Supplementary Fig.1 Konno *et al.*

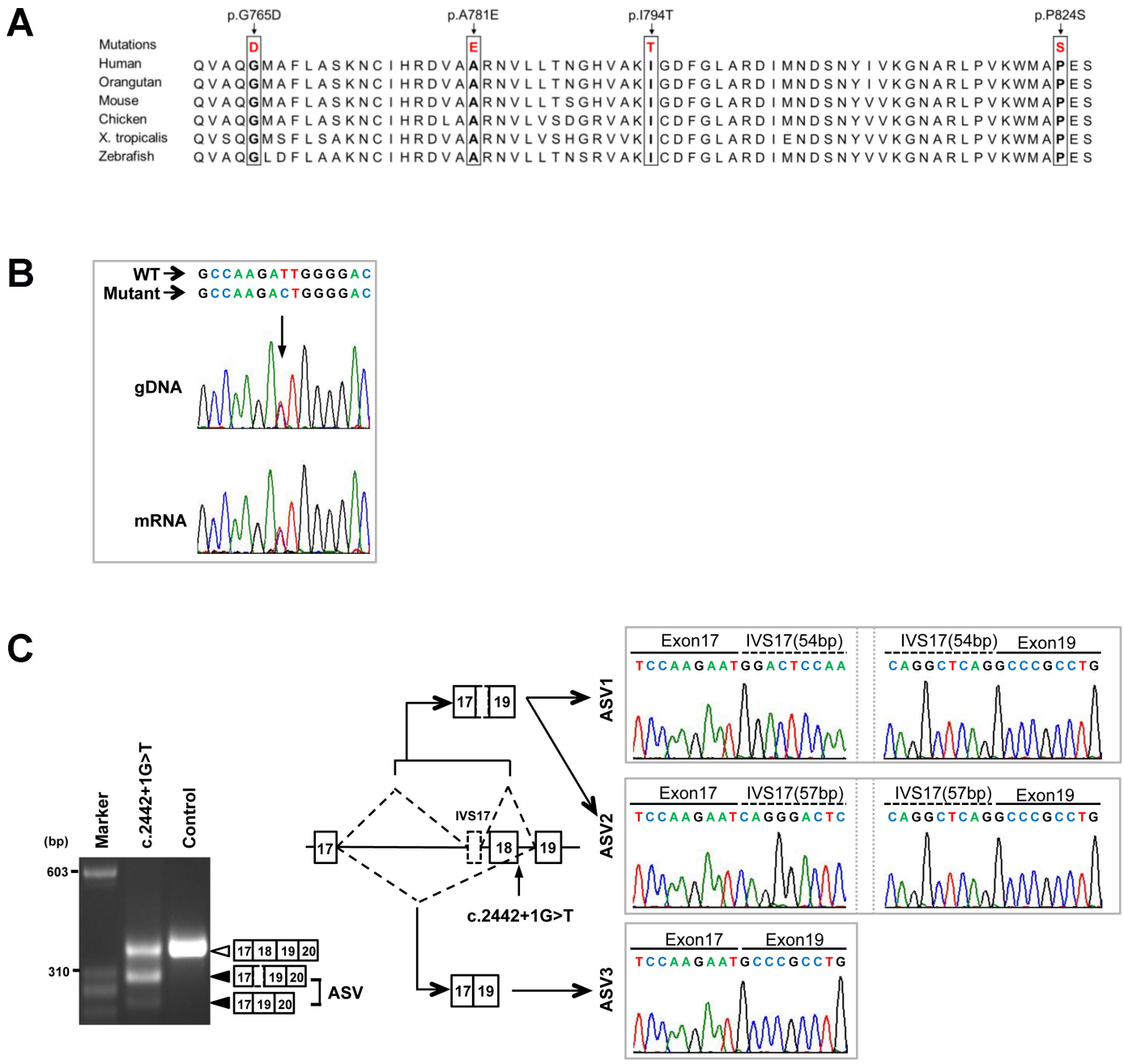

Figure e-1. Identification of CSF-1R mutations

(A) The evolutionarily conserved of amino acids associated with four missense mutations in our study are shown. The mutated amino acids are in red.

(B) Electropherogram of the sequencing of amplified genomic DNA and RT-PCR products of patient with missense mutation (p.I794T). The mutant allele with the missense mutation is expressed at a level comparable to the normal allele.

(C) Electrophoresis of RT-PCR amplicons spanning from exons 17 to 20 of patient with splice-site mutation (c.2442+1G>T) showing aberrant splice variants (closed triangles) in addition to normal transcript (opened triangle). We identified three aberrant splice variants (ASVs 1-3) by subcloning and sequencing. Exon18 was skipped in all of the aberrant variants and a portion of the intervening sequence (IVS)17 sequence was included in two variants (ASVs 1 and 2).