GENEReviews

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Brugada Syndrome

[*Sudden Unexpected Nocturnal Death Syndrome*]

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Summary

Disease characteristics. Brugada syndrome is characterized by ST-segment abnormalities in leads V_1-V_3 on ECG and a high risk of ventricular arrhythmias and sudden death. Brugada syndrome presents primarily during adulthood; age at diagnosis ranges from two days to 85 years. The mean age of sudden death is approximately 40 years. Clinical presentations may also include sudden infant death syndrome (SIDS) (death of a child during the first year of life without an identifiable cause) and the sudden unexpected nocturnal death syndrome, a typical presentation in individuals from Southeast Asia.

Diagnosis/testing. Diagnosis is based on clinical findings. *SCN5A*, the gene encoding the α-subunit of the sodium channel, is the only gene currently known to be associated with Brugada syndrome. A second locus has been found, but the causative gene has not yet been identified. Molecular genetic testing of *SCN5A* identifies mutations in approximately 20%-25% of individuals with Brugada syndrome. Such testing is available clinically.

Management. *Treatment of manifestations:* implantable cardioverter defibrillator (ICD) in individuals with a history of syncope or cardiac arrest; isoproterenol for electrical storms. *Prevention of primary manifestations:* quinidine (1-2 g daily). Treatment of asymptomatic individuals is controversial. *Surveillance:* ECG monitoring every one to two years for at-risk individuals with a family history of Brugada syndrome. *Agents/circumstances to avoid:* high fever, anesthetics, antidepressant drugs, and antipsychotic drugs with sodium-blocking effects. *Testing of relatives at risk:* identification of relatives at risk using ECG or (if the disease-causing mutation in the family is known) molecular genetic testing enables use of preventive measures and avoidance of medications that can induce ventricular arrhythmias.

Genetic counseling. Brugada syndrome is inherited in an autosomal dominant manner. Most individuals diagnosed with Brugada syndrome have an affected parent. The proportion of cases caused by *de novo* mutations is estimated at 1%. Each child of an individual with Brugada syndrome has a 50% chance of inheriting the mutation. Prenatal testing for pregnancies at increased risk may be available through laboratories offering custom prenatal testing if the disease-causing mutation in the family is known.

Diagnosis

Clinical Diagnosis

The diagnosis of Brugada syndrome is **confirmed** in an individual with the following:

- **Type 1 ECG** (elevation of the J wave ≥2 mm with a negative T wave and ST segment that is coved type and gradually descending) in more than one right precordial lead (V_1-V_3)*, with or without administration of a sodium channel blocker (i.e., flecainide, pilsicainide, ajmaline, or procainamide)

  and

- **A and/or B:**

  A At least one of the following findings:

  - Documented ventricular fibrillation
  - Self-terminating polymorphic ventricular tachycardia
  - A family history of sudden cardiac death
  - Coved-type ECGs in family members
  - Electrophysiologic inducibility
  - Syncope or nocturnal agonal respiration

  B An *SCN5A* mutation

* No other factor(s) should account for the ECG abnormality.

Brugada syndrome should be **strongly considered** in individuals with either of the two following ECG types:

- **Type 2 ECG** (elevation of the J wave ≥2 mm with a positive or biphasic T wave; ST segment has a saddle-back configuration and is elevated ≥1 mm) in more than one right precordial lead under baseline conditions with conversion to type 1 ECG following challenge with a sodium channel blocker (considered equivalent to a positive finding in A.).

  Note: Drug-induced ST-segment elevation to a value greater than 2 mm should raise the possibility of Brugada syndrome when one or more of the clinical criteria are present (see A.). Based on current limited knowledge, whether an individual with a negative drug test (i.e., no change observed in the ST segment in response to a sodium channel blocker) has Brugada syndrome is unknown because the sensitivity of the test is 80% with the sodium channel blocker ajmaline, and probably lower for the other class 1 blockers [Hong, Brugada et al 2004].

- **Type 3 ECG** (elevation of the J wave ≥2 mm with a positive T wave; ST segment has a saddle-back configuration and is elevated <1 mm) in more than one lead under baseline conditions with conversion to type 1 ECG following challenge with a sodium channel blocker (considered equivalent to a positive finding in A.).

Note: Drug-induced conversion of type 3 ECG to type 2 ECG is inconclusive.

See Figure 1 for a characteristic ECG observed in an individual with Brugada syndrome.

