Extended Abstract Journal of Invasive and Non-Invasive Cardiology 2020
Vol.3 No.1

The utility of genetic testing in cardiac arrhythmias

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Introduction: The common life threatening cardiac arrhythmias, Long QT (LQTS type 1-13) and Brugada (BrS type 1-12) present with syncope/palpitations/ seizures/aborted cardiac arrest. They have incomplete penetrance and variable expressivity. The three common genes (KCNQ1, KCNH2 and SCN5A) account for 75% of all LQTS cases and SCN5A gene in BrS accounts for 25% of all cases.

Aim: To identify the causative variation in the associated genes responsible for causing cardiac channelopathies in Indian patients.

Materials & Methods: Hundred patients who fulfilled the inclusion criteria of the study were enrolled. Mutation analysis was performed in most probable candidate gene by direct sequencing using primers flanking exon-intron boundaries. If a mutation was not identified, NGS was performed to identify mutations in other cardiac genes in patients. Parents and siblings were screened if a mutation was identified in the proband. Novel mutations were evaluated for pathogenicity using ACMG guidelines, bioinformatics and molecular modelling softwares.

Results: Mutations was identified in 23 of 100 (23%) patients by Sanger sequencing, 20 had LQTS and 3 had BrS. Among the LQT syndromes, mutations were identified in 17 in KCNQ1 (LQTS1), one in KCNH2 (LQTS2) and two in SCN5A (LQTS3). Among the LQTS1 patients, ten were identified with biallelic mutations. The three BrS patients had mutations in SCN5A (BrS1). Ten of 23 mutations were novel. NGS identified mutation in 22 (49%) of 45 patients negative for mutations by Sanger and with significant family history and/or strong clinical indication. Of which, 20 had LQTS and two had BrS. Out of these 46 mutations, 18 were novel. Cascade screening identified mutations in two symptomatic and forty asymptomatic family members. Genetic counseling was provided to the proband and family members.

Conclusion: Genotyping is important for confirming type of LQTS/BrS, which has implications for management, cascade screening and risk assessment.

Biography

Bijal Vyas Bhatia has completed her MS in Medical Genetics from Virginia Commonwealth University, US and her PhD in Cardiac Genetics from Indraprastha University affiliated to Institute of Medical Genetics and Genomics, Sir Ganga Ram Hospital. She has been working in the field of Cardiac Genetics with specialization in Long QT and Brugada syndromes for last five years. She has written original research articles and review papers based on these syndromes. Her study was the first cohort study on cardiac arrhythmias in Indian patients that lead to establishment of genetic testing for arrhythmias in India.

This work is partly presented at Joint Event on 2nd Edition of EuroSciCon Congress on Heart Disease and Interventional Cardiology, February 25-26, 2019 in Paris, France