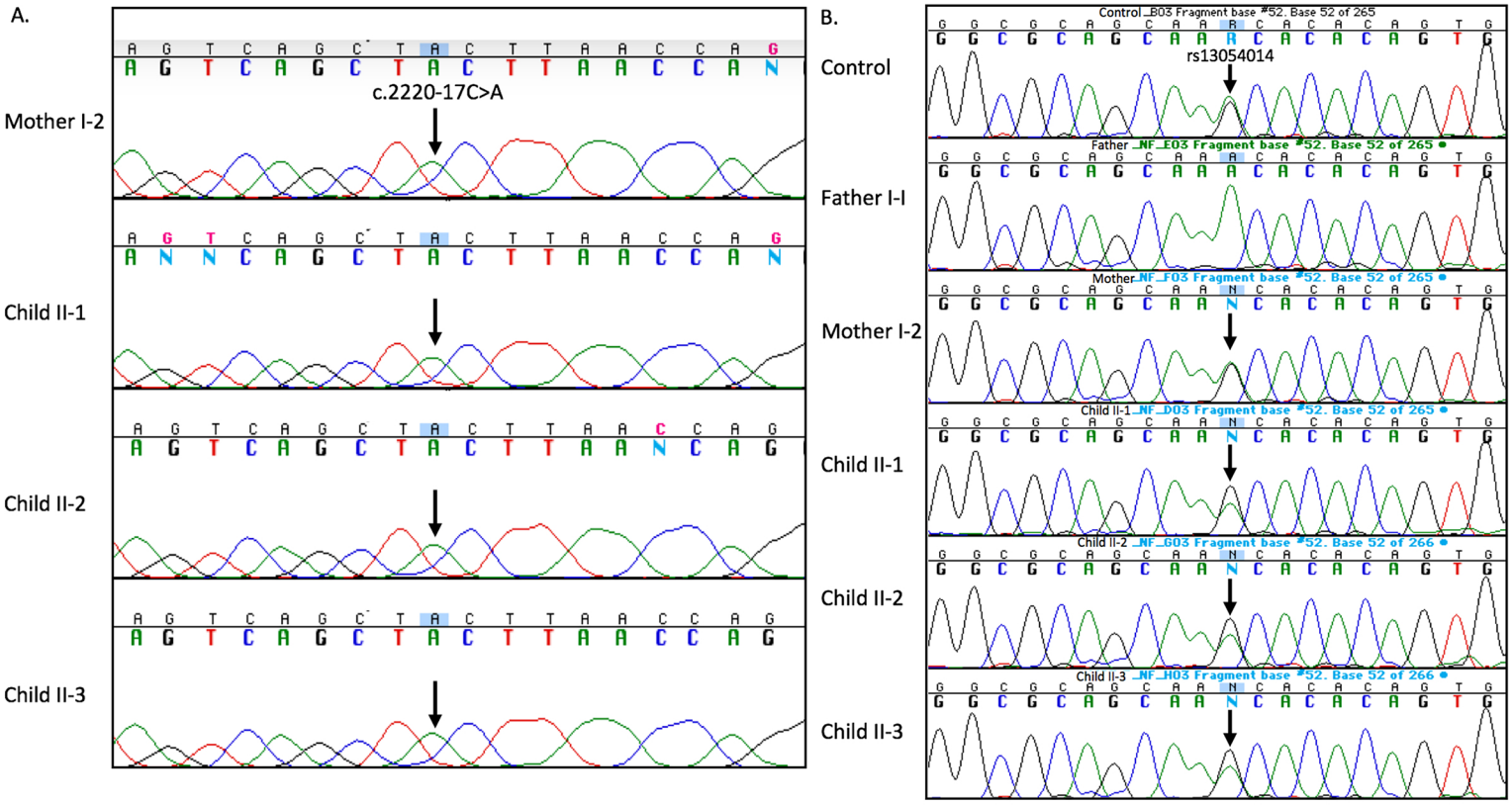
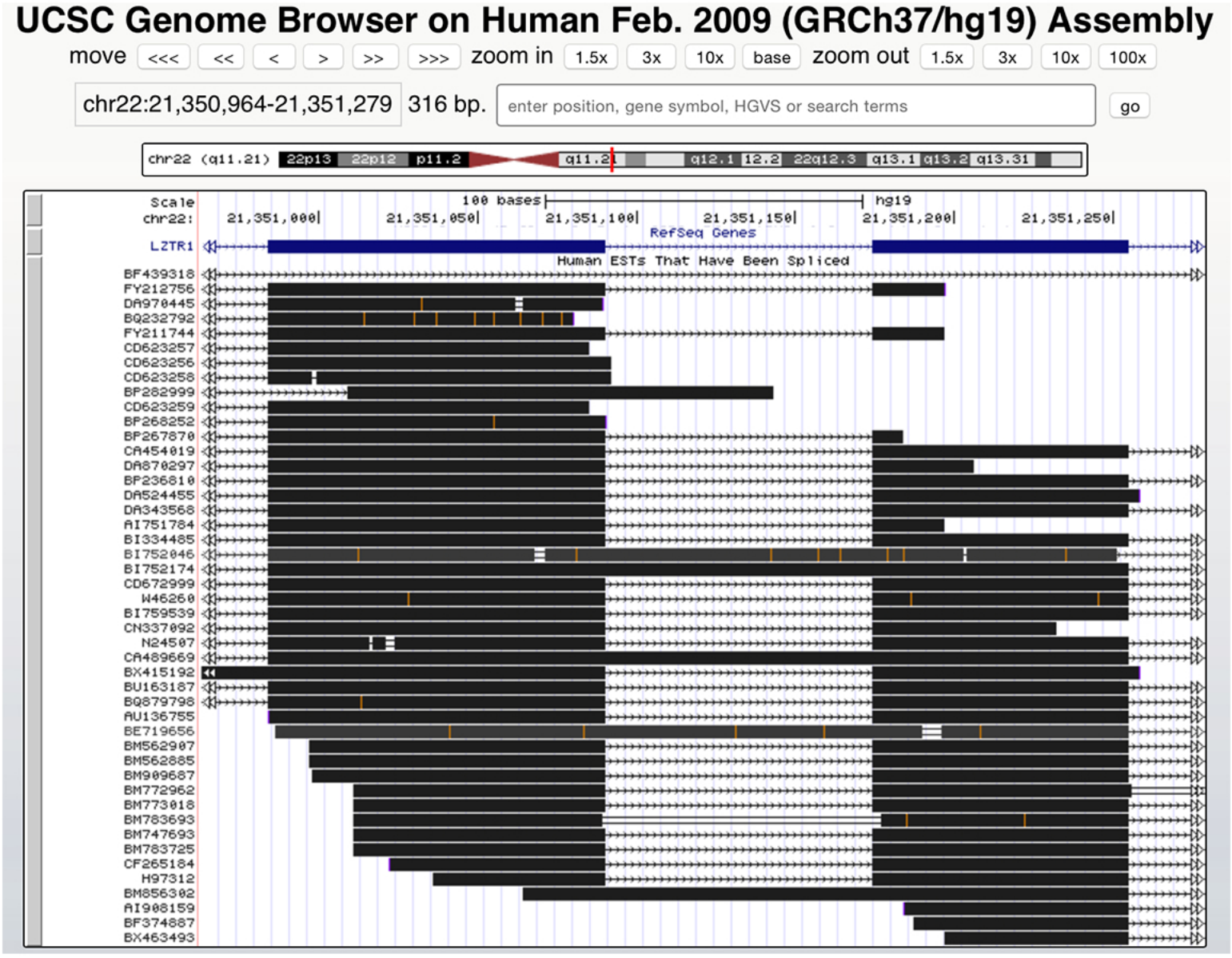
Supplemental Figure 1, Johnston et al.