Molecular Genetic Testing

GeneReviews designates a molecular genetic test as clinically available only if the test is listed in the GeneTests Laboratory Directory *by either a US CLIA-licensed laboratory or a non-US clinical laboratory. GeneTests does not verify laboratory-submitted information or warrant any aspect of a laboratory's licensure or performance. Clinicians must communicate directly with the laboratories to verify information.*—ED.

Gene. *SCN5A*, the gene encoding the α-subunit of the sodium channel, is the only gene currently known to be associated with Brugada syndrome.

Other loci. In 2002, a second locus for Brugada syndrome was found on chromosome 3; the responsible gene has not yet been identified. Further locus heterogeneity may exist, but linkage studies are difficult for the following reasons:

- Brugada syndrome causes sudden death in young individuals who may not have reproduced, reducing family size and thus limiting the power of linkage studies.
- Clinical diagnostic tools are limited in their ability to identify affected individuals.
- The effects of *SCN5A* mutations range from ST elevation in precordial leads to other alterations in the ECG that are prevalent in the general population (e.g., conduction disease) (see Genetically Related Disorders).

Clinical testing

- **Mutation scanning/sequencing of entire coding region.** Mutations in *SCN5A* have been identified in approximately 20%-25% of individuals with Brugada syndrome. Priori et al (2002) identified mutations in 22% (28/130) of consecutively screened probands. Schulze-Bahr et al (2003) determined that 38% of individuals with Brugada syndrome and a positive family history had identifiable mutations, whereas no individuals with Brugada syndrome and a negative family history had identifiable mutations.

Table 1 summarizes molecular genetic testing for *SCN5A*-related disorders.

Table 1. Molecular Genetic Testing Used in *SCN5A*-Related Disorders

Test Method	Mutations Detected	Mutation Detection Frequency [1, 2]	Test Availability
Mutation scanning/sequence analysis	*SCN5A* sequence variants	20%-25%	Clinical Testing

1. Proportion of affected individuals with a mutation(s) as classified by test method
2. Wilde et al 2002

Interpretation of test results. For issues to consider in interpretation of sequence analysis results, click here.

Testing Strategy

Confirmation of the diagnosis in a proband. In afpproximately 75% of cases the diagnosis can be confirmed without molecular genetic testing.

Predictive testing for at-risk asymptomatic adult family members requires prior identification of the disease-causing mutation in the family.

Genetically Related (Allelic) Disorders

Mutations in *SCN5A* are also responsible for the following:

- **Long QT syndrome 3 (LQT3)** (see Romano-Ward syndrome [RWS]). RWS is characterized by QT prolongation and T-wave abnormalities on ECG; these abnormalities are associated with tachyarrhythmias, including the ventricular tachycardia torsade de pointes (TdP), which may cause syncope or degenerate into ventricular fibrillation, resulting in aborted cardiac arrest (if the patient is defibrillated) or sudden death. In several reported families, some relatives had a LQT syndrome phenotype and others had a Brugada syndrome phenotype, supporting the concept that the two disorders are part of a spectrum of "sodium channelopathies" [Bezzina et al 1999; Priori, Napolitano, Schwartz et al 2000; Veldkamp et al 2000; Grant et al 2002].
- **Progressive conduction system disease (PCCD, Lenegre disease, isolated cardiac conduction disease).** PCCD manifests as slowed intramyocardial conduction and, in some cases, progressive atrioventricular (AV) block from first-degree to complete AV block [Schott et al 1999, Tan et al 2001, Wang et al 2002].

Clinical Description

Natural History

Individuals with Brugada syndrome have a high risk of ventricular arrhythmias and sudden death. Males in whom sustained ventricular arrhythmias are easily induced and who have a spontaneously abnormal ECG have a poor prognosis (i.e., a 45% likelihood of having an arrhythmic event at any time during life).

Brugada syndrome manifests primarily during adulthood, with a mean age of sudden death of approximately 40 years. Currently, the most common presentation is that of a man in his 40s with malignant arrhythmias and a previous history of syncopal episodes. Syncope is a common presenting symptom [Mills et al 2005, Benito & Brugada 2006, Karaca & Dinckal 2006].

The youngest individual diagnosed with the syndrome was two days old and the oldest age 85 years [Huang & Marcus 2004].

