

Headline: Found Variants in Splice Regions which could be Pathogenic

Motivation

- Brand new tool just made by Illumina & Stanford: SpliceAI
- John's Doctor said best theory: lipid metabolism and/or immune involvement

What we did

- Ran Splice AI on GPU on all of John's Lipid Metabolism Genes (1,000)

Found

- 18 genes flagged as potentially: 'pathogenic'
 - Lipid Metabolism: ACSM5, CES5A, PIGN, CES5A, ACP6, PDXDC1, ACSM5, OSBPL10, PIK3R3, SAA1, PEX5, ACSM1, RBL2, RETSAT, PI4KA, AGMO
 - Immunity: ACP6, CD55, CHUK, SPTA1

Next Steps

- Share these genes with Doctor to inspect