Sana Sfar et al., J Bioengineer & Biomedical Sci 2015, 5:3
http://dx.doi.org/10.4172/2155-9538.C1.006

OMICS International
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2nd Biomedical Engineering Conference and Expo

November 30-December 01, 2015 San Antonio, USA

A novel CASR mutation in a Tunisian FHH/NSHPT family associated with a mental retardation

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The calcium-sensing receptor (CASR), a plasma membrane G-protein coupled receptor, is expressed in parathyroid gland and kidney, and controls systemic calcium homeostasis. Inactivating CASR mutations have previously been identified in patients with familial hypocalciuric hypercalcemia (FHH) and neonatal severe hyperparathyroidism (NSHPT). The aim of the our study is to determine the underlying molecular defect of FHH/NSHPT disease in a consanguineous Tunisian family. Mutation screening was carried out using RFLP-PCR and direct sequencing. We found that the proband is homozygous for a novel 15bp deletion in the exon 7 (c.1952_1966del) confirming the diagnosis of NSHPT. All the FHH members were found to be heterozygous for the novel detected mutation. The mutation, p.S651_L655del, leads to the deletion of 5 codons in the second transmembrane domain of the CASR which thought to be involved in the processes of ligand-induced signaling. This alteration was associated with the evidence of mental retardation in the FHH carriers and appears to be a novel inactivating mutation in the CASR gene. Our findings provide additional support for the implication of CASR gene in the FHH/NSHPT pathogenesis.

Biography

Sana SFAR is currently working as an Assistant professor at the Faculty of Pharmacy of Monastir-TUNISIA genetics and research Scientist in the research Unity "Congenital malformations epidemiology, etiology and therapeutic in Tunisia. She has completed her PhD in Biological Sciences and Biotechnology from the High Institute of Biotechnology of Monastir. She has published 13 articles in peer-reviewed journals and also an Editorial Board Member of the Global Journal of Genitourinary cancer. Her current research interests include genetics and molecular diagnostics of multiple diseases with genetic background.

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