Although Brugada syndrome is more prevalent among affected males, the disease may affect both males and females. Females with Brugada syndrome are not spared from the disease or from sudden death [Hong, Brugada et al 2004].

Clinical presentations may also include sudden infant death syndrome (SIDS) (death of a child during the first year of life without an identifiable cause) [Priori, Napolitano, Giordano et al 2000; Antzelevitch 2001; Skinner et al 2005] and sudden unexpected nocturnal death syndrome (SUNDS) [Vatta et al 2002], a syndrome seen in Southeast Asia in which young persons die from cardiac arrest with no identifiable cause. The same mutation in *SCN5A* was identified in individuals with Brugada syndrome and SUNDS, thus supporting the hypothesis that they are the same disease [Hong, Berruezo-Sanchez et al 2004].

Brugada syndrome can overlap with conduction disease. The presence of first-degree AV block, intraventricular conduction delay, right bundle branch block, and sick sinus syndrome in Brugada syndrome is not unusual [Smits et al 2005].

Several parameters have been investigated to improve risk stratification. The only parameter currently used for clinical decision-making is inducibility during electrophyisiologic study (EPS). EPS inducibility in the asymptomatic individual is highly predictive of subsequent

events; however, the data are controversial and several groups do not use EPS for risk stratification; thus, decisions regarding when to implant a defibrillator vary widely among physicians and investigators [Eckardt et al 2005, Glatter et al 2005, Ikeda et al 2005, Al-Khatib 2006, Delise et al 2006, Gehi et al 2006, Imaki et al 2006, Ito et al 2006, Ott & Marcus 2006, Tatsumi et al 2006].

Genotype-Phenotype Correlations

Few studies have investigated genotype-phenotype correlations.

- The *SCN5A* mutations that cause LQT3 are associated with a gain of function, in contrast with the loss of function associated with Brugada syndrome and progressive conduction system disease. However, mutations that are associated with both diseases in the same family have been described.
- Within Brugada syndrome, the only data available indicate that individuals with Brugada syndrome who have an identifiable *SCN5A* mutation have a longer PR interval [Smits et al 2002] and may experience more bradyarrythmias [Makiyama et al 2005] than individuals with Brugada syndrome who do not have an identifiable *SCN5A* mutation.

Penetrance

Penetrance in Brugada syndrome is approximately 30% (i.e., ~30% of individuals with an *SCN5A* mutation present with an ECG diagnostic of Brugada syndrome). Using sodium channel blockers, approximately 80% of individuals with an *SCN5A* mutation manifest the characteristic ECG changes [Hong, Brugada et al 2004].

Nomenclature

Vatta et al (2002) and Hong, Berruezo-Sanchez et al (2004) determined that SUNDS and Brugada syndrome are phenotypically, genetically, and functionally the same disorder. SUNDS was originally described in individuals from Southeast Asia. Other names for SUNDS include sudden and unexpected death syndrome (SUDS), bangungut (Philippines), non-lai tai (Laos), lai-tai (Thailand), and pokkuri (Japan).

Prevalence

Brugada syndrome was identified relatively recently; thus, it is difficult to determine its prevalence and population distribution. Because the ECG is dynamic and often concealed, it is difficult to estimate the true incidence of Brugada syndrome in the general population.

Data suggest that Brugada syndrome occurs worldwide. The prevalence of the disease in endemic areas is on the order of 1:2,000 inhabitants. In countries in Southeast Asia in which SUNDS is endemic, it is the second cause (following accidents) of death of men under age 40 years.

Data from published studies indicate that Brugada syndrome is responsible for 4%-12% of unexpected sudden deaths and for up to 20% of all sudden death in individuals with an apparently normal heart.

As recognition of Brugada syndrome increases in the future, a sizeable increase in the number of identified cases could be expected. A prospective study of an adult Japanese population (22,027 individuals) showed 12 individuals (prevalence of 0.05%) with ECGs compatible with Brugada syndrome. A second study of adults in Awa (Japan) showed a prevalence of 0.6% (66 individuals out of 10,420). However, a third study in Japanese children showed only a 0.0006% (1:163,110) prevalence of ECGs compatible with Brugada syndrome [Hata et al 1997].

Therefore in the absence of symptoms and/or molecular genetic testing of *SCN5A*, these studies provide an estimate of the prevalence of the Brugada ECG pattern (and not Brugada syndrome) in the population studied. The results suggest that Brugada syndrome manifests primarily during adulthood, a finding in concordance with the mean age of sudden death (35 to 40 years).

