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Supplemental information

A recurrent *de novo* splice site variant involving *DNM1* exon 10a causes developmental and epileptic encephalopathy through a dominant-negative mechanism

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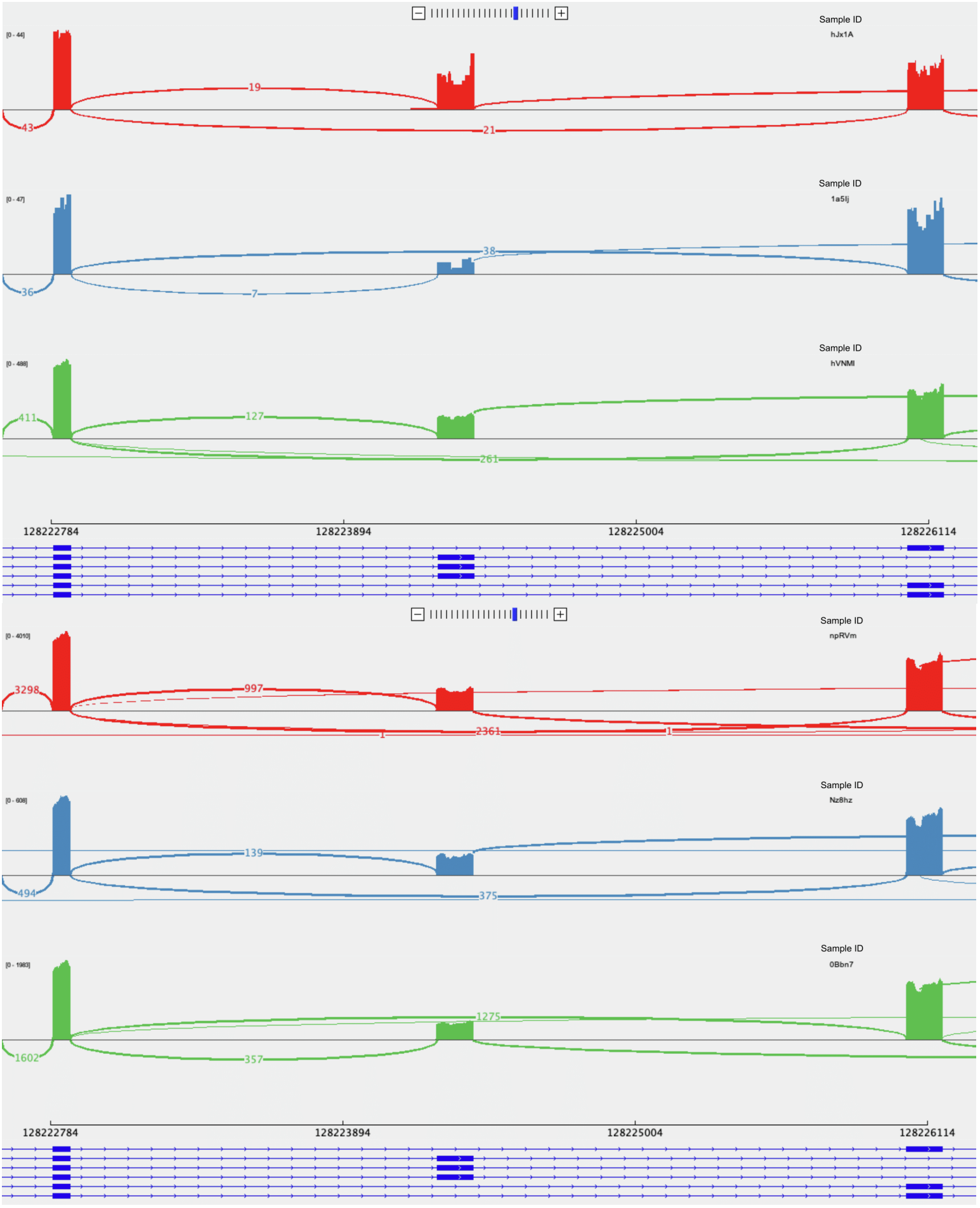
