Insights In Blood Pressure

Mutations in domain III and IV of SCN5a in Tunisian Brugada patients

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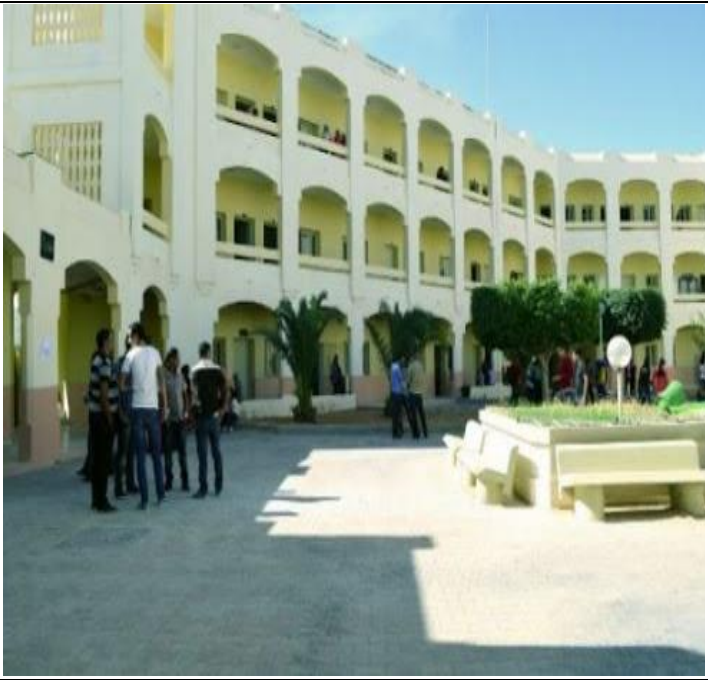
Abstract:

Background 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 leadsV1-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



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

Publications:

1. Skewed X-chromosome inactivation pattern in SRY positive XX maleness: a case report and review of literature
2. Genetic Diversity Using Random Amplified Polymorphic DNA (RAPD) Analysis for Aspergillus niger isolates
3. Au–Ag–Cu nanoparticles alloys showed antifangal activity against the antibiotics-resistant Candida albicans
4. Induce mutations for Bavistin resistance in Trichoderma harzianum by UV-irradation
5. Endocrine disruption and ovarian morphometric responses in rats following exposure to tetradifon

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