Differential Diagnosis

For current information on availability of genetic testing for disorders included in this section, see GeneTests Laboratory Directory. —ED.

Other conditions that can be associated with ST-segment elevation in right precordial leads include the following (adapted from Wilde et al 2002 with permission).

Abnormalities that can lead to ST-segment elevation in the right precordial leads

- Right or left bundle-branch block, left ventricular hypertrophy
- Acute myocardial ischemia or infarction
- Acute myocarditis
- Hypothermia, causing Osborn wave in ECGs and sometimes resembling Brugada syndrome
- Right ventricular ischemia or infarction
- Dissecting aortic aneurysm
- Acute pulmonary thromboemboli
- Various central and autonomic nervous system abnormalities
- Heterocyclic antidepressant overdose
- Duchenne muscular dystrophy
- Friedreich ataxia
- Thiamine deficiency
- Hypercalcemia
- Hyperkalemia
- Cocaine intoxication
- Mediastinal tumor compressing the right ventricular outflow tract (RVOT)
- Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C)

Other conditions that can lead to ST-segment elevation in the right precordial leads

- Early repolarization syndrome
- Other normal variants (particularly in males)

Most of the above conditions can give rise to a type 1 ECG, whereas ARVD/C and Brugada syndrome can give rise to type 2 and type 3 ECGs. Therefore, it is important to distinguish between these two disorders.

Brugada syndrome should always be considered in the differential diagnosis of sudden cardiac death and syncope in persons with a structurally normal heart. The implications of establishing the diagnosis of Brugada syndrome in a symptomatic person are limited as placement of a defibrillator is the next step in management; however, determining which at-risk family

members have Brugada syndrome is essential to their medical care. Asymptomatic first-degree relatives should be evaluated with an ECG. Further studies (EPS, ajmaline test) should be considered based on clinical findings and ECG results.

Management

Evaluations Following Initial Diagnosis

To establish the extent of disease in an individual diagnosed with Brugada syndrome, the following evaluations are recommended:

- Electrocardiogram
- Induction with sodium blockers in patients with type 2 ECG or type 3 ECG and suspicion of the disease
- Electrophysiologic study to assess risk of sudden cardiac death. The data are controversial. However, in asymptomatic individuals, no other risk stratification parameter is presently available.

Treatment of Manifestations

Brugada syndrome is characterized by the presence of ST-segment elevation in leads V_1 to V_3. Implantable cardioverter defibrillators (ICDs) are the only therapy currently known to be effective in patients with syncope or cardiac arrest [Brugada et al 1999, Wilde et al 2002].

Electrical storms respond well to infusion of isoproterenol (1-3 µg/min), the first line of therapy before other antiarrhythmics [Maury et al 2004].

Controversy exists regarding the treatment of asymptomatic individuals. Recommendations vary:

- Observation until the first symptom develops (the first symptom can also be sudden cardiac death)
- ICD if the family history is positive for sudden cardiac death
- Use of electrophysiologic study to determine which individuals should have an ICD

Prevention of Primary Manifestations

Quinidine (1-2 g daily) has been shown to be beneficial in preventing symptoms and resolving ECG features [Belhassen et al 2004, Hermida el at 2004, Probst et al 2006].

Prevention of Secondary Complications

Persons with Brugada syndrome should avoid high fever, anesthetics, antidepressant drugs, and antipsychotic drugs with sodium-blocking effects.

During surgery and in the postsurgical recovery period persons with Brugada syndrome should be electrocardiographically monitored.

Surveillance

At-risk individuals with a family history of Brugada syndrome should undergo electrocardiographic monitoring every one to two years [Oe et al 2005]. The presence of type I ECG changes should be further investigated.

Agents/Circumstances to Avoid

The following can unmask the Brugada ECG [Antzelevitch et al 2002]:

- Febrile state
- Vagotonic agents
- α-adrenergic agonists [Miyazaki et al 1996]
- β-adrenergic antagonists
- Tricyclic antidepressants
- First-generation antihistaminics (dimenhydrinate)
- Cocaine toxicity

The following should be avoided [Antzelevitch et al 2003]:

- Class 1C antiarrhythmic drugs including flecainide and propafenone
- Class 1A agents including procainamide and disopyramide

Testing of Relatives at Risk

If the disease-causing mutation has been identified in the proband, molecular genetic testing of at-risk relatives is appropriate because of the insensitivity of ECG changes in establishing the diagnosis [Priori et al 2003]. Identification by ECG and/or molecular genetic testing of individuals at risk allows preventive measures such as avoidance of medications that can induce ventricular arrhythmias and closer follow-up (e.g., annual ECG).

