COMMENTARY

Brugada Syndrome: An Easily Overlooked Arrhythmic Disorder

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Abstract

Brugada Syndrome (BrS) is an autosomal dominant and congenital heart disorder that often remains undiagnosed. BrS can present with no symptoms, and when symptoms do occur, the presentation can be similar to that of many other illnesses. The disorder is often detected on an electrocardiogram (EKG). When patients display symptoms, they can present as palpitations, fainting, dizziness, syncope, seizures, and lightheadedness. BrS can present as one cause of sudden cardiac death (SCD); hence, early detection and management is crucial. BrS is managed with an Implantable Cardioverter/Defibrillator (ICD) device, a pacemaker, or a combination of antiarrhythmic medicines.

Keywords: *Brugada syndrome, arrhythmias, sudden cardiac death, ventricular tachycardia, ventricular fibrillation, rare disorders.*

INTRODUCTION

Brugada Syndrome (BrS), first described by Josep and Pedro Brugada in 1992, is a rare genetic disorder of the heart that affects the electrical conduction system, resulting in dysrhythmia. The disease usually presents among adults and many times results in sudden cardiac death (SCD) [1-3]. Although many patients do not display any symptoms, BrS increases the risk of SCD, and about 20% of SCDs among patients with structurally normal hearts are associated with this disorder. [2]. The symptoms, when present, can include fainting spells, dizziness, breathing difficulties, and seizures [2].

Etiological and Genetic Factors

BrS is an easily underdiagnosed disorder, and the epidemiology is often inaccurate. However, studies suggest that it occurs in about five out of every ten thousand individuals and many times coexists or associates with other genetic disorders such as Schizophrenia and Ehlers-Danlos syndrome [4]. Association with other genetic disorders can aid in the early detection of BrS, particularly when patients begin to exhibit heart rhythm abnormalities.

Genetic examination of BrS reveals that it is associated with a pathogenic variant on the 3rd chromosome that results in a sodium channelopathy. This rare variation can exist even with an anatomically normal heart. While numerous gene variants are linked to BrS and other heart conditions, the *SCN5A* variant is the most frequently observed. Additionally, the *SCN10A* gene is commonly associated with BrS and is found in ~17% of patients [4, 5]. Scientists have identified around 20 genes that may be linked to BrS; however, these genes are found in only 30% of sufferers, making BrS a heterogeneous condition [6, 7].

These pathogenic variations may occur in the heart's sodium, potassium, or calcium channels, or in proteins that regulate these ion channels. *SCN5A* gene mutations are present in 20-30% of cases, while alterations in other genes are present in 5% of patients [6]. The genetic variation results in abnormalities in heart rhythm, with symptoms often manifested during sleep, at rest, or following large meals [8]. Due to its tendency to cause sudden death during sleep, it is also referred to as sudden unexplained nocturnal death syndrome (SUDS) [8]. Among survivors, arrhythmias recur in approximately eight percent of cases, and the risk of SCD can persist for decades after diagnosis [5, 6].

In addition to *SCN5A* aberration, other risk factors include family history of SCD (and/or BrS), demonstration of Type 1 BrS pattern on an electrocardiogram (EKG), presence of ventricular tachycardia during an electrophysiology (EP) testing, and demographic factors such as male sex, age above forty, and Asian ancestry [9-11].

Clinical Manifestations

BrS presents with a wide range of symptoms, from being entirely asymptomatic to causing syncope or even sudden cardiac death. Some signs can resemble those of other conditions, which often leads to delayed or missed diagnosis of this potentially fatal disorder.

While fainting or syncope is the most recognized symptom, others include dizziness, heart palpitations, unexpected arrhythmias, and seizure episodes [9]. Atrial fibrillation (A-fib) is the second most common arrhythmia in BrS, following ventricular fibrillation (V-fib) [3].

