THE FIRST CLINICAL CASE OF RARE FORM OF FOCAL EPILEPSY CAUSED BY THE NOVEL MUTATION IN THE NPRL3 GENE IN RUSSIAN FEDERATION AND KAZAKHSTAN


The nucleotide variant c.481C>T which leads to stop codon p.Q161* in heterozygous state was revealed in exon 5 of the NPRL3 gene. This variant was not described in gnomAD and HGMD databases and was considered as pathogenic according to ACMG criteria. An interesting fact is that the most frequent pathogenic variants in the NPRL3 gene (among the 21 described variants in the HGMD) are nonsense mutations and frameshift deletions.

Conclusion This paper describes the first clinical case of rare form of focal epilepsy caused by the novel mutation in the NPRL3 gene in Russian Federation and Kazakhstan.

91 FABRY’S DISEASE WITH MINIMAL MANIFESTATIONS IN GIRLS (CLINICAL CASES)

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92 TYPICAL PROBLEMS OF PARENTS OF CHILDREN WITH CLEFT LIP AND PALATE

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