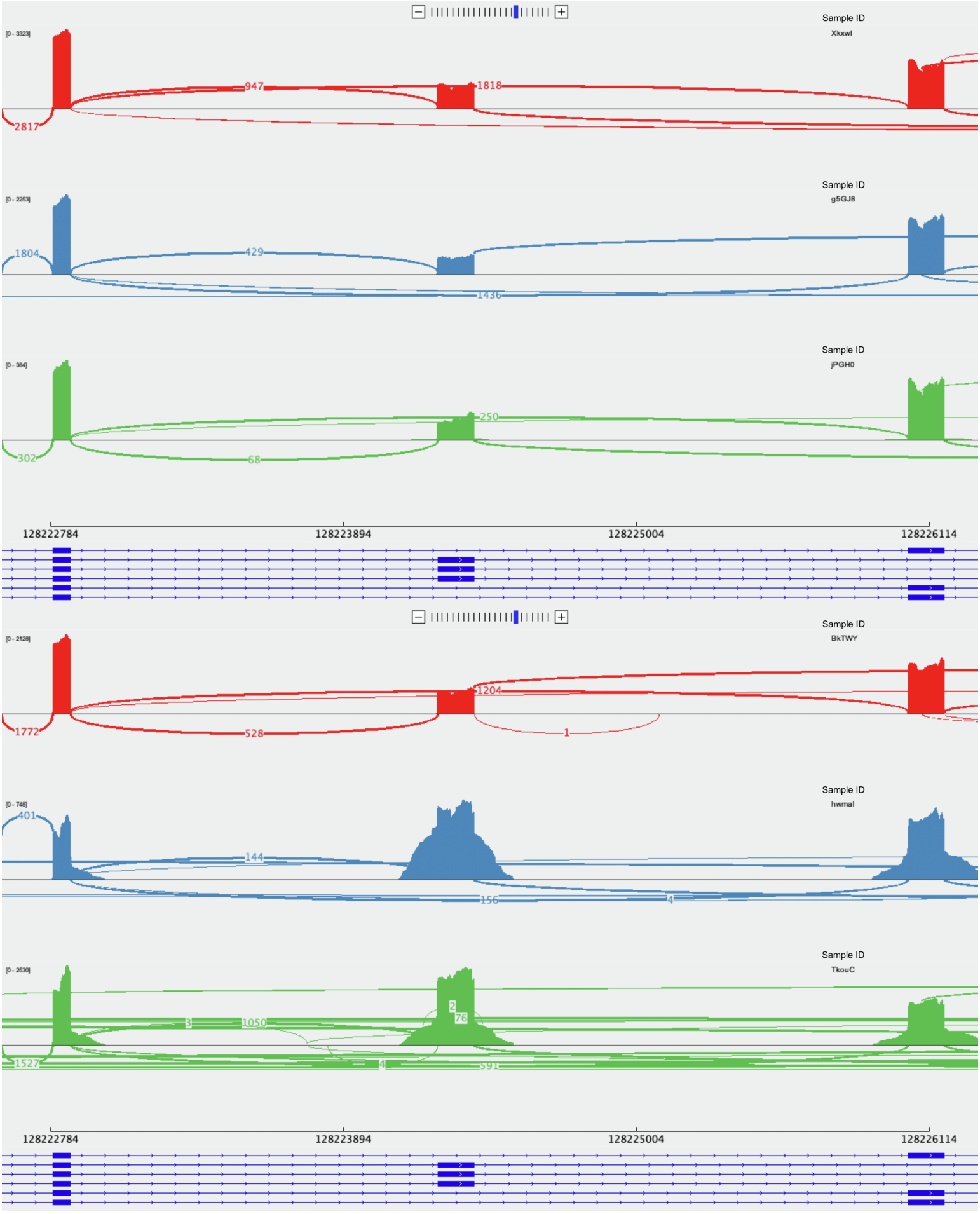
Figure S1. Sashimi plots from bulk tissue RNAseq in 39 individuals. *DNM1* exons 9, 10b, and 10a are displayed with corresponding splice junctions. Raw read quantity of the exon 9-10a junction consistently exceed that of exon 9-10b.

