

Case report: Brugada syndrome and the importance of family screening

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Introduction: Brugada syndrome is a genetic disorder characterized by abnormal electrocardiogram (ECG) findings and an increased risk of sudden cardiac death, particularly in young adults. The condition is most often linked to mutations in the SCN5A gene, which affect sodium channels in the heart. Clinical manifestations can range from asymptomatic cases to life-threatening arrhythmias. Approximately 20-30% of cases are linked to genetic mutations, and it predominantly affects males.¹

Case report: This case describes a 45-year-old male patient diagnosed with Brugada syndrome, confirmed via ECG after episodes of ventricular tachycardia (VT) in 2017. Despite normal echocardiograms and coronary angiography, the patient exhibited a characteristic ST-segment elevation in leads V1-V2. Given the high risk of sudden cardiac death, he underwent the implantation of an implantable cardioverter-defibrillator (ICD) in March 2018. The procedure was successful, with no complications, and proper positioning of the ICD lead was confirmed with chest X-ray. Following the ICD implantation, the patient was scheduled for follow-up in three months and was given instructions on wound care and activity restrictions. He was advised to avoid specific medications and situations known to trigger arrhythmias, in line with current management guidelines for Brugada syndrome. In terms of family screening, ajmaline testing was arranged for his two children, his brother, and his brother's two sons. All tested negative except for his son, who showed borderline results, indicating the need for continuous monitoring. Genetic testing may have also been performed to further assess the risk of Brugada syndrome within the family.

Conclusion: This case underscores the importance of ICD implantation in patients at high risk of sudden cardiac death and highlights the value of genetic testing and family screening to identify and manage potentially affected relatives.

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LITERATURE

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