Clinical Quiz

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A family's name for a familial disease

Clinical Presentation

A 25-year-old male with electrocardiographic (ECG) changes (**Figure 1**). His family history includes death at an early age of male relatives (16 and 17 years) (**Figure 2**), and another 2 had implantable defibrillator fitted for syncopal attacks.

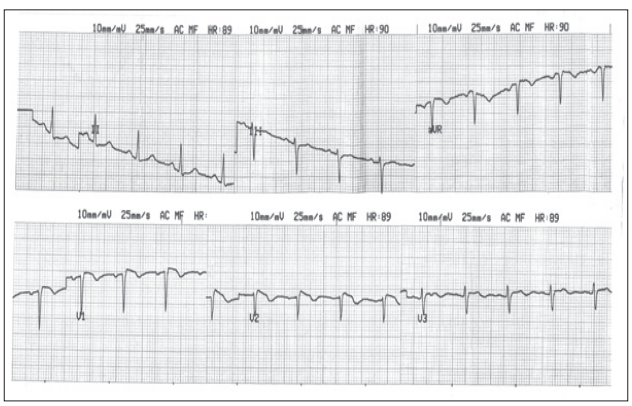

Figure 1 - Note the precordial leads recording V1 - V3.

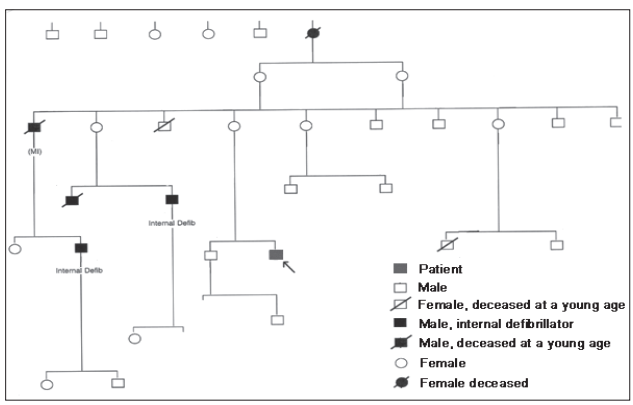

Figure 2 - Family pedigree.

Questions

1. Which is the syndrome he was investigated for?
2. Are you aware of its type of inheritance?
3. What ECG changes will make you think of the syndrome in patients presenting with syncopal attacks?
4. What is the management modality of choice?

Clinical Quiz

Answers

1. Brugada syndrome.
2. Familial, displaying autosomal dominant inheritance with incomplete penetrance
3. ST-segment elevation in the right precordial leads followed by negative T-waves or rapid polymorphic ventricular tachycardia (VT), unrelated to ischemia and structural heart disease.
4. Implantable cardioverter-defibrillators.

Discussion

Brugada syndrome (BS) is a familial syndrome, which displays autosomal dominant inheritance with incomplete penetrance, and the incidence ranges between 5 and 66/10,000.[1] It is now recognized with increased frequency worldwide, and its highest prevalence appears in southeast Asia.[2] It has a male predominance (male:female ratio 8:1), and arrhythmic events usually appear. It is a relatively new disease. In 1986, the first case referred to the Brugada brothers was a 3-year-old boy with recurrent episodes of syncope.[2] His ECG showed elevations of the ST-segment in the right precordial leads (V1 - V3). His sister, having also displayed syncopal attacks and ECG abnormalities, had died at the age of 2. It only became known as BS in the early nineties, and immediately received international attention because of its relation with sudden cardiac death (SCD). The typical ECG abnormalities of BS are: 1. Attenuated J-wave mostly in the precordial leads (V1 - V3), which takes the form of ST-segment elevation and is often followed by negative T-wave; 2. Very closely coupled extrasystoles; 3. Rapid polymorphic VT (at times very similar to ventricular fibrillation), all these changes must be unrelated to ischemia, electrolytic imbalances or structural heart disease;[3] 4. History of SCD of relative (age <45 years); 5. Syncopal episodes or nocturnal agonal respiration, or both.[1] Of note, 3 out of the 6 male patients, presented as case-series by the Brugadas, had prolonged QTc.[1,4] The genetic research has identified one gene (SCN5A), encoding for the α-subunit of the sodium channel, as responsible for the BS.[1] Therefore, it seems logical that medications with potent sodium channel-blocking activity such as flecainide and procainamide, can be used in drug challenge tests to elicit the ECG phenotype of BS in patients with intermittently normal ECG, but with appropriate clues from the patient's history. Obviously, due to the increased risk of precipitating ventricular arrhythmia, challenge tests should always be performed with the patient under continuous monitoring and resuscitation facilities at hand distance.[1]

The differential diagnosis of BS is extensive, and includes mostly diseases and factors that can lead to ST-segment elevation in the right precordial leads. With these, it is worth mentioning the arrhythmogenic right ventricular cardiomyopathy, which can present with the same ECG phenotype as BS, and its structural heart abnormalities may only be found at the time of the autopsy.[1] Implantable cardioverter-defibrillators, together with a high level of clinical suspicion is necessary for the diagnosis, and still represents as the only effective mode of dealing with BS.[3]

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References

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