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Sample ID
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Sample ID
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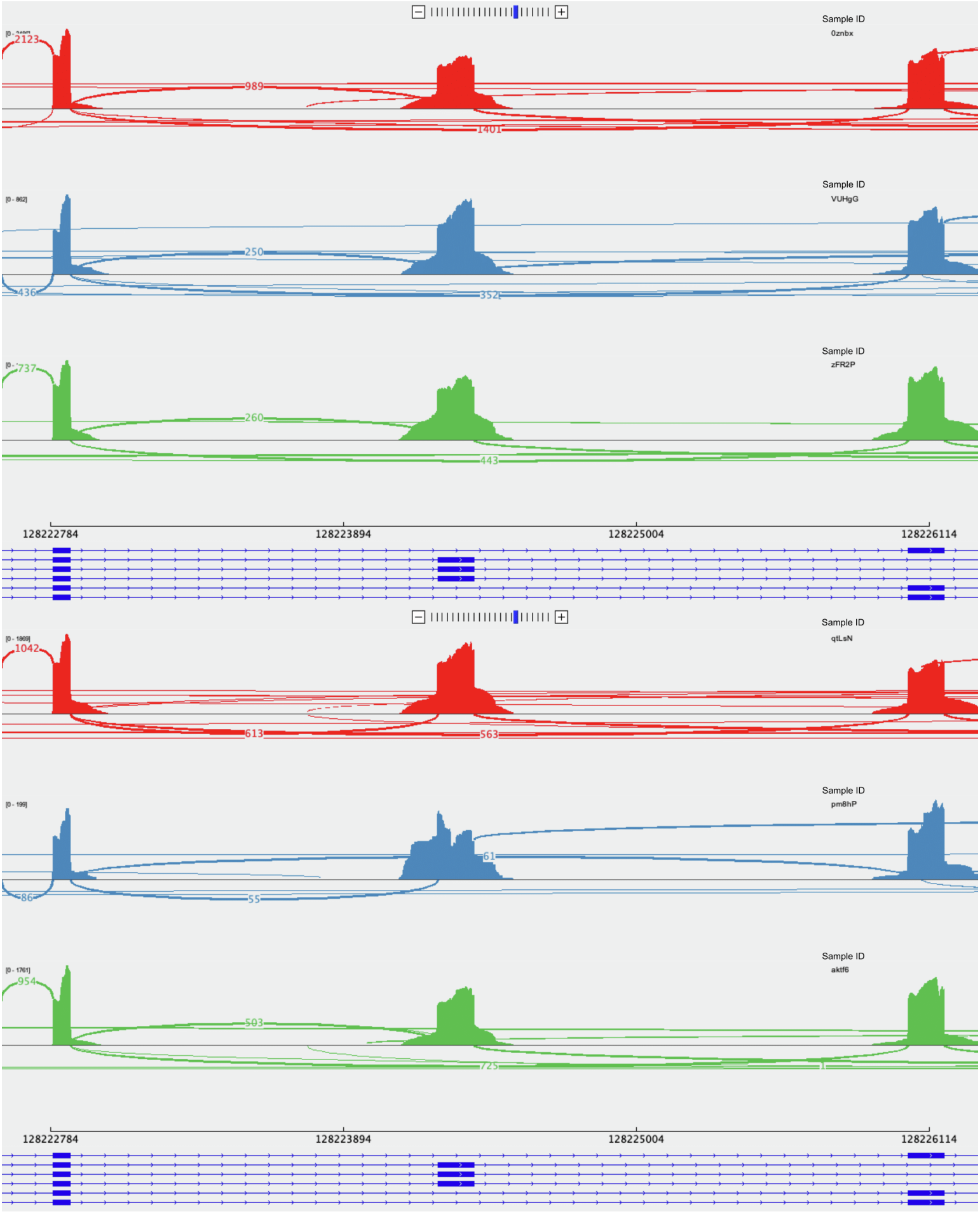
