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Familial hemiplegic migraine with prolonged coma and hyperthermia: ATP1A2 gene mutation case report in a single Saudi family

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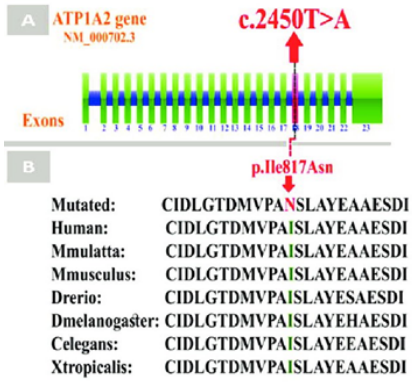
Familial Hemiplegic Migraine (FHM) is a rare disorder presented commonly with coma, hyperthermia and headache. FHM is usually associated with fully reversible motor weakness as a specific symptom of aura. Seizure and fever are the secondary features observed. Three sisters diagnosed with type 2 FHMs presenting features, such as coma and hyperthermia. The brain (magnetic resonance imaging) revealed focal subtle cortical swelling, Electroencephalography showed unilateral slowing, while no signs of infectious disease were observed. Molecular and genetic tests using whole exome sequencing identified a novel heterozygous mutation (c.2450T > A p.Ile817Asn) in the exon 18 of the ATP1A2 gene (NM_000702.3). The Sanger's sequencing results confirmed the variant was segregated with the disease phenotype within the family. The current study report for the first time, a Saudi family with migraine coma having a novel heterozygous ATP1A2 mutation.



Recent Publications

1. Safiyyah Asiri, W. A. (2019). Prevalence and outcomes of Guillain-Barré syndrome among pediatrics in Saudi Arabia: a 10-year retrospective study. Neuropsychiatr Dis Treat., 15: 627-635.

2. Waleed Altwaijri, F. A.-R. (2019). Familial Hemiplegic Migraine with Prolonged Coma and Hyperthermia: ATP1A2 Gene Mutation Case Report in a Single Saudi Family. JBCGenetics., 2(1): 85-90.