**Table 1. Metrics obtained from the analysis of GTEx v7 datasets to observe the impact of variants prioritized as splice impacting.** Our analysis identified 4 variants in autosomal recessive genes that were present in a carrier state in individuals in GTEx v7 and had observable impacts on splicing in these individuals. Metrics were calculated from aligned RNAseq datasets from tissues with a transcript per million value > 5 for the gene of interest. *Cases,* individuals within the GTEx dataset carrying prioritized variant. *Controls*, a group of 10 randomly selected individuals within the GTEx dataset that do not carry the prioritized variant.

| **Variant** | **Gene** | **Tissue** | **Metric Type** | **Controls** Mean (95% CI) | **Cases** |
|---|---|---|---|---|---|
| 20-3899342-G-A | *PANK2* | Fibroblasts | Intron Retention | 0.12 (0.10-0.14) | 0.32 |
| 12-88448136-G-A | *CEP290* | Thyroid | NRC | 0.15 (0.02-0.27) | 0.91 |
| 10-73567463-C-T | *CDH23* | Ovary | NRC | 0.003 (0-0.01) | 0.11 |
| 2-110922263-G-A | *NPHP1* | Testis | NRC* | 0.51 (0.48-0.55) | 0.7 |

*NRC* = normalized read count (described in Supporting Information). *switch in usage of two canonical exon junctions in alternative isoforms.