DOI: https://doi.org/10.3329/cardio.v15i1.61916

Case Report

Brugada Syndrome in a Female Patient from Bangladesh: A Rare Occurrence

Debasish Roychoudhury, Jacob Sam Koruth
Mount Sinai Hospital, New York

Key Words : *Brugada syndrome, Bangladeshi female, Sudden cardiac death, Genetics, Autism.*

Abstract:

Brugada syndrome is inherited in an autosomal dominant pattern which is associated with ventricular fibrillation and sudden cardiac death in a patient with structurally normal heart. The diagnosis is clinched on characteristic EKG pattern of >2 mm ST segment elevation followed by negative T wave in right precordial leads V1,2 either occurring spontaneously or with pharmacological provocation with a sodium channel blocker. Prevalence of this disease varies by geographic location and sex with highest preponderance in South East Asian countries and in males. Among South-East Asian countries, there has been only one case report so far from Bangladesh and that too in a male patient.

We present a case of female patient from Bangladesh with Brugada syndrome. We also report an association of Autism with Brugada syndrome in patient's only female child.

(Cardiovasc j 2022; 15(1): 86-87)

Case Report:

37 years old female was diagnosed with Brugada syndrome in Bangladesh in 2015 following an episode of aborted sudden cardiac death with cardioversion (presumed to be ventricular tachycardia). She underwent single chamber ICD implantation on 9/22/15 with no reported shocks since implant. She moved to United States in June, 2018 and underwent cardiac and electrophysiologic evaluation.

She denied any other syncopal episodes or near syncopal episodes in the past. Denied any chest pain, shortness of breath, palpitation or dizziness. Her past medical history is significant for diabetes and hypertension. No family history of sudden cardiac death or syncopal episode in first degree relative. She has one daughter who also has autism. Her medications include Metformin 500 mg po bid, Metoprolol Succinate ER 50 mg po qd. Her EKG showed normal sinus rhythm; rSr' pattern in V1/V2 with T wave inversion-Brugada type1 pattern (Fig. 1). Her echocardiogram revealed normal LV systolic function with LVEF 65% ; minimal anterior mitral leaflet prolapse with mild MR; mild TR with mild pulmonary hypertension (PA pressure 31-36 mmHg).

Patient was advised to take Acetaminophen early during any febrile illness to reduce risk of ventricular arrhythmias and to use caution prior to starting new medications that may need to be avoided with Brugada syndrome. She was subsequently referred for genetic testing for herself and her daughter. Patient was found to have pathogenic variant in SCN5A gene variant c.2678G>A(p.Arg893His); zygosity was heterozygous; variant classification was likely pathogenic. Her daughter was also tested and had same genetic disorder. She is being closely followed by pediatric cardiology.

Discussion:

In 1992 a report regarding 8 persons being resuscitated from sudden cardiac death which was caused by documented ventricular fibrillation (VF) was first published. An EKG characterized by ST elevation in right precordial leads was identified in these individuals and all of them had structurally normal heart. In 1996 this

Address of correspondence: Debasish Roychoudhury, NewYork Presbyterian Medical Group/Queens. Mount Sinai Hospital. New York, USA. Email: roychoudhury03@gmail.com

arrhythmogenic entity was named Brugada syndrome.[1] As far back as 1917, reports from Philippines described sudden deaths in healthy young males during sleep with no pathological evidence to explain the cause of the death.[2] This same disease entity was known by all different names in different parts of Southeast Asian countries where the disease is most prevalent with the notable exception of Bangladesh. It is responsible for 8-10% of all causes of sudden death and 20 % of sudden death in patients with structurally normal heart. Also, sex predilection is much higher in males about 8-10 times more in men than women.[3]

Medline search reveals only one case report published so far which is a male patient with genetic testing published in 2014.[4] Our patient is a woman from Bangladesh whose genetic testing was done in USA. Furthermore, we report a very rare genetic association between Brugada and autism in her daughter.

Differences in sex predilection could be due to transmembrane ion current expression between sexes with higher concentration of testosterone noted in men with Brugada.

Genetic alteration in SCN5A gene associated with Brugada syndrome was first described in 1998. This gene encodes the alpha subunit of the cardiac sodium channel. This genetic variation is responsible for 30% of all cases. It has an autosomal dominant pattern of inheritance. The prevalence is approximately 1 in 5000 to 1 in 2000(3). Mutations in as many as 23 genes have been described in some cases of Brugada syndrome and can also be a cause for Autism Spectrum Disorders. The risk for autism in the general population is 1:50 to 1:80. Genes SCN2A on chromosome 2q24.3 and SCN5A on chromosome 3p22.2 are 2 of the more than 18 genes associated with Brugada syndrome and autism. Approximately 30% of Brugada syndrome are associated with mutations in the SCN5A gene. 65-70% of cases are still in the unknown or uncertain group.[5,6]

Conclusion:

Brugada syndrome is a rare genetic disorder with characteristic EKG pattern and propensity for sudden cardiac death due to ventricular fibrillation. It is mostly prevalent in South Asian countries although Bangladesh is so far an exception. It has 8-10 times more predilection for male than female population. Our case is unique in that it hails from Bangladesh and furthermore in a female patient. There is only one such genetically determined case report published prior to this. Our patient also had a daughter with autism and genetic predilection for Brugada syndrome. Although such association is known in the genetic literature, no such case report has been published so far to our knowledge.

Our case report enhances the knowledge of this rare genetically inherited disease particularly in Bangladesh which is the harbinger of sudden cardiac death as illustrated in this case and also it highlights the modality of treatment with ICD as in our patient. It also elucidates a very rare genetic association between Brugada syndrome and autistic spectrum disorder in a female child also from Bangladesh.

We hope to be able to contribute through our case report to raising awareness of this rare disease entity in Bangladesh and also worldwide

Conflict of Interest - None.

References:

1. Brugada P, Brugada J. Right bundle branch block, persistent ST segment elevation and sudden cardiac death: a distinct clinical and electrocardiographic syndrome. A multicenter report. J Am Coll Cardiol. 1992;20(6):1391-1396. doi:10.1016/0735-1097(92)90253-j
2. Guazon MP. Algunas notas sobre bangungut. Revista Filipina de Medicina Y Farmacia. 1917;8:437–442
3. Brugada J, Campuzano O, Arbelo E, Sarquella-Brugada G, Brugada R. Present Status of Brugada Syndrome: JACC State-of-the-Art Review. J Am Coll Cardiol. 2018;72(9):1046-1059. doi:10.1016/j.jacc.2018.06.037
4. Sayeed MZ, Salam MA, Haque MZ, Islam AK. Brugada syndrome with a novel missense mutation in SCN5A gene: a case report from Bangladesh. Indian Heart J. 2014;66(1):104-107. doi:10.1016/j.ihj.2013.12.003
5. Sarquella-Brugada G, Campuzano O, Arbelo E, Brugada J, Brugada R. Brugada syndrome: clinical and genetic findings. Genet Med. 2016;18(1):3-12. doi:10.1038/gim.2015.35
6. Delise P, Probst V, Allocca G et al. Clinical outcome of patients with the Brugada type 1 electrocardiogram without prophylactic implantable cardioverter defibrillator in primary prevention: a cumulative analysis of seven large prospective studies. EP Europace. 2017;20(FI1):f77-f85. doi:10.1093/europace/eux226