OPINION

The brugada syndrome is a rare arrhythmia disorder with complex inheritance

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ABSTRACT

For the past 10 years, the enormous variety of the human genome has been exposed by using new sequencing technology to examine hundreds of entire exomes. Therefore, extreme care should be exercised to prevent misunderstanding when linking uncommon genetic variations to illness vulnerability. An uncommon genetic arrhythmia disorder called Brugada Syndrome (BrS) puts young adults at a significant risk of dying suddenly from cardiac arrest. Incomplete penetrance and the autosomal dominant mode of transmission have long been associated with mendelian inheritance in families. The 23 genes that were previously linked to the disease have all been found through a candidate gene technique, with the exception of one. So far, only a few uncommon *SCN5A* gene coding variations have been found to be strongly linked to the condition thus far. The complicated pattern of inheritance of BrS is illustrated by genotype/phenotype investigations done in families with *SCN5A* mutations, though. The recent discovery of common polymorphic alleles that are highly related to disease risk has verified this genetic complexity. Due to the implications of both common and rare variations on BrS susceptibility, a good genetic model for BrS predisposition should be defined before using molecular diagnosis. Although there is still a long way to go before BrS can be treated specifically for each patient, the significant phenotypic variability shown in familial variants of the disease may have some connection to this particular genetic architecture.

Key Words: *Vulnerability; Brugada syndrome; Genotype; Polymorphic; Mendelian inheritence*

INTRODUCTION

The Brugada Syndrome (BrS), a rare genetic arrhythmia condition, was originally identified in 1992 and raises the risk of ventricular fib--rillation in young adults who appear healthy. It is thought to be responsible for 4%–12% of SCD cases in the general population and for at least 20% of SCD cases in people with structurally normal hearts. A unique Electrocardiographic (ECG) pattern identified during three consecutive consensus conferences serves as the foundation for clinical diagnosis. This ECG pattern, formerly known as "type 1," is defined as a 0.2 mV ST segment elevation with a coved-type morphology in one of the right precordial leads V1 and V2, located in the second, third, or fourth intercostal space, occurring either spontaneously or following a provocative drug test with intravenous class Class I antiarrhythmic drug administration. Infected patients' ECG patterns may change briefly. Unmasking medications such as ajmaline, flecainide, and procainamide, which have higher sensitivity than flecainide and procainamide and can be used to disclose this pattern, can be used to solve this problem.

The ECG pattern's great degree of variability makes it difficult to determine with accuracy how often it occurs in the general population. Results from epidemiological studies on the prevalence of BrS around the world have been inconsistent. BrS prevalence is thought to be 5 per 10,000 in western Europe and the USA, whereas it appears to be 20 per 10,000 in Southeast Asia.

Aborted SCD is frequently the initial indication of BrS, with a mean age of 45 years at diagnosis and a 4-times higher frequency in men

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than in women. The affected patients are discovered in one-third of cases after syncope, which is typically preceded by vagal symptoms. It is difficult for the practitioner to differentiate between arrhythmic and non-arrhythmic etiologies since the syncope could be caused by non-sustained VF or a vaso-vagal episode without obvious clinical relevance Most individuals are asymptomatic at the time of diagnosis. With a 48% rate of suitable device therapy at 10 years in patients with prior aborted sudden death, defibrillator implantation remains the only effective therapy in high-risk patients. When implants are placed in asymptomatic patients, this rate drops to 12%, with many affected people remaining asymptomatic their whole lives. Additionally, there is a 30% risk of device-related problems at a 10-year follow-up, primarily as a result of lead dysfunction, ineffective therapy, and infection. Due to these severe adverse effects and the extremely low arrhythmic risk for asymptomatic people, precise risk assessment and/or effective pharmacological therapy are necessary. Risk categorization in BrS is limited by a small number of clinical characteristics. The usefulness of ventricular stimulation is still debatable, and the two main factors permitting risk categorization for SCD continue to be symptoms and spontaneous ECG pattern. Medical treatments that could lower arrhythmia incidence and stop SCD are still required. Since promising trials were reported in small patient populations, quinidine has been predicted to be "the medicine" for BrS since. However, a number of subsequent studies fell short of proving its advantageous effects.

Evidence is mounting that implantable defibrillators are a reliable and efficient treatment for symptomatic individuals. For asymptomatic individuals, numerous clinical markers have also been presented. However, risk prediction in this patient population is still difficult due to the scarcity of reproducible and trustworthy data.