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Comparing the SCN5A Gene Sequence and Expression in Three Different Brugada Syndrome Profiles of Patients: New Insights for Genotype-Phenotype Correlation

Hajer Foddha[1,2], Houria Daimi[1], Abdelhak Foddha[3], Nadia Leban[1], Rahma Touhami[1,2], Habib Gamra[3], Ali Saad[4], Jemni Ben Chibani[1] and Amel Haj Khelil[1,2]

[1]Biochemistry and Molecular Biology Laboratory, Department of Clinical Biology, Faculty of Pharmacy, University of Monastir, 5000, Tunisia
[2]Department of Molecular and Cellular Biology and Biotechnology, Higher Institute of Biotechnology, University of Monastir, 5000, Tunisia
[3]Cardiology A Service, Fattouma Bourguiba Hospital, Monastir, 5000, Tunisia
[4]Cytogenetics, Molecular Genetics and Reproduction Biology Service, Farhat Hached Hospital, Sousse, 4000, Tunisia

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ABSTRACT Brugada Syndrome (BrS) is a rare inherited cardiac arrhythmia. In this paper, the researchers aim to identify the molecular defect in three unrelated patients with different clinical BrS profiles. Therefore, the researchers analysed, by real time PCR, the expression amount of SCN5A gene, which is the most described in BrS, in the three selected patients. Thereafter, the researchers sequenced the total exome and the untranslated regions of SCN5A. The results showed a variable decrease in SCN5A expression and no deleterious mutation, but 11 exonic and 3' UTR variants. The number of these variants is correlated to the gene expression and the phenotype severity in each patient. These results confirm the implication of SCN5A, and the researchers hypothesise that in the absence of causal mutation, the presence of one or numerous variants in the SCN5A gene could be involved in the disease. This finding may provide new insights in genotype-phenotype correlation and personalised diagnosis.