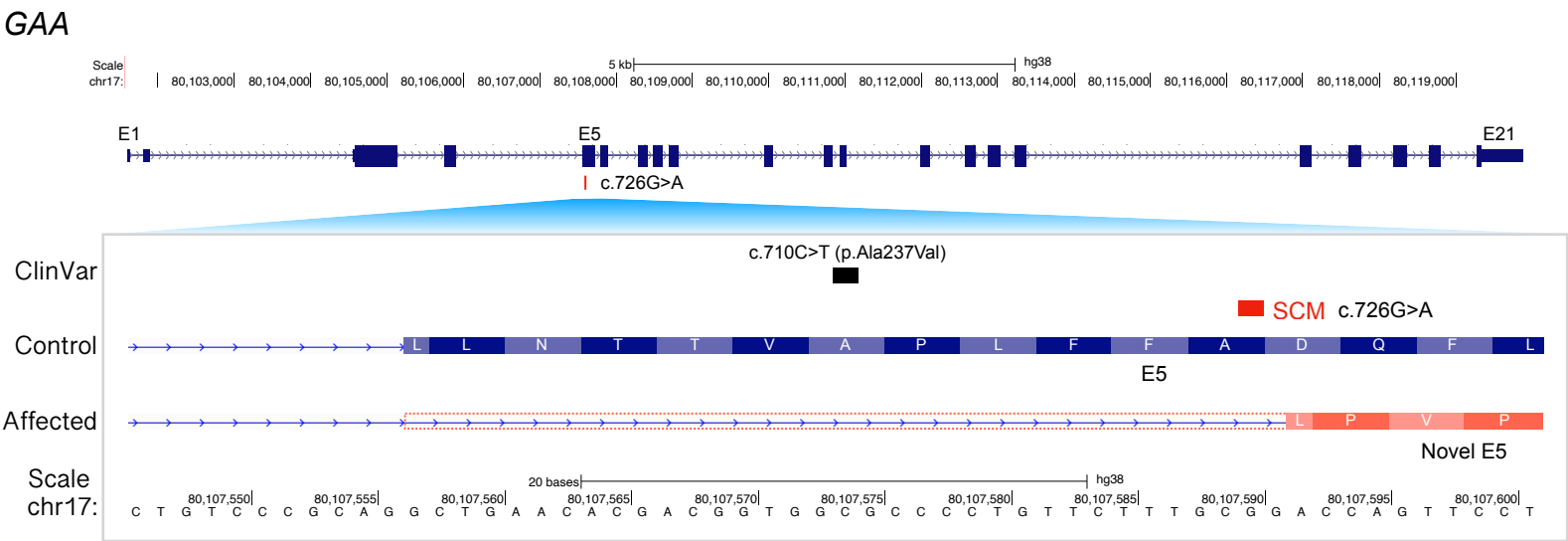




**Supplementary Fig. 1 Schematics of exon shrinkage by SCM found in GAA.**

The upper panel shows the gene structure of *GAA* obtained from the gene annotation data of GENCODE v29. The lower panel is a close-up view of the SCM and exon 5 (E5). The ClinVar track shows a known pathogenic variant, c.710C>T, for Pompe disease. The gene structure for a control individual is followed by that for an affected individual with the SCM (c.726G>A) which induces an exon shrinkage. The dotted box indicates the region of the shrinkage. The shrunken region changes the downstream reading frame.

SUPPLEMENTARY FIGURE 1.