

Brugada Syndrome

A Guide for Patients and Health Care Providers

A note from the SADS Foundation.

We provide this information with the hope that informing physicians, other health care providers, and the public improve recognition, resulting in the reduction and ultimately elimination of cardiac arrest and sudden death from the Brugada Syndrome.

Why do I need to know about Brugada Syndrome?

Because this is a relatively recently identified condition we are uncertain of the true incidence of Brugada. It is thought responsible for between 4 and 12% of all unexpected sudden deaths and possibly as many as 50% of all sudden deaths in patients with an apparently normal heart.

Physicians need to know:

- When to consider Brugada as a possible diagnosis.
- When to refer patients for diagnosis & treatment.
- About genetic testing for Brugada Syndrome.
- How to develop a family pedigree and screen family members for Brugada Syndrome.

Patients and Parents need to know:

- The warning signs and symptoms of Brugada Syndrome.
- Who to see for proper testing.
- How to protect their children and themselves.
- How to expand their family pedigree and contact other family members who may be at risk.

What is Brugada Syndrome?

Brugada Syndrome is an inheritable disease that results in a disturbance of the heart's electrical system. Like LQTS, Brugada Syndrome is due to an abnormality in a cardiac ion channel. Thus far the ion channel abnormality (mutation) identified in Brugada Syndrome is of the cardiac sodium channel. However, many patients with clinical Brugada Syndrome in whom genetic testing has been done do not have this mutation thus, additional, yet undiscovered defects exist. Our window to this electrical activity of the heart is through an electrocardiogram (ECG). The ECG in Brugada Syndrome can be normal at times and abnormal at other times. When abnormal there are changes in the ST segments. Fever and certain medications can bring out these ECG changes.



What are the symptoms of Brugada Syndrome?

This is a disease that affects males more than females with an average age of onset of 40 years. Symptoms, like the abnormal ECG appearance, occur more commonly with a fever. The symptoms of Brugada Syndrome are syncope (fainting) and sudden death due to a fast polymorphic ventricular tachycardia (fast and unstable rhythm from the ventricle that does not allow the heart to pump adequately). If the ventricular

arrhythmia stops by itself then the symptom is fainting. If it does not stop then a cardiac arrest or sudden death can occur. Other symptoms include seizures, unexplained nighttime urination or strange breathing with sleep. Symptoms are more common during sleep. Some patients do not have symptoms. The symptoms of Brugada Syndrome can and are confused with the common faint.

When should the diagnosis be suspected?

- In any young person with **unexplained syncope (fainting), unexplained seizures, or unexplained cardiac arrest or sudden death.**
- When there is a **family history** of unexplained syncope, unexplained seizures, or sudden death in young people.
- When the autopsy is normal following the sudden and unexpected death of a young person

How is the diagnosis made?

- Abnormal resting ECG
 - Multiple ECGs may be needed
 - ECGs at times of fever
- Drug provocation study (procainamide)
- Genetic testing
 - Only positive in 30% of patients

How is Brugada Syndrome inherited, and who in a known or suspected family should be tested?

Brugada Syndrome is usually inherited by autosomal dominant transmission. This means that it generally should affect boys and girls equally, and that each child of an affected parent has a 50% chance of inheriting the genetic abnormality. In a really large family, close to 50% of the children would inherit the genetic abnormality. In average size families, it can range from all to none as each child has an independent 50/50 chance of inheriting the particular disease gene. Once a family member is identified with Brugada Syndrome, it is extremely important that other family members be tested for the syndrome. It is especially important to know which parent and grandparent has the abnormality, since brothers and sisters, aunts, uncles, nephews, nieces, and cousins on the affected side are potentially at risk.

Screening ECGs in family members of a patient with Brugada Syndrome is important but because the ECG can be different at different times it is more difficult and additionally ECGs, ECGs with fever or drug provocation testing may be needed.

What about genetic testing?

Since 2004, LQTS genetic testing has been a commercially available, clinical diagnostic test. As research about Brugada Syndrome continues more mutations will be identified and incorporated into the test and will increase the usefulness of genetic testing in Brugada Syndrome. Your physician will order the initial test on one family member (so-called index case). Once/if a family member has a gene mutation identified, testing of other family members for that specific mutation is available, and can assist in clarifying those family members with non-definitive ECG findings. If a genetic diagnosis of Brugada Syndrome is established for the index case, then the genetic test is definitive and will rule in or rule out the diagnosis of Brugada Syndrome for family members and relatives.

What is the treatment and who should be treated?

There are few options for treatment for Brugada Syndrome. At present, the ICD is used for patients with symptoms and a definitive diagnosis. An ICD is not the answer for every patient. There are some data to suggest Quinidine may help some patients. Close monitoring during times of fever is helpful. Research is ongoing concerning other drug therapy for Brugada Syndrome.

How can parents protect their kids?

- See the doctor regularly for follow-up. If on a medication the dose needs to be adjusted as the child grows. Make sure you see the doctor at least once a year, more frequently during very rapid growth, and discuss the need for dose changes. ICD patients need to be seen at least twice a year to check the device.
- Get additional medical advice if you are not comfortable with how things are going. Ideally, every patient/family with Brugada Syndrome should be cared for by a heart rhythm specialist (cardiac electrophysiologist). Do not hesitate to obtain a second opinion if you have any questions about your or your child's treatment.
- Make sure your family has an AED (automatic external defibrillator) and/or your child's school district has AED programs in their schools.
- A useful website for more information about Brugada Syndrome: <http://www.brugada.org>

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