Supplemental Online Figure 1. Sanger sequence analyses of aberrant splice products. A. Total lymphoblast RNA was reverse transcribed and PCR-amplified with primers from exons 18 and 21 of *LZTR1*. This section of sequence shows a 3' portion of intron 18 that includes the c.2220-17C>A variant, for which the mother is heterozygous and the children compound heterozygous. The analysis showed retention of intron 18 (583 bp) in the 885 bp RTPCR product (Figure 3A). The RTPCR sequence was exclusively from the allele with the splice variant showing that all of the cDNA with the aberrant splice product has only the A allele at the c.2220-17C>A position. B. Total lymphoblast RNA was reverse transcribed and PCR-amplified with primers from exons 1 and 8 of *LZTR1*. The product was sequenced and SNP (rs13054014) at position chr22:21,337,325 was used to assess the extent of nonsense mediated decay (NMD) in transcripts overall. In a control individual and the mother, without the nonsense allele, equal levels of the A and G peak are evident. In the children the A allele in *cis* with the nonsense variant is reduced in comparison to the G allele, demonstrating a relative reduction in cDNA from the nonsense allele, confirming NMD.

Supplemental Figure 2, Johnston etal



Screen shot of UCSC browser showing exons 19 and 20 with a natrually-occuring, but minor isoform supported by 5 spliced EST sequences. This isoform is responsible for the 386 bp band in the RTPCR result shown in figure 1A.