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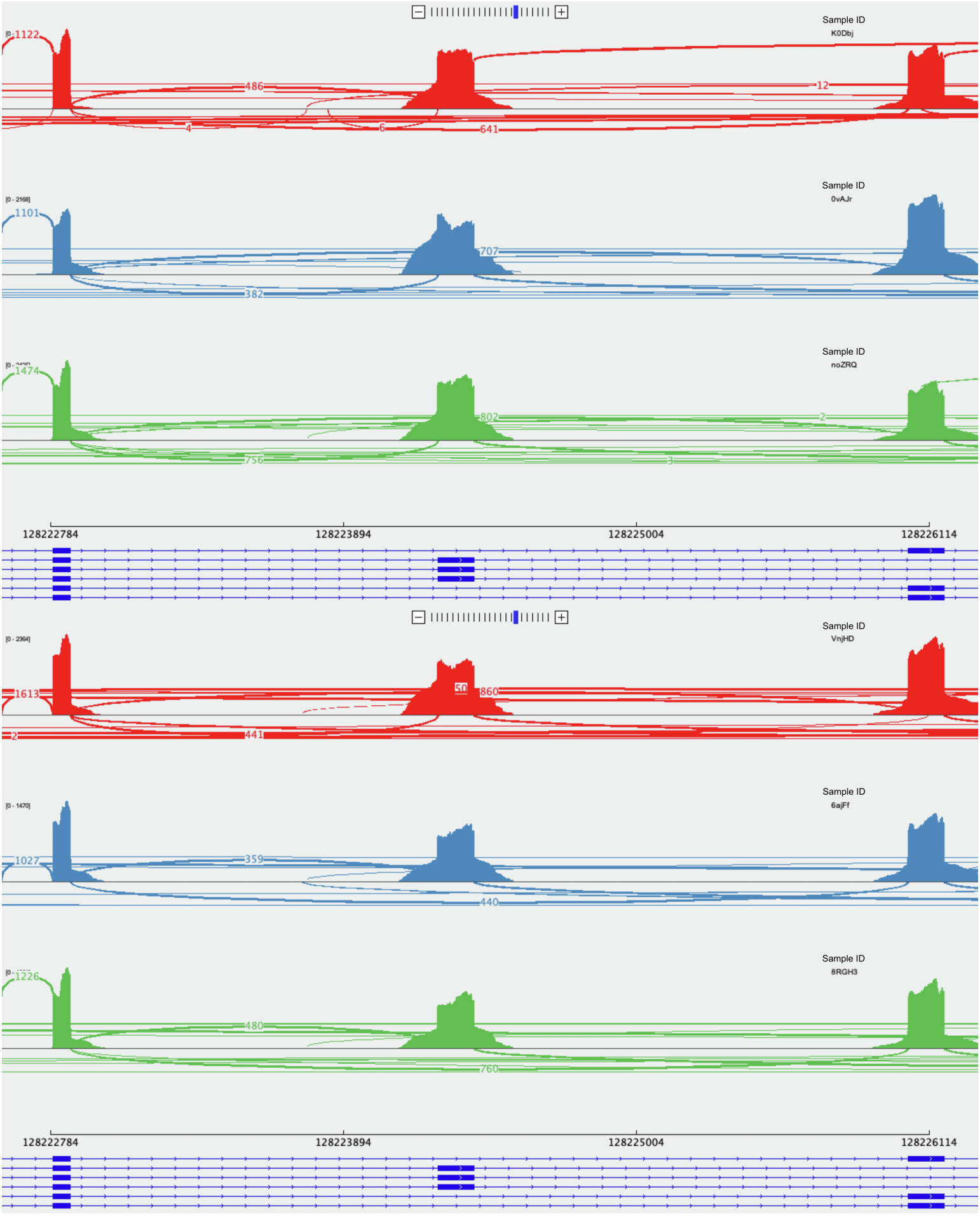
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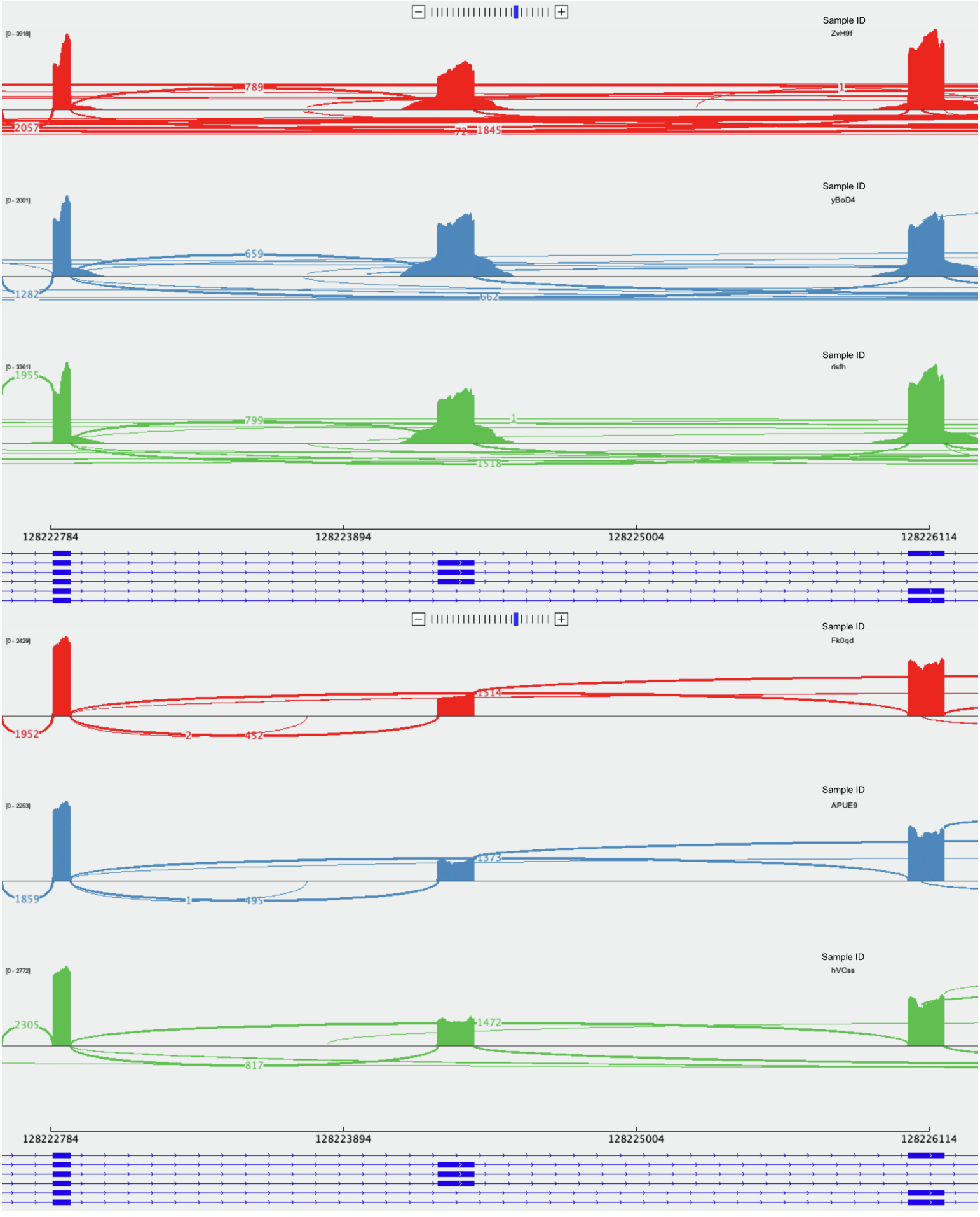
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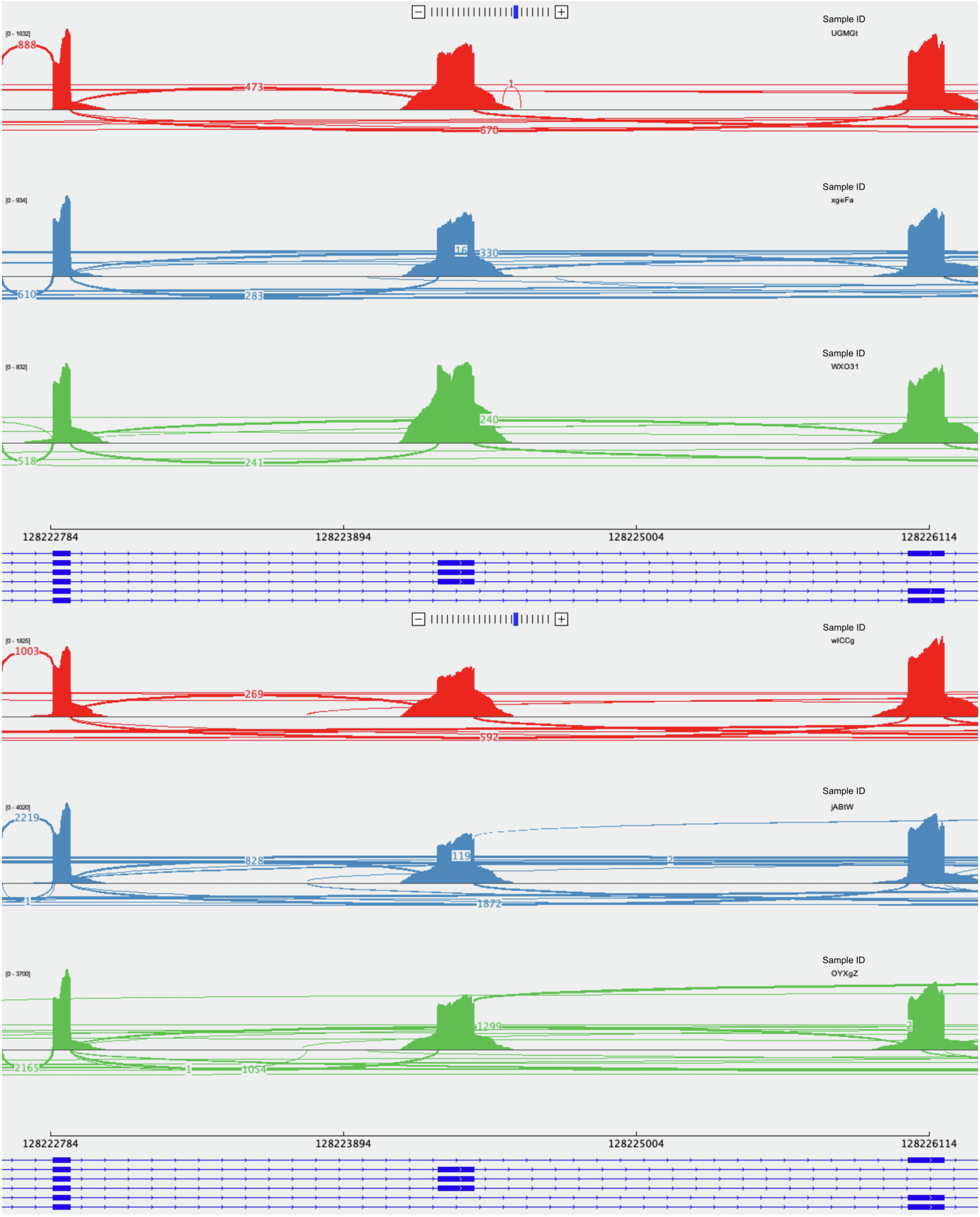