See Genetic Counseling for issues related to testing of at-risk relatives for genetic counseling purposes.

Therapies Under Investigation

Search ClinicalTrials.gov for access to information on clinical studies for a wide range of diseases and conditions. Note: There may not be clinical trials for this disorder.

Other

Genetics clinics are a source of information for individuals and families regarding the natural history, treatment, mode of inheritance, and genetic risks to other family members as well as information about available consumer-oriented resources. See the GeneTests Clinic Directory.

Support groups have been established for individuals and families to provide information, support, and contact with other affected individuals. The Resources section (below) may include disease-specific and/or umbrella support organizations.

Genetic Counseling

Genetic counseling is the process of providing individuals and families with information on the nature, inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. The following section deals with genetic risk assessment and the use of family history and genetic testing to clarify genetic status for family members. This section is not meant to address all personal, cultural, or ethical issues that individuals may face or to substitute for consultation with a genetics professional. To find a genetics or prenatal diagnosis clinic, see the GeneTests Clinic Directory.

Mode of Inheritance

Brugada syndrome is inherited in an autosomal dominant manner.

Risk to Family Members

Parents of a proband

- Most individuals diagnosed with Brugada syndrome have inherited the disease-causing mutation from a parent.
- A proband with Brugada syndrome may have the disorder as the result of a *de novo* gene mutation. The proportion of cases caused by *de novo* mutations is very low (~1%).
- Recommendations for the evaluation of parents of a proband with an apparent *de novo* mutation include electrocardiographic analysis, attention to a family history of sudden death, and (if the mutation in the proband has been identified) molecular genetic testing.

Note: Although most individuals diagnosed with Brugada syndrome have inherited the mutation from a parent, the family history may appear to be negative because of failure to recognize the disorder in family members, decreased penetrance, early death of the parent before the onset of symptoms, or late onset of the symptoms in the affected parent.

Sibs of a proband

- The risk to the sibs of the proband depends on the genetic status of the proband's parents.
- If a parent of the proband is affected or has an *SCN5A* mutation, the risk to the sibs of inheriting the mutation is 50%.
- If a disease-causing mutation cannot be detected in the DNA of either parent, two possible explanations are germline mosaicism in a parent or a *de novo* mutation in the proband. To date, *de novo* mutations or germline mosaicism have not been described in Brugada syndrome.

Offspring of a proband. Each child of an individual with Brugada syndrome has a 50% chance of inheriting the mutation.

Other family members of a proband. The risk to other family members depends on the status of the proband's parents. If a parent is affected and/or has an *SCN5A* mutation, his or her family members are at risk.

Related Genetic Counseling Issues

See Testing of Relatives at Risk for information on testing at-risk relatives for the purpose of early diagnosis and treatment.

Considerations in families with an apparent *de novo* mutation. When neither parent of a proband with an autosomal dominant condition has the disease-causing mutation or clinical evidence of the disorder, it is likely that the proband has a *de novo* mutation. However, possible non-medical explanations including alternate paternity or undisclosed adoption could also be explored.

Family planning. The optimal time for determination of genetic risk is before pregnancy. Similarly, decisions about testing to determine the genetic status of at-risk asymptomatic family members are best made before pregnancy. It is appropriate to offer genetic counseling

(including discussion of potential risks to offspring and reproductive options) to young adults who have an SCN5A mutation or who are at risk of having one.

DNA banking. DNA banking is the storage of DNA (typically extracted from white blood cells) for possible future use. Because it is likely that testing methodology and our understanding of genes, mutations, and diseases will improve in the future, consideration should be given to banking DNA of affected individuals. DNA banking is particularly relevant in situations in which the sensitivity of currently available testing is less than 100%. See Testing for a list of laboratories offering DNA banking.

Prenatal Testing

No laboratories offering molecular genetic testing for prenatal diagnosis of Brugada syndrome are listed in the GeneTests Laboratory Directory. However, prenatal testing may be available for families in which the disease-causing mutations have been identified. For laboratories offering custom prenatal testing, see Testing.