BrS can be classified into three variations. Type 1, identified on EKG by a distinctive 2-millimeter J-point

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Received: May 13, 2025; Revised: July 20, 2025; Accepted: August 12, 2025
DOI: https://doi.org/10.37184/jlnh.2959-1805.3.51

  ISSN: 2960-2963 (Online) *Journal of Liaquat National Hospital 2026; 4(1): 96-99*

elevation, a coved-shaped ST-segment, and a subsequent negative T-wave in one of the right precordial leads (V1–V3), is the most common and most severe. Type 2 features a gradually descending ST-segment elevation that remains at or below 1 mm above the baseline, followed by a T-wave that creates a saddle-back appearance. Type 3 exhibits characteristics of both Type 1 and Type 2, with ST-segment elevation in the right precordial leads measuring less than 2 mm [10].

BrS is less commonly observed in pediatric age groups than in adult populations, although recent evidence indicates an increasing prevalence among children [12]. Although symptoms can begin in infancy or childhood, most individuals experience symptom onset in their 40s or 50s; hence, it is rare for a pediatric member of a family to be the 1st symptomatic individual with BrS [12]. Most children do not display any symptoms, but when they do, pediatric patients typically present with atrial arrhythmias and sinus node dysfunction. Less commonly, they may experience ventricular arrhythmias and sudden cardiac death (SCD). Deaths related to ventricular fibrillation are also common. Furthermore, severe breath-holding spells (a type of syncope), especially if repetitive or accompanied by loss of consciousness and/or sphincter control, may be suggestive of BrS [10, 13].

Variations in the *SCN5A* gene have been identified in 10-15% of sudden infant death syndrome (SIDS) cases. Many families affected by SIDS also have a history of sudden cardiac events [12]. BrS is an autosomal dominant disorder, meaning that only one parent needs to pass along the gene to their offspring. There is a 50% chance that a parent carrying the mutated gene will pass it on to their children, although not all offspring of a parent with BrS will develop the syndrome. While BrS prevalence shows no gender difference before adolescence, it occurs more frequently in adult males, affecting approximately 70-80% of cases. Studies suggest this higher prevalence may be linked to elevated testosterone levels in affected males compared to healthy peers [11, 13]. Testosterone is believed to shorten the action potential in the right ventricular epicardium, potentially leading to ventricular dysrhythmias [13].

Diagnosis and Treatment

BrS is diagnosed by a combination of genetic testing, EKG, and electrophysiology (EP) studies. Electrolyte panels and additional tests are essential in ruling out other causes of signs and symptoms [14, 15]. Diagnosing BrS can be challenging because its characteristic EKG patterns do not consistently appear during routine monitoring. When present, the hallmark findings include ST-segment elevation in the right precordial leads (V1–V3) and T-wave inversion [1]. Typically, the EKG pattern specific to BrS emerges only during symptomatic episodes, requiring recordings during arrhythmia, though there are exceptions to this rule. The transient nature of these patterns requires repeated EKG assessments for accurate detection. Sodium channel blockers such as Ajmaline or Flecainide are also used to unmask latent BrS by inducing symptoms. Although genetic testing has become more widely available, it is typically only recommended for first-degree relatives [5]. Evidence suggests that patients with the SCN5A gene are at greater risk of developing or presenting with arrhythmic episodes compared with those without this gene [16].

Management of BrS includes a Pacemaker/Implanted Cardioverter-Defibrillator (ICD) or a subcutaneous ICD (S-ICD), which are the gold standard treatments for the condition. Having an ICD has been a proven method to prevent SCD and reduce morbidity and mortality among sufferers [9, 10]. Combining catheter ablation with ICD implantation has been associated with better outcomes [16]. Additionally, genetic testing and counseling play a crucial role in management, as they help establish risk assessment and stratification once a diagnosis has been confirmed [17].