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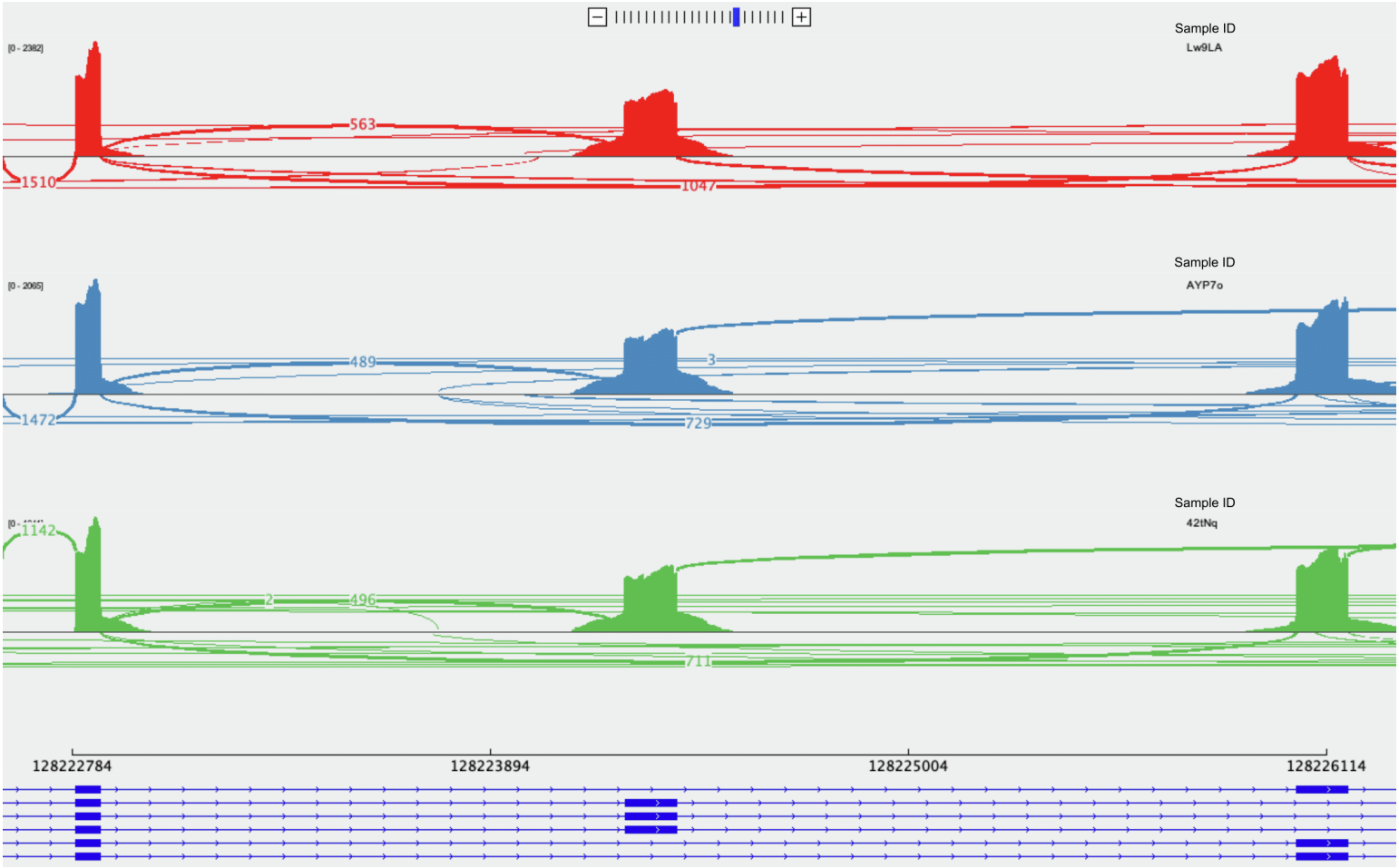
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711
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128225004
128226114

Figure S2. Analysis of expression data for *DNM1* transcripts containing exon 10a (blue) and 10b (green) in 255 adult samples from the GTEx Portal. (A) Distribution of expression quantity, in transcripts per million (TPM), of *DNM1a* and *DNM1b*. *DNM1a* expression is a mean of 5.7 times higher than that of *DNM1b*. (B) Per-sample transcript quantities for *DNM1a* and *DNM1b*, showing the increased expression of *DNM1a* in the GTEx data.

