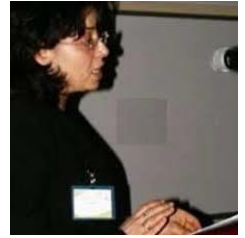


Mutations in domain III and IV of SCN5a in tunisian brugada patients

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Abstract

Mutations in the gene SCN5a is the most common genetic cause of Brugada Syndrome (BS), a rare inherited cardiac channelopathy, characterized by ST-segment elevation in the right pericardial leads V1-V3 and right bundle-branch block. BS presents with syncope and/or cardiac arrest due to ventricular fibrillation in normal structural heart. SCN5a encodes an α -subunit of the cardiac voltage-gated sodium channel (Nav1.5) at 3p21 with 28 exons encoding 2016 amino acids. More than 400 mutations are described and up of 150 mutations are located in domains III and IV of SCN5a. Here, we investigated nine Tunisian families in whom BS has been identified clinically. The aim was the optimisation of a genetic screening method for causative mutation in domain III and IV of SCN5a using High Resolution Melting (HRM). Sixteen patients were recruited from departments of cardiology of Sousse and Sfax, genetic family investigation, clinical data and informed consents are acquired. Genomic DNA extraction was performed by the phenol-chloroform method. Genetic analysis for the exons coding to the whole domains DIII and DIV have been carried out by polymerase chain reaction (PCR). HRM technical assessment has been optimized. HRM results have been verified by sequencing. As primary results, interpretation of clinical data and genetic family investigations are in favor of molecular basis for this channelopathy, which will be confirmed by HRM and sequencing. This study will define hotspot mutations in domain III and IV of SCN5a in Tunisian Brugada patients and families, to improve the genetics counseling and therapeutic management.

Biography:

Pr. Dr. Nouha Bouayed Abdelmoula graduated in medical, chromosomal and molecular genetics in Tunisian and French universities is a full university-hospital professor of medicine at the faculty of medicine of Sfax (Tunisia), genetic counselor, senior research supervisor and director of the research unit UR17ES36 entitled genomic of signalopathies at the service of medicine (MESRS). As an expert in genetics, She is the author of many scientific articles, and the designer on multiple issued and pending Md and PhD thesis. Mme Oldez Kaabi is one of his PhD thesis candidates working in the field of Brugada Syndrome genetics.

Speaker Publications:

1. "NFB-16. mToropathies and subependymal giant cell Astrocytomas: Predictive value of germinal TSC1/2 mutations screening in familial cases."
2. "TBIO-27. Rasopathies and Brain Tumorigenesis: are SOS1 mutations are concerned?"

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