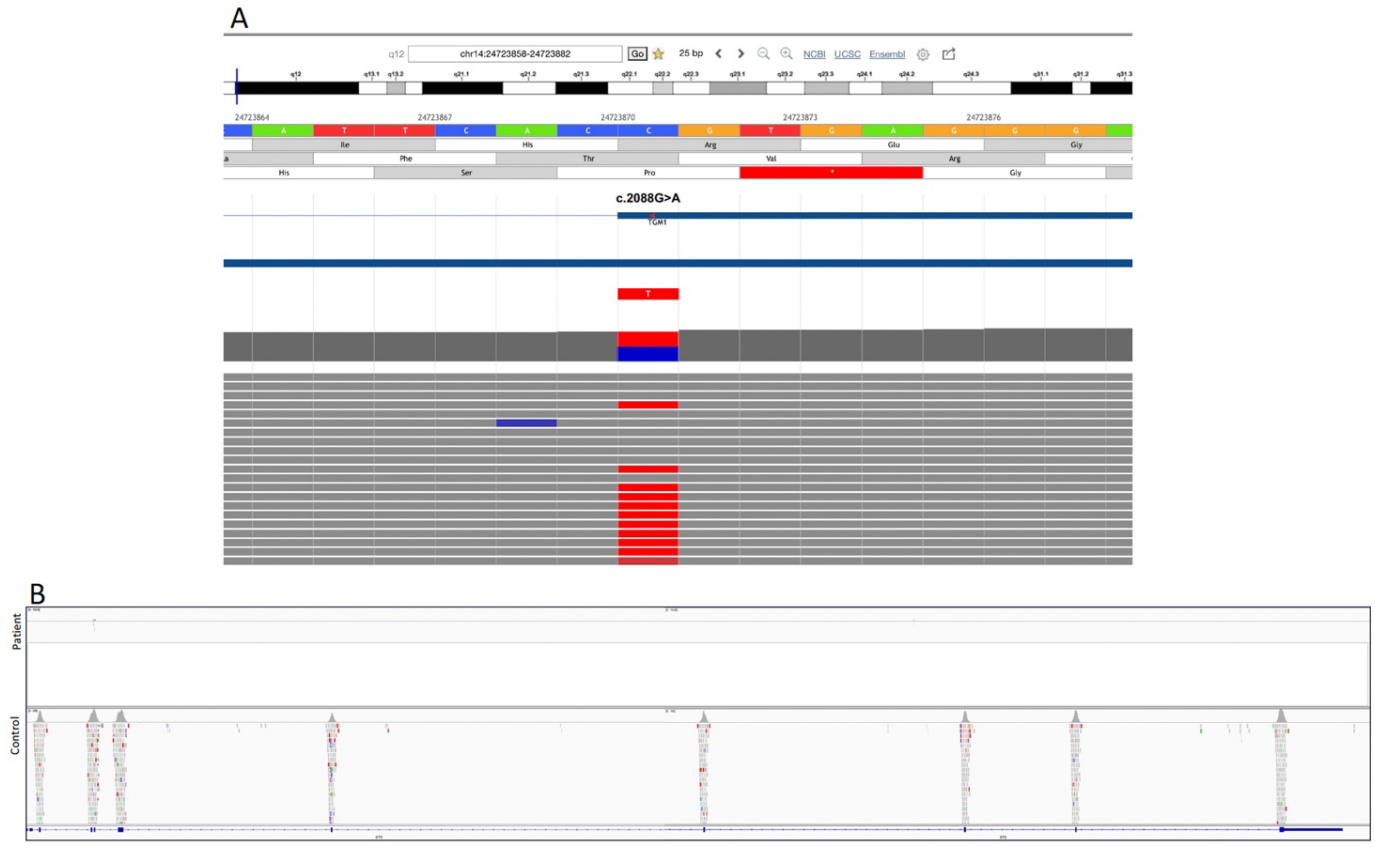

Figure S1

Integrative Genomics Viewer visualization of NGS data. (A) Position of the synonymous variant c.2088G>A p.(Thr696=) in the TGM1 exon 13 sequence; (B) Coverage of the 9 exons of STS gene (NM_ 000351.7) in a patient hemizygous for a whole gene deletion, compared with a normal male. In the patient, there are no visible reads for any exons of the STS gene.