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1. tablo içeren bir resim

   Açıklama otomatik olarak oluşturuldutablo içeren bir resim

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   Açıklama otomatik olarak oluşturulduI have applied filtre. Rules of filtersare popfreq<=0.0017 for homozygous mutation, <0.0006 for comphet mutation, <0.0002 for heterozygous mutation. In case i can’t find the gene, the thresholds are changable.
2. tablo içeren bir resim

   Açıklama otomatik olarak oluşturuldutablo içeren bir resim

   Açıklama otomatik olarak oluşturuldumetin içeren bir resim

   Açıklama otomatik olarak oluşturulduAfter popfreq limitations, i first looked at exonic genes since intronic genes are useless part of the genome and splicing variants are less likely to be the pathogenic variant. And filtered them according to their CADD\_phred values. As far as i learned, if CADD phred value is more than 30, it is pathogenic. After that filtre i lessened the possibilities into 3. NBAS, SEC14L3,SELO genes. All homozygous mutations and all but 1 (NBAS) heterozygous mutations are eliminated at that point.
3. metin içeren bir resim

   Açıklama otomatik olarak oluşturuldumetin içeren bir resim

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   Açıklama otomatik olarak oluşturulduThen i searched all possibilities in the databases like OMIM and GENECARD and compared the disorder of these genes’ mutations with our patients symptomps.

When i searched for SELO gene, both OMIM and GENECARD direct me into SELENOO gene, the location of the gene is same with what is said in the excel file, so i assumed that it is the same gene. SELENOO gene is related with different bacteria infections, doesn’t match with our patient’s symptoms.

For SEC14L3 gene, we can say that it is not the pathogenic variant either. Because bardet biedl syndrome is related with central nervous system related endocrine systme disorders. doesn’t fit into our patient profile.

Last gene almost perfectly shows us what is the problem we have disorders of short stature, optic nerve atrophy, pelger huet anomaly( which is a immune system disorder, can be related with the low immunoglobin counts in the patients history.), facial dysmorphism, short feet and hands both in the description of gene and patient history. In genecard it says gene mutation related with liver disorders but our patient has normal liver biopsy. We can assume that elevation could be caused by mutation and there can be another factors that prevent liver failure since we have a really good match.

metin içeren bir resim

Açıklama otomatik olarak oluşturulduWe can also see in the excel table that exonic function column says stopgain. So it is not a synonymous aka harmless mutation.

ANSWER IS **NBAS** GENE.