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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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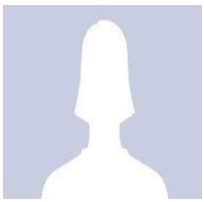

Abstract

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.

Biography:

Safiyyah Asiri has completed her medical school at the age of 24 years from King Khalid University, KSA. She worked as a resident in King Abdulaziz Medical City, for 5 years and was awarded as the best resident among her level and the resident of the year for three consecutive years. She has 2 publications and ongoing two projects. Currently she is working as a senior registrar at King Abdullah Specialized Children Hospital, Riyad.

Speaker Publications:

1. Safiyyah Asiri, W. A. (2019). "Prevalence and outcomes of Guillain-Barré syndrome among pediatrics in Saudi Arabia: a 10-year retrospective study". Neuropsychiatric 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.

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