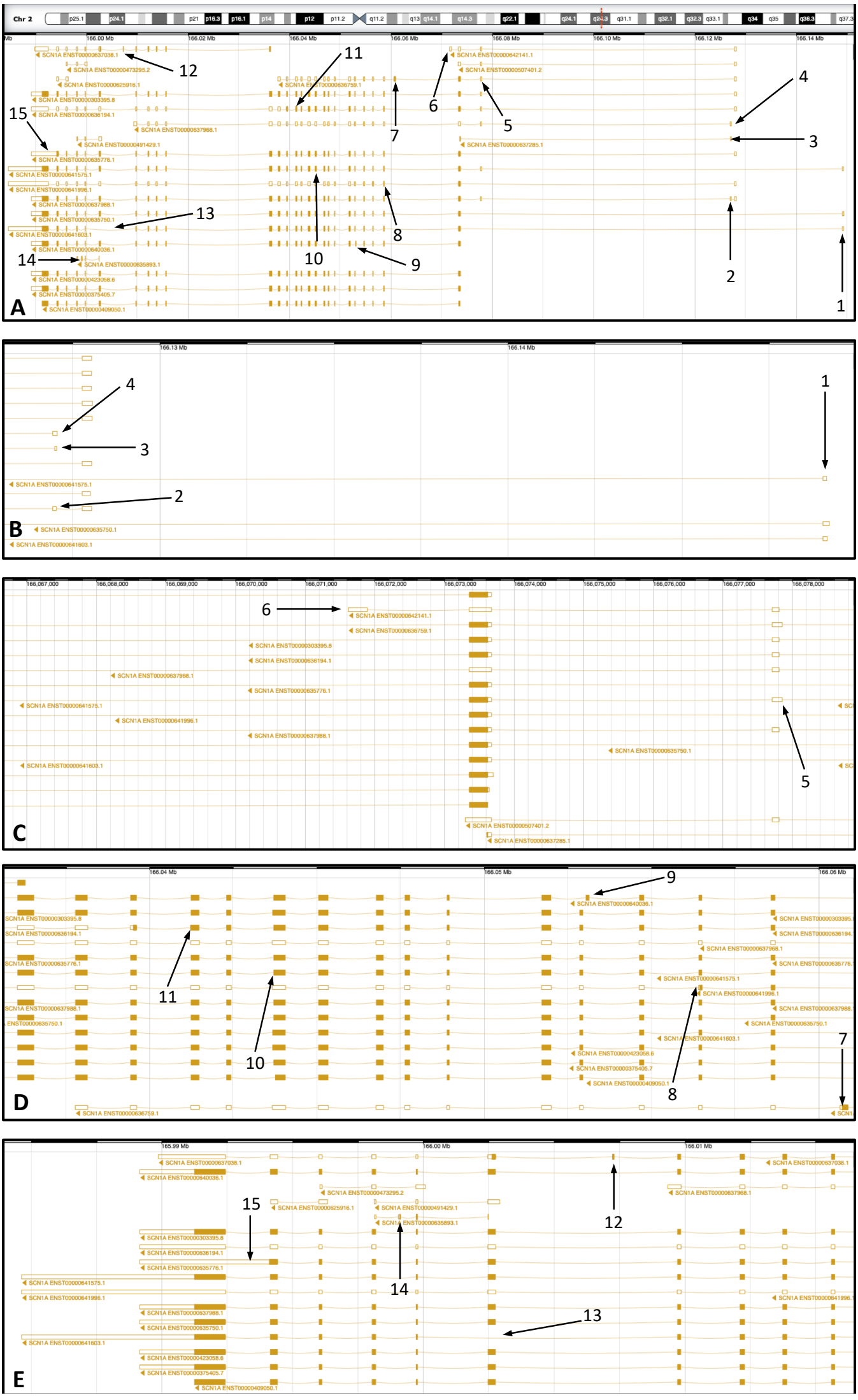
Supplementary Data 1

The first sheet lists the 23 variants found in the ClinVar database that are incorporated into the exon annotations produced in this study. Additional variants found within 8bp of existing splice sites were filtered out and are presented in sheet 2. Note that ClinVar is primarily based on RefSeq annotation; most of the variants in the table are still considered as intergenic or intragenic by ClinVar and the classifications of pathogenicity presented there will not necessarily change according to these annotations. Genome coordinates are for GRCh38.

Supplementary Figure 1

Figure showing the position of all features identified within *SCN1A* as viewed in the Genoverse browser <https://wtsi-web.github.io/Genoverse/>). The 5' end is to the right and the 3' end is to the left. See Table 1 in the main manuscript for a description of each of the 15 identified features. Panel A below shows the entirety of *SCN1A* and the 15 features, which are illustrated with arrows. Panels B-E show a higher resolution of these features starting at the 5' UTR (panel B) and ending with the 3' UTR (panel E).


A
B
C
D
E

Supplementary Data 2

The complete list of Ensembl gene IDs and HGNC gene names (https://www.genenames.org/) that were reannotated as part of the study. Also listed is the number of number of transcripts found before reannotation (GENCODE v20 transcripts) and after reannotation (GENCODE v28 transcripts). The Mendelian Inheritance in Man accession ID for each gene is listed in the final column (https://www.omim.org/)

Supplementary Data 3

Results from the fitDNM package analysis across the 450bp of CDS from exons 7,9,12 and 14 in *SCN1A*. Refer to Table 1 in the main manuscript.