37th European Neurology Congress

May 02-03, 2023

Webinar

Natural history of HSP56 informed by a common ancient CYP2U1 founder mutation

Raneem H. Alghamdi[1]*, Tariq Masoodi[1], Mohammed Alowain[1], Hesham M. Aldhalaan[1], Fathiya Al Murshedi[2], Moeenaldeen Alsayed[1], Mohammad Al-Muhaizea[1], Sateesh Maddirevula[1], Niema A. Ibrahim[1], Firdous M. Abdulwahab[1], Mais O. Hashem[1] and **Fowzan S Alkuraya[1,3]**

[1]King Faisal Specialist Hospital and Research Center, Saudi Arabia

[2]Sultan Qaboos University Hospital, Oman

[3]Alfaisal University, Saudi Arabia

Biallelic mutations of CYP2U1 are known to cause a complicated form of hereditary Spastic Paraplegia (SPG56). Only 26 families with SPG56 have been reported to date with 29 variants in CYP2U1. Here, we report 18 additional families (25 patients) all but two are homozygous for a founder Arab variant c.947A>T:p.Asp316Val. This largest cohort to date on SPG56 demonstrates a remarkably consistent phenotypic expression of the founder variant with predictable and universal loss of ambulation but variable degree of loss of speech, intellectual disability and white matter changes. Two families had novel variants in CYP2U1 (c.890T>C:p.Phe297Ser and c.967 997del) and the phenotype was compatible with that seen in the rest of the cohort. Haplotype analysis allowed us to estimate the age of the founder mutation at 1040 years, which makes this as one of the most ancient intellectual disability mutations in Arabia and explains its high carrier frequency and wide geographic distribution.

Biography

Raneem Alghamdi is a Pediatric Neurology Resident currently working at King Faisal Specialist Hospital & Research Center, Riyadh, Saudi Arabia. He is the current Deputy Chief Resident and the Quality Chief of Pediatric Neurology Trainees. He has a strong interest in epilepsy and neurogenetics and has ongoing research publications in both fields.

Received:March 15, 2023; **Accepted**: March 18, 2023; **Published**: May 02, 2023