Muhammad Imran Naseer et al., J Neurol Neurophysiol 2016, 7:6(Suppl)
http://dx.doi.org/10.4172/2155-9562.C1.037

A novel mutation in PGAP2 gene causes developmental delay, intellectual disability, epilepsy and microcephaly in consanguineous Saudi family

Muhammad Imran Naseer, Mahmood Rasool, Peter Natesan Pushparaj, Mohammed M Jan, Adeel G Chaudhary, Adel M Abuzenadah and **Mohammad H Al Qahtani**
King Abdulaziz University, Saudi Arabia

PGAP2 (Post-GPI Attachment to Proteins 2) gene is involved in lipid remodeling steps of glycosylphosphatidylinositol (GPI)-anchor maturation. At the surface of the cell this gene is required for proper expression of GPI-anchored proteins. Mutations in the gene cause hyperphosphatasia mental retardation syndrome-3. We have identified a large consanguineous family from Saudi origin segregating developmental delay, intellectual disability, epilepsy and microcephaly. Whole exome sequencing with 100x coverage was performed on two affected siblings of the family. Data analysis in the patient revealed a novel missence mutation c.191C>T in PGAP2 gene resulting in Alanine to Valine substitution (Ala64Val). Whole exome sequencing data analysis, confirmed by subsequent Sanger sequencing validation, identifies a novel mutation c.191C>T in exon 3 of PGAP2 gene. The mutation was ruled out in 100 unrelated healthy controls. We suggest that this pathogenic mutation disrupts the proper function of the gene proteins resulting in the disease state.

Biography

Muhammad Imran Naseer has completed his PhD from Gyeongsang National University, Jinju, South Korea and 1 year Post-doc from same university in the field of Molecular Neuroscience. In 2011, he joined CEGMR as Assistant Professor at King Abdulaziz University. He is the PI of two projects funded by the KACST and last year he was promoted as Associate Professor. He has published more than 50 papers in reputed journals and has been serving as an Editorial Board Member of repute.

mimrannaseer@yahoo.com

Notes: