Leila Vahedi, J Neurol Disord 2015, 3:4
http://dx.doi.org/10.4172/2329-6895.C1.006

OMICS International
conferenceseries.com

International Conference on
Epilepsy & Treatment
September 21-22, 2015 Baltimore, USA

First case report of Rett syndrome in the Azeri Turkish population and brief review of the literature

Leila Vahedi
Tabriz University of Medical Sciences, Iran

Rett syndrome is a dominant X-linked male-lethal disorder largely caused by mutations in the gene encoding methyl-CpG binding protein 2 (MECP2). Clinical manifestations include neurodevelopmental disorder characterized by early-onset intractable seizures, severe developmental delay, intellectual disability and abnormal electroencephalograms. Afflicted females show normal development until the age of 6 to 18 months followed by gradual loss of speech abilities, microcephaly, social impairment, ataxia and stereotypic hand movements. We report a 7-year-old girl who was born of a non-consanguineous marriage presenting with mental retardation and delayed development. Physical examination revealed loss of speech, repetitive hand-wringing movement, short stature (120 cm), strabismus, microcephaly and autistic behavior. The diagnosis was confirmed by sequencing MECP2 gene with heterozygous mutation C385A in exon 2. The current study aimed to report the first case of Rett syndrome in the Azeri Turkish population.

Biography

Leila Vahedi has completed her MD from Tabriz University School of Medicine and started her PhD studies in 2013 at Tabriz University of Medical Sciences. She is the student of Liver and Gastrointestinal Diseases Research Center. She has published more than 10 papers in reputed journals and has been serving as an Editorial Board Member of repute.

vahedi.l49@gmail.com

Notes: