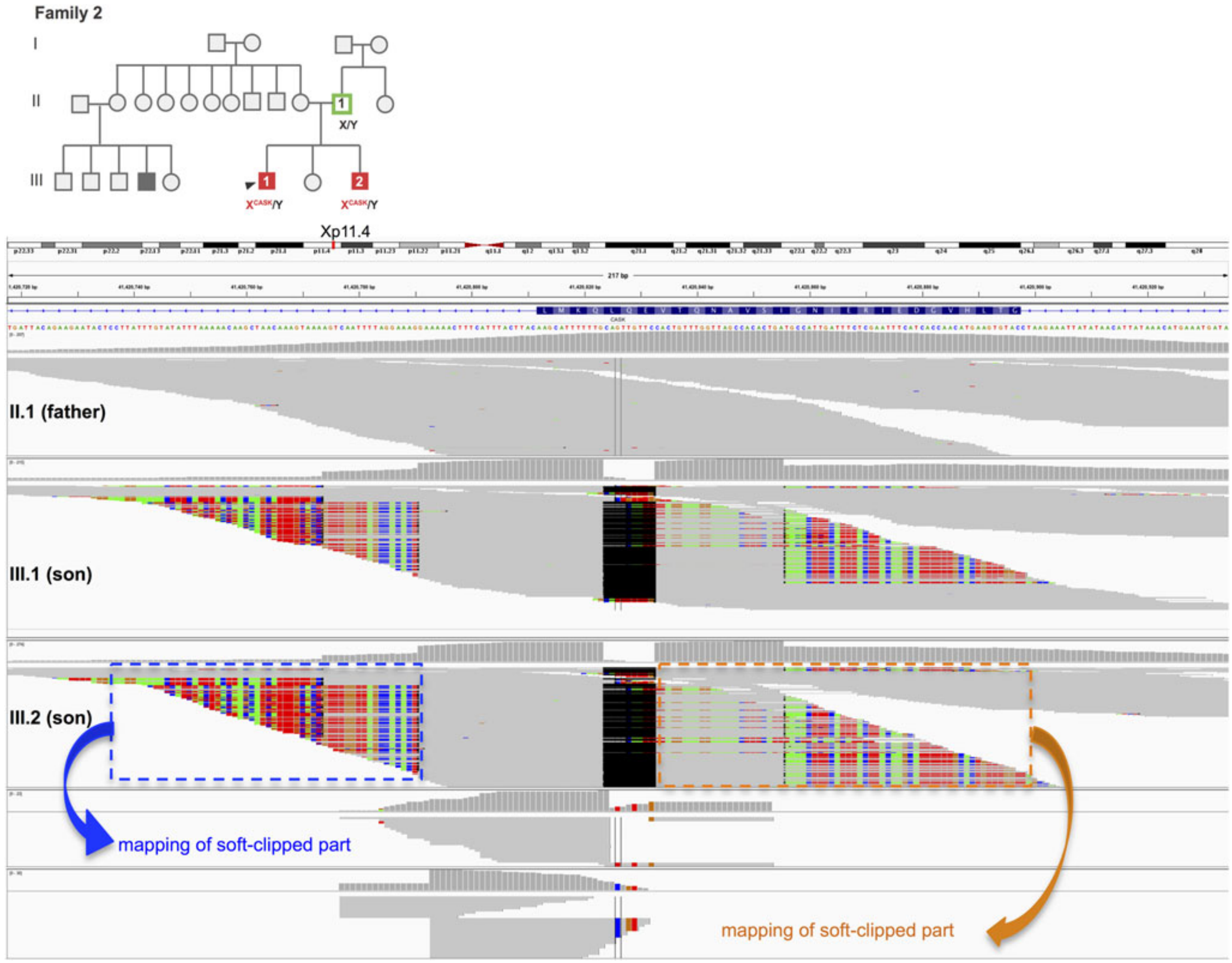
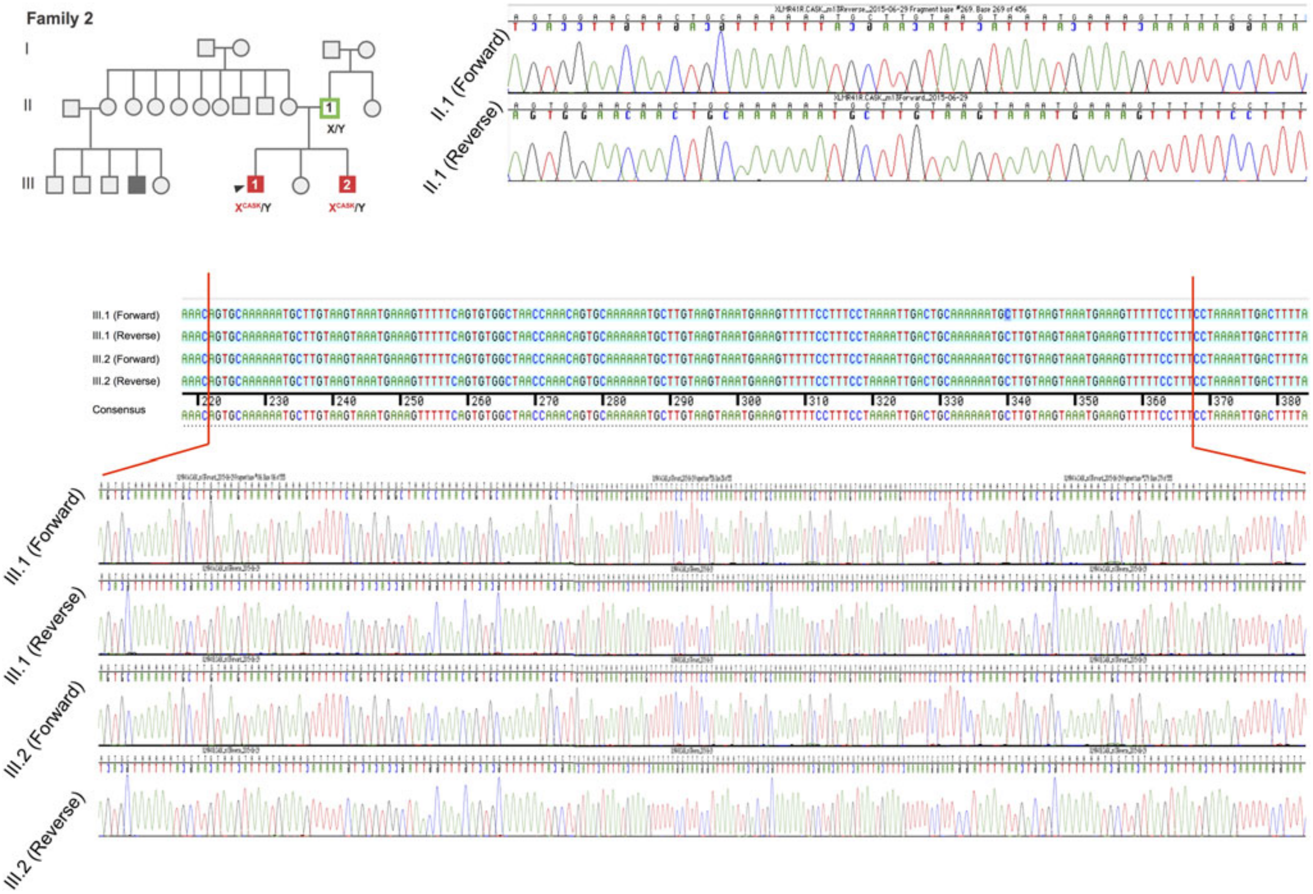


SUPPLEMENTARY FIG. S3. **(A)** IGV plots show read mapping of three individuals in family 2. Mapping of soft-clipped bases (shown in four colors *red* (T), *green* (A), *blue* (G), and *orange* (C) onto the reference genome hg19 at two *bottom panels* illustrates the evidence of short duplication. **(B)** Sanger sequencing chromatograms from three individuals in family 2 in the region containing a complex genomic alteration that resulted in two additional copies of a 34 bp sequence that spans exon–intron junction of CASK in the affected family members are shown. Both forward and reverse reads are shown for all the individuals.



SUPPLEMENTARY FIG. S3. (*Continued*).