Preimplantation genetic diagnosis (PGD) may be available for families in which the disease-causing mutations have been identified. For laboratories offering PGD, see Testing.

Molecular Genetics

Information in the Molecular Genetics tables is current as of initial posting or most recent update. —ED.

Table A. Molecular Genetics of Brugada Syndrome

Gene Symbol	Chromosomal Locus	Protein Name
SCN5A	3p21	Sodium channel protein type 5 subunit alpha

Data are compiled from the following standard references: Gene symbol from HUGO; chromosomal locus, locus name, critical region, complementation group from OMIM; protein name from Swiss-Prot.

Table B. OMIM Entries for Brugada Syndrome

600163	SODIUM CHANNEL, VOLTAGE-GATED, TYPE V, ALPHA SUBUNIT; SCN5A
601144	BRUGADA SYNDROME

Table C. Genomic Databases for Brugada Syndrome

Gene Symbol	Locus Specific	Entrez Gene	HGMD
SCN5A	SCN5A	6331 (MIM No. 600163)	SCN5A

For a description of the genomic databases listed, click here.

Note: HGMD requires registration.

Molecular Genetic Pathogenesis

The relationship between *SCN5A* and Brugada syndrome was identified in 1998. *SCN5A*, the α-subunit of the cardiac sodium channel gene, is responsible for the phase 0 of the cardiac action potential. The identification of *SCN5A* disease-causing mutations and the decrease in availability of sodium current suggest that a shift in the ionic balance in favor of a larger transient outward current (I_{to}) during phase 1 of the action potential causes the disease.

Normal allelic variants: The genomic sequence encompasses more than 100 kb, and the gene contains 28 exons.

Pathologic allelic variants: More than 100 different mutations in *SCN5A* have been reported to date [Moric et al 2003, Tan et al 2003]; approximately half of them have been biophysically characterized. Several different mutations affecting the structure, function, and trafficking of the sodium channel have been identified.

Normal gene product: *SCN5A* encodes the α-subunit of the cardiac sodium channel gene. The protein contains four internal repeats, each with five hydrophobic segments (S1, S2, S3, S5, S6) and one positively charged segment (S4). S4 segments are probably the voltage sensors and are characterized by a series of positively charged amino acids at every third position (adapted from Human Genome Browser). This integral membrane protein mediates the voltage-dependent sodium ion permeability of excitable membranes. Assuming opened or closed conformations in response to the voltage difference across the membrane, the protein forms a sodium-selective channel through which Na+ ions may pass in accordance with their electrochemical gradient. It is a tetrodotoxin-resistant Na+ channel isoform. The channel is responsible for the initial upstroke of the action potential in the ECG. The protein is expressed in human atrial and ventricular cardiac muscle but not in adult skeletal muscle, brain, myometrium, liver, or spleen.

Abnormal gene product: The common feature is the decrease in Na+ current availability by two main mechanisms: lack of expression of the mutant channel or accelerated inactivation of the channel.

Resources

GeneReviews provides information about selected national organizations and resources for the benefit of the reader. GeneReviews is not responsible for information provided by other organizations. Information that appears in the Resources section of a GeneReview is current as of initial posting or most recent update of the GeneReview. Search GeneTests*for this disorder and select* Resources *for the most up-to-date Resources information.*—ED.

National Library of Medicine Genetics Home Reference
Brugada syndrome

Ramon Brugada Sr Foundation
Email: foundation@brugada.org
www.brugada.org

Canadian SADS Foundation
15-6400 Millcreek Drive Suite 314
Mississauga L5N 3E7
Canada
Phone: 877-525-5995; 905-826-6303
Fax: 905-826-9068
www.sads.ca

Cardiac Arrhythmias Research and Education Foundation (CARE)
26425 NE Allen Street Suite 103
P.O. Box 369
Duvall WA 98019
Phone: 800-404-9500; 425-788-1987
Fax: 425-788-1927
Email: care@longqt.org
www.longqt.org

SADS Australia
Email: info@sads.org.au
www.sads.org.au

SADS UK
www.sadsuk.org

Sudden Arrhythmia Death Syndromes (SADS) Foundation
508 East South Temple Suite 20
PO Box 58765
Salt Lake City UT 84102
Phone: 800-786-7723; 801-531-0937
Fax: 801-531-0945
Email: sads@sads.org
www.sads.org

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Medical Genetic Searches: A specialized PubMed search designed for clinicians that is located on the PubMed Clinical Queries page. PubMed

Published Statements and Policies Regarding Genetic Testing

No specific guidelines regarding genetic testing for this disorder have been developed.

