**International Journal of Medical and Health Research**
**ISSN: 2454-9142**
**Received: 03-08-2019; Accepted: 05-09-2019**
**www.medicalsciencejournal.com**
**Volume 5; Issue 11; November 2019; Page No. 11-12**

# Brugada syndrome in children: Stepping into unchartered territory

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**Abstract**
Brugada syndrome (BrS) is an autosomal dominant inherited channelopathy. It is associated with a typical pattern of ST-segment elevation in the precordial leads V1–V3 and potentially lethal ventricular arrhythmias in otherwise healthy patients. It is frequently seen in young Asian males, in whom it has previously been described as sudden unexplained nocturnal death syndrome. Although it typically presents in young adults, it is also known to present in children and infants, especially in the presence of fever. Our understanding of the genetic pathogenesis and management of BrS has grown substantially considering that it has only been 24 years since its first description as a unique clinical entity. However, there remains much to be learned, especially in the pediatric population. This review aims to discuss the epidemiology, genetics, and pathogenesis of BrS, new innovations in the diagnosis, prognostication, risk stratification, and management of BrS.

**Keywords:** Brugada syndrome, Brugada pattern, children

## Introduction

### Definition of Brugada Syndrome

Brugada [brew-GAH-dah] syndrome is a potentially life-threatening heart rhythm disorder that is often inherited. If your child has Brugada syndrome, they will have an increased risk of abnormal heart rhythms from the lower chambers of the heart, also called ventricular [ven-TRIK-yoo-lar] arrhythmias [uh-RITH-me-uh]. An arrhythmia is when the heart beats irregularly. This can disrupt circulation and lead to fainting or sometimes death. Problems most often happen during sleep.

Brugada syndrome is rarely diagnosed in children. This syndrome may explain some cases of sudden infant death syndrome.

### Disease Burden of Brugada Syndrome

ECG findings are a requisite for the diagnosis of BrS, it is hard to know the true prevalence of BrS as presumably many patients are asymptomatic. Different studies propose varying prevalence rates ranging from 1 in 5000 to 1 in 2000. BrS is thought to be responsible for 4–12% of sudden cardiac death in children and young athletes. In some studies, it is implicated in 20% of sudden unexplained death in the young.10%–20% of sudden infant death syndrome is due to identifiable inherited channelopathies including BrS.

### Molecular Genetics of Brugada Syndrome

Genetic abnormalities have been discovered in several arrhythmic disorders, including the long QT (LQT) syndrome, arrhythmogenic right ventricular dysplasia, conduction system disease (Lenegre's disease), and the Brugada syndrome . About 20% of patients with Brugada syndrome have documented SCN5A mutations . Genetic studies have demonstrated that some cases of Brugada syndrome and chromosome 3–linked long-QT syndrome (LQT3) are allelic disorders othe cardiac sodium channel gene (SCN5A, 3p21). Three types of SCN5A mutations have been identified in the Brugada syndrome: splice-donor, frame-shift, and missense . All of these lead to a reduction in the fast sodium channel current. In LQT3 the defect in the sodium channel causes a persistent late sodium current. An autosomal dominant SCN5A gene abnormality has also been shown to underlie a progressive cardiac conduction system disease (Lev's or Lenegre's disease).

### Inheritance of Brugada Syndrome

The genetic form of Brugada syndrome (not the acquired form) is inherited in an autosomal dominant manner. This means that having one mutated copy of the responsible gene in each cell is enough to cause signs or symptoms. Almost all people with Brugada syndrome have a parent with the condition. In about 1% of cases, an affected person has a new mutation in the responsible gene and has no family history of the condition. Each child of an affected person has a 50% chance to inherit the mutated gene.

Unsymptomatic parents of an affected person should be evaluated with electrocardiography, and any family history of sudden death should be discussed. If genetic testing reveals a mutation in the affected person, genetic testing of the parents is recommended. A family history may *appear* to be negative due to reduced penetrance, death of a parent before symptoms start, or late onset of symptoms in an affected parent.

### Symptoms of Brugada Syndrome

Many children who have Brugada syndrome remain asymptomatic because they didn't produce any noticeable symptoms. If there are any symptom, they often occur at night and include

- Irregular Heartbeat
- Fainting
- Palpitations
- Chaotic Heartbeat ( Very Fast)
- Dyspnea

- Seizures
- Sudden Cardiac arrest

**Diagnosis of Brugada Syndrome**

The diagnosis of Brugada syndrome is based on a

- Thorough clinical evaluation,
- A complete medical and family history that may include a family history of sudden cardiac death, and a specialized test known as an electrocardiogram (ECG).
- Molecular genetic (DNA) testing is available for mutations in all genes to confirm the diagnosis but only about 30-35% of affected individuals have an identifiable gene mutation after a comprehensive genetic test. Sequence analysis of the *SCN5A* gene is the first step in making a molecular genetic diagnosis because mutations in this gene are the most common cause of Brugada syndrome (nearly 25%).

**Prognosis and Management of Brugada syndrome**

The prognosis is poor, whether the patient is symptomatic or asymptomatic, with a 10% per year mortality.

- Antiarrhythmic drugs, such as beta blockers and amiodarone, appear to be of little use in prolonging survival. Use of beta blockers is still associated with a 10% per year death rate. Beta blockade worsens whereas beta stimulation usually reduces ST segment elevation. Class IC agents can be used to express the ST segment elevation in latent cases.
- The treatment of choice is implantation of an implantable cardioverter-defibrillator (ICD).

**Note: C**ompared with no therapy, beta blockers, or amiodarone, ICDs statistically ($P = 0.0009$) prevent sudden cardiac death (personal communication, Josep and Pedro Brugada).

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