Figure. S1

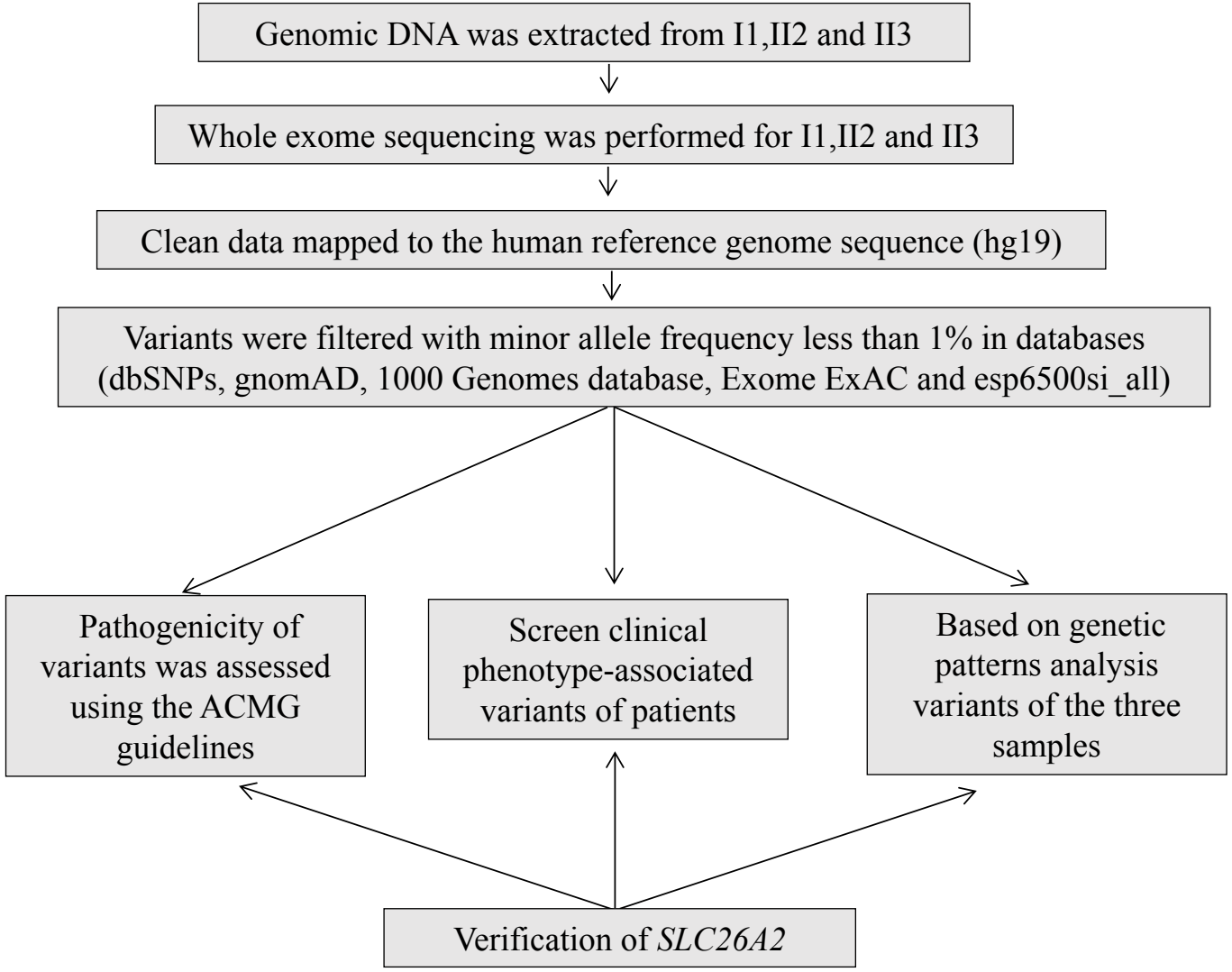
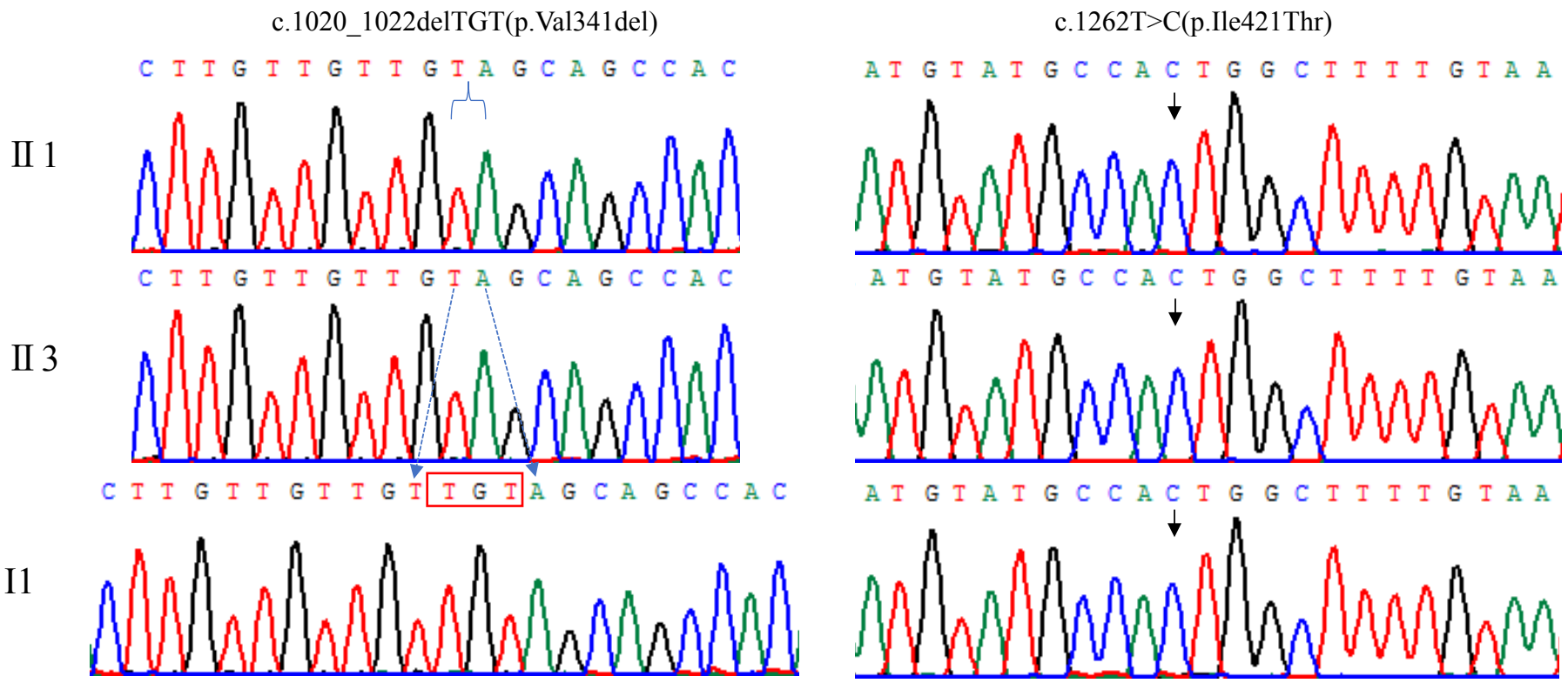

Genomic DNA was extracted from I1,II2 and II3
Whole exome sequencing was performed for I1,II2 and II3
Clean data mapped to the human reference genome sequence (hg19)
Variants were filtered with minor allele frequency less than 1% in databases (dbSNPs, gnomAD, 1000 Genomes database, Exome ExAC and esp6500si_all)
Pathogenicity of variants was assessed using the ACMG guidelines
Screen clinical phenotype-associated variants of patients
Based on genetic patterns analysis variants of the three samples
Verification of SLC26A2

Figure. S2


c.1020_1022delTGT(p.Val341del)
c.1262T>C(p.Ile421Thr)
Ⅱ 1
Ⅱ 3
I1
C T T G T T G T T G T A G C A G C C A C
C T T G T T G T T G T A G C A G C C A C
C T T G T T G T T G T T G T A G C A G C C A C
A T G T A T G C C A C T G G C T T T T G T A A
A T G T A T G C C A C T G G C T T T T G T A A
A T G T A T G C C A C T G G C T T T T G T A A