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Suggested Readings

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Chapter Notes

Author Notes

Ramon Brugada, MD

Clinical interest. As a clinical and noninvasive cardiologist, Dr. Brugada is interested in the management of patients with inherited disorders of the heart.

Research interest. Dr. Brugada's research interests are focused on molecular genetics of cardiovascular disease with an emphasis on genetics of cardiac arrhythmias. His research achievements include the identification of the chromosomal locus on 10q22 for familial atrial fibrillation, the gene for familial idiopathic ventricular fibrillation (Brugada syndrome), and the gene for short QT syndrome.

Honors include the American College of Cardiology Young Investigator Award (1st Prize), the Merck-ACC Adult Cardiology Research Fellowship Award, the Fritz-Acker Award of the German Society of Cardiology, the 3rd Mirowski Award, and the Josep Trueta Award to Medical research of the Academy of Sciences of Catalonia. In 2000, he received the Doris Duke Clinical Scientist Development Award and the Fourjay Foundation Award to continue his research project on familial atrial fibrillation. He has received a Scientist Development grant from the AHA and he is the PI on an RO1 grant from the NIH to continue his projects. In 2005 he moved to the Montreal Heart Institute/University of Montreal as Associate Professor, Cardiologist, Research Scientist and Director of the Clinical Cardiovascular Genetics Center. He is the recipient of a Canadian Research Chair in Genetics of Cardiovascular Electrophysiology.

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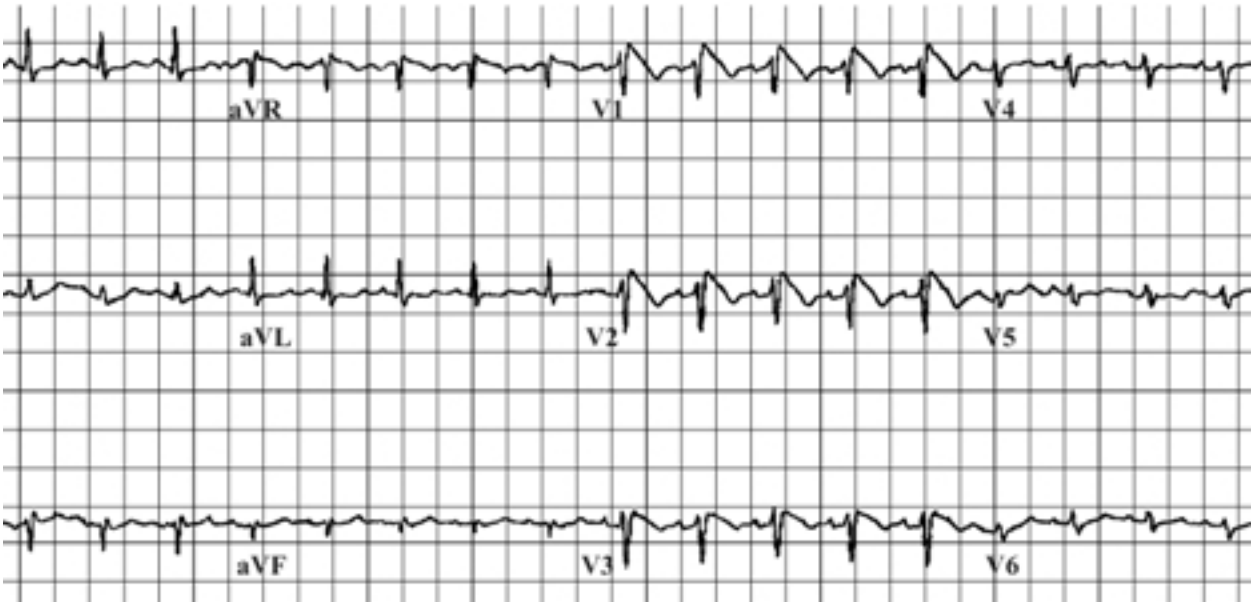

Figure 1. Characteristic ECG in Brugada syndrome. Note presence of ST-segment elevation in leads V_1-V_3, coved type.