Symptomatic treatment approaches are essential in the management of BrS. Fever and arrhythmia can be controlled with antipyretics (*e.g.*, acetaminophen, ibuprofen) and antiarrhythmic drugs (*e.g.*, Quinidine, a class-I medication), respectively [10, 18]. Although antipyretics do not treat the syndrome itself, they are essential for managing fever, which is known to trigger arrhythmias [18]. Risk stratification and treatment depend on symptoms, EKG/EP findings, and family history of BrS.

DISCUSSION

BrS is a severe heart rhythm disorder that requires ongoing collaborative care between patients and providers. Although knowledge about BrS continues to grow, many aspects of the condition, including its causes and optimal management strategies, remain evolving.

Despite significant advancements in diagnostics and management, challenges persist in diagnosing and treating BrS. EKG patterns can be intermittent, leading to false-negative results. Provocative testing, such as the administration of ajmaline, can induce arrhythmias and related complications, requiring close supervision. Furthermore, while genetic testing can be valuable, it has limitations and may not definitively rule out the diagnosis.

The use of ICDs also carries certain risks; for instance, infections at the implant site may occur, and equipment malfunction remains a potential concern [8, 9].

Living with BrS involves strategies beyond medical care. Long-term outcomes depend on lifestyle changes such as a healthy diet, physical activity, avoiding medications that could worsen the condition (www.brugadadrugs.org), recognizing early warning signs, and continuous communication with healthcare providers. Even with appropriate care, BrS can still present risks. Arrhythmias may persist, and in rare cases, sudden cardiac death can occur if the ICD fails to restore a normal heart rhythm. Some patients may also experience side effects from medications, requiring multiple adjustments, which can be physically and emotionally taxing. Patients with additional chronic health conditions may also experience better outcomes with BrS if they receive appropriate management and adopt lifestyle interventions targeting modifiable risk factors [19].

Device-related complications can also arise. These may include inappropriate shocks, lead malfunctions, device shifting, or battery issues. Though ICD batteries typically last several years, they may need early replacement, often involving another surgical procedure.

The psychological toll of BrS can be just as significant as the physical impact. The stress of living with a potentially life-threatening condition can lead to anxiety, depression, and body-image concerns, all of which can reduce quality of life. Living with ICD can be stressful; hence, appropriate regular counseling and follow-up are essential. Family members may also struggle with the emotional burden of the diagnosis. Regular emotional check-ins and support from mental health professionals can be critical in helping patients and their families.

Ongoing research studies, especially those that explore both clinical outcomes and patient experiences and employ mixed-methods, are essential for understanding diseases like BrS [20, 21]. Public health initiatives aimed at increasing awareness among the public and healthcare professionals are crucial and can help in the early detection of this rare disorder. Public health campaigns can specifically focus on promoting healthier lifestyles, recognizing warning signs, screening family members, encouraging genetic testing and counseling, and guiding the public toward appropriate disease management.

CONCLUSION

Brugada syndrome is an uncommon but serious inherited arrhythmic disorder that is often missed because of its variable presentation and transient electrocardiographic findings. Although the heart is structurally normal, affected individuals remain at risk for malignant ventricular arrhythmias and sudden cardiac death. Early recognition, careful risk assessment, and timely treatment, particularly ICD placement in high-risk patients, are key to reducing morbidity and mortality. Greater awareness among clinicians, along with family screening and continued research, is essential to improve diagnosis and long-term management of BrS.

CONFLICT OF INTEREST

The author declares no conflict of interest.

ACKNOWLEDGEMENTS

Declared none.

AUTHOR'S CONTRIBUTION

Author conceptualized, performed literature review, drafted, and finalized the final manuscript.

GENERATIVE AI AND AI-ASSISTED TECHNOLOGIES IN THE WRITING PROCESS

During the preparation of this work the author(s) limitedly used ChatGPT (GPT-4, OpenAI) to get language suggestions and do minor proofreading in some parts of the manuscript. After using this tool/service, the author(s) reviewed and edited the content as needed and take(s) full responsibility for the content of the published article.

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