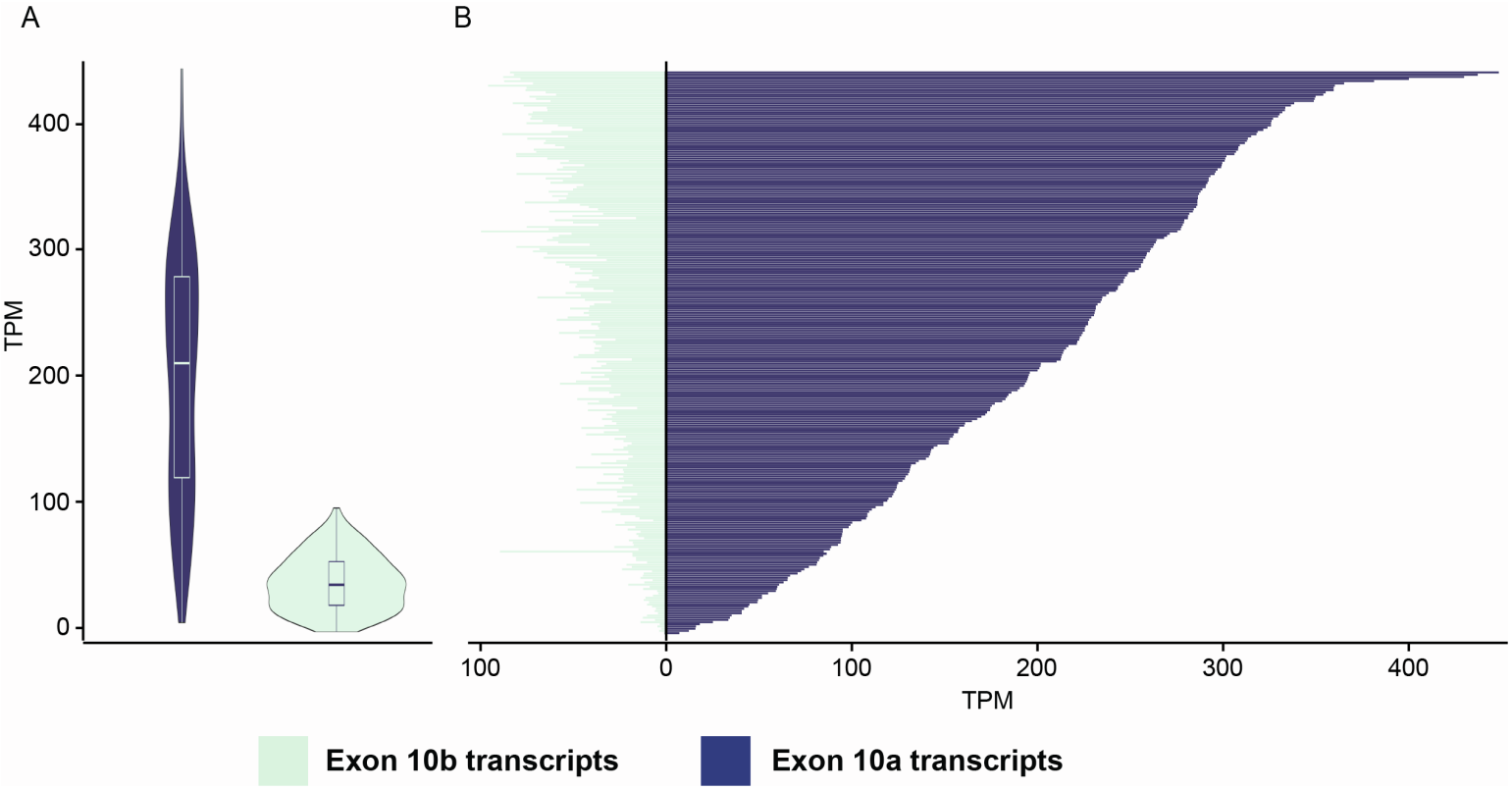

Figure S3. Analysis of missense variation in *DNM1* isoforms *DNM1a* (blue) and *DNM1b* (green) in the gnomAD population data. (A-B) The transcript-specific observed vs expected missense variation (A) and missense Z-scores (B) do not differ between *DNM1a* and *DNM1b*. (C-D) Minor allele frequency for missense variants in the population are similarly distributed across both isoforms (C) although exon 10-specific population missense variation (D) is higher in exon 10b than 10a.