gene\_variant | -/G | . |
| chr1 | 111390641 |  |  |  |  | G/- |  |
| chr4 | 85465390 |  |  |  |  | TT/- |  |
| chr9 | 33796609 | *PRSS3* | NM\_001197098.1 | . | intron\_variant | CTTGC/- | c.20-28\_20-24delGCCTT |
| chr10 | 125780759 | *CHST15* | NM\_001270764.1 | . | intron\_variant | CC | c.1347+13C>G |
| chr10 | 125780759 | *CHST15* | NM\_001270765.1 | p.(P454A) | missense\_variant | CC | c.1360C>G |
| chr10 | 135105877 | *TUBGCP2* | NM\_006659.3 | . | intron\_variant | CCCCGTGTCCCCGTGTCCCGGGGAGCCCTACCTGTACGGGAGAGGGCAGCATGCACACTCCGTGTCCCCCATGTCCCTCCGTGTCCCCGTGTCCCTCCGTGT/- | c.1214+24\_1214+125delACACGGAGGGACACGGGGACACGGAGGGACATGGGGGACACGGAGTGTGCATGCTGCCCTCTCCCGT |
| **chr14** | **76147885** | ***TTLL5*** | **NM\_015072.4** | **p.(E61D\*19)** | **splice\_region\_variant** | **TAG/AA** | **c.182-3\_182-1delinsAA** |
| chr16 | 71004385 | *HYDIN* | NM\_001270974.1 | . | intron\_variant | -/AAGGCCAGGAGGATGTTATTGCATGCAGAGAAAAGAGCATGAGTACATAGCAGGGTCCAGC | c.5619+37\_5619+38insGCTGGACCCTGCTATGTACTCATGCTCTTTTCTCTGCATGCAATAACATCCTCCTGGCCTT |
| chr17 | 46115083 | *COPZ2* | NM\_016429.2 | p.(Q19HR) | protein\_altering\_variant | -/CTG | c.56\_57insCAG |
| chr17 | 73781075 | *UNK* | NM\_001080419.2 | . | intron\_variant | C/- | c.104+23delC |
| chr20 | 33033065 | *ITCH* | NM\_001257137.1 | . | intron\_variant | -/T | c.1089-10dupT |
| chr20 | 62597247 | *ZNF512B* | NM\_020713.2 | . | intron\_variant | GACCTGGGACGAGCCCCCATACCTTTTCTTACCACTGTTCCCTGACCTGGGACGAGCCCCCATACCTTTTCTCACCACTGTTCCTCCCCGACCTGGGACGAGCCCCCATACCTTTTCTCACCACTGTTCCCCGACCTGGGACGAGCCCCCATACCTTTTCTTACCACTGTTCCTCCCTGACCTGGGACGAGCCCCCATACCTTTTCTTACCACTGTTCCTCCCC/- | c.1034+23\_1034+246delGGGGAGGAACAGTGGTAAGAAAAGGTATGGGGGCTCGTCCCAGGTCAGGGAGGAACAGTGGTAAGAA |