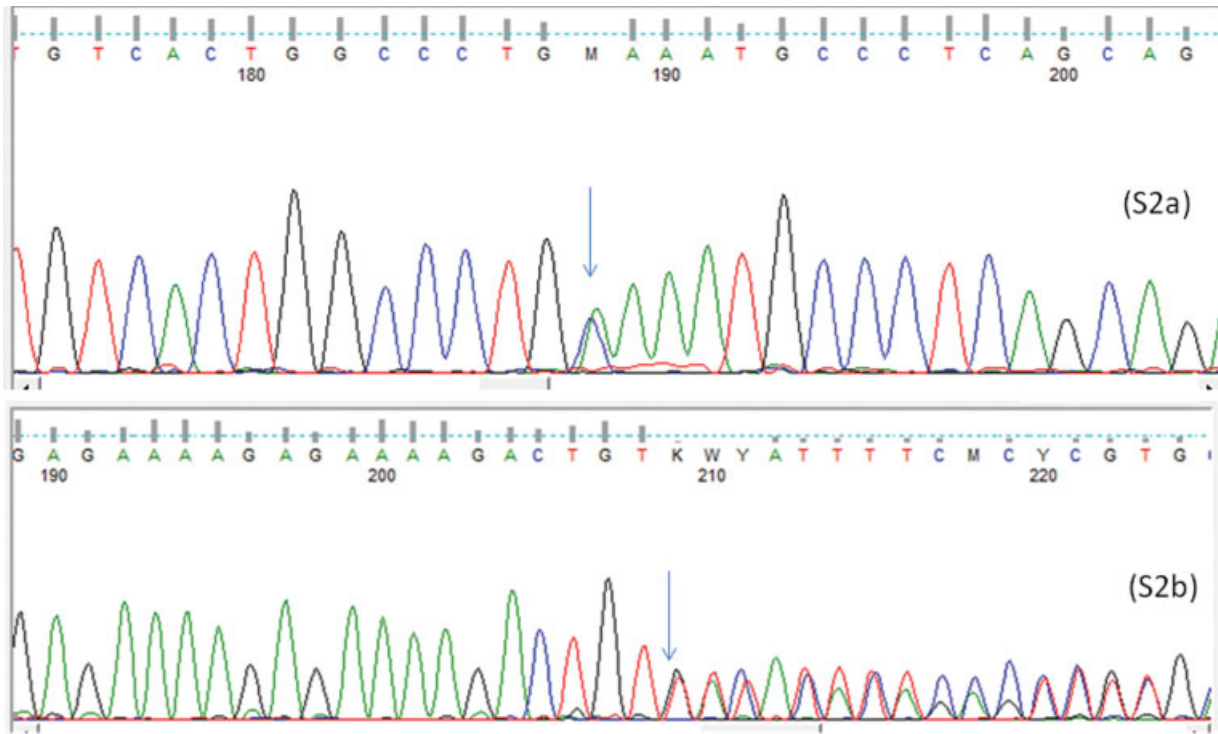
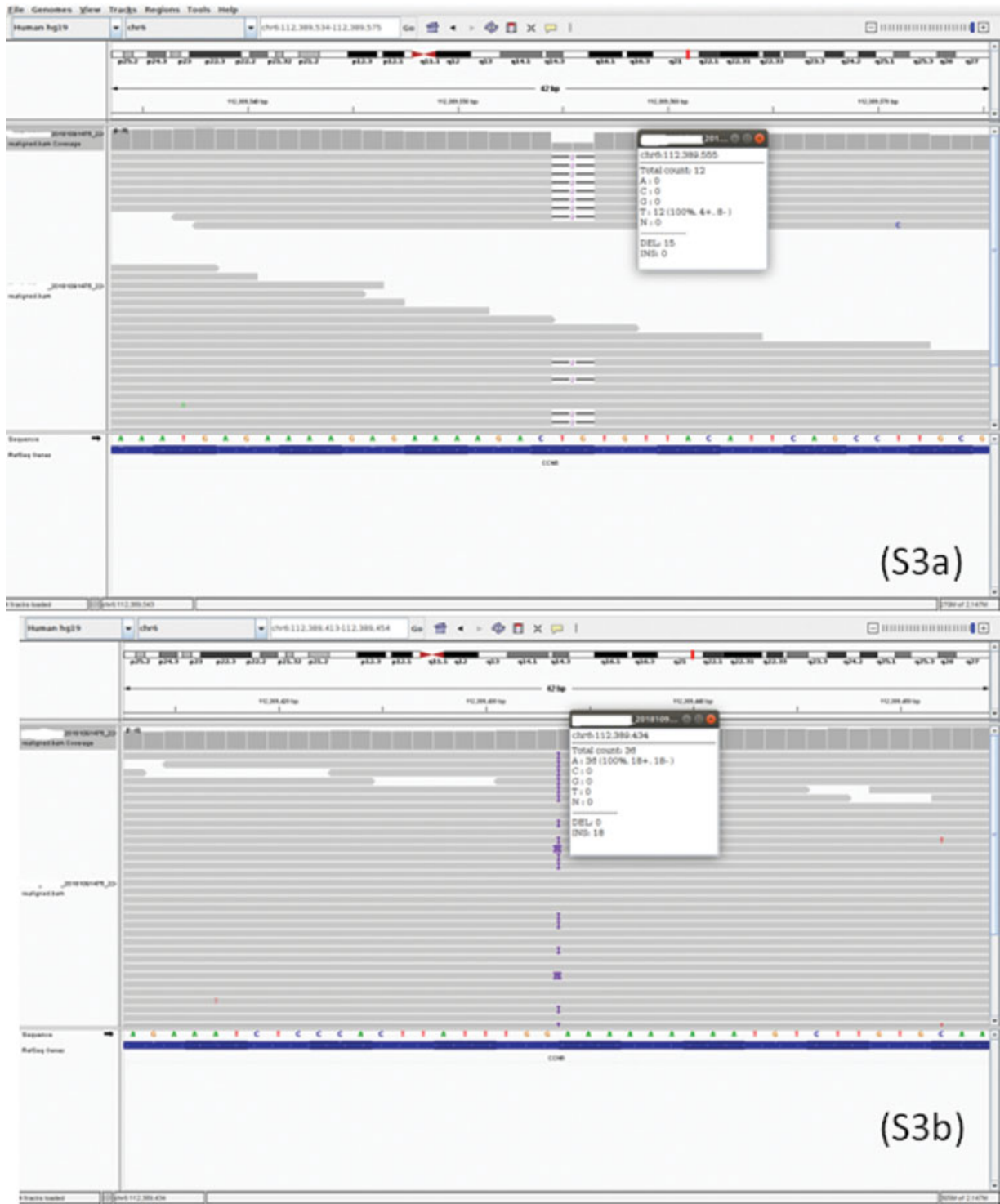


Supplementary Fig. S1 Sequence chromatogram (S1a) and alignment to the reference sequence, showing the variation in exon 4 of the WISP3 gene (*CCN6*; c.740_741del; p.Cys247Leufs*31) detected in homozygous condition in patient 1 along with the integrative genomics viewer image (S1b) for this mutation.



Supplementary Fig. S2 Sanger chromatogram images for pathogenic variants in patient 2; (S2a) for c.156C>A and (S2b) for c.740_741del.



Supplementary Fig. S3 Integrative genomics viewer images for pathogenic variants in patient 3; (S3a) for c.740_741del and (S3b) for c.624dup.