

CASE STUDY

Brugada Syndrome: Killer Genetic Heart Disorder

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ABSTRACT

Brugada syndrome is a rare inherited arrhythmic disorder, which predisposes to ventricular arrhythmia that is responsible for sudden and unexpected nocturnal death syndrome.

Keywords: Brugada syndrome, Sudden and unexpected nocturnal death syndrome, Ventricular arrhythmia.

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INTRODUCTION

Brugada syndrome is a hereditary arrhythmic disorder (autosomal dominant inheritance), caused by mutation in the SCN5A gene. It is a cardiac sodium channel abnormality that predisposes to ventricular arrhythmia and responsible for sudden cardiac death. Prevalence is approximately 0.15% in adults and 0.005% in children in Asia and less than 0.02% in the West. Its incidence in Japan is high, i.e., 14.2 per 100,000 person per year. It is identified in both genders and all races, but is most common in young male patients (75%). It remains life-long asymptomatic in individuals, but sudden cardiac death may occur as early as the first year of life. Fever is the most common precipitating factor for arrhythmic cardiac events. In patient with structurally and functionally normal heart, electrocardiogram (ECG) appears like right bundle branch block with ST segment elevation in V1–V3.

CASE REPORT

A young 40-year-old male presented in emergency with history of palpitation and chest heaviness for 2 to 3 hours

with fainting attack and mild fever without chills and shivering for 1 day. No history of perspiration, breathlessness, and cough was present.

He gave a history of several on–off fainting attacks in the past 12 years, and there was no definite interval for these symptoms. He had been treated as a case of myocardial infarction/unstable angina on clinical basis without relief of symptoms in past. No previous ECG was available. He also gave a history of his younger brother's death in the night due to unexplained cause at the age of 30 years. However, his father is asymptomatic.

The patient was conscious, oriented with pulse 78 beats/minute, regular, normovolumic, normal in character, synchronous, arterial wall not palpable, and with no radiofemoral delay. Blood pressure was 124/84 mm Hg in the right arm supine position. Temperature was 100°F at the time of admission. Respiratory rate was 20/minute, regular, and abdominothoracic. The peripheral capillary oxygen saturation (SpO₂) was 98%. All systemic examinations were clinically within limits. Hemogram, electrolytes, and chest X-ray were normal. The ECG showed saddleback ST segment elevation in V1–V2 (Fig. 1). Cardiac biomarkers were negative 6 hours postadmission. No functional or structural abnormality was detected in two-dimensional echocardiography. Therefore, ECG changes gave us clues to make a diagnosis of Brugada syndrome and was thought to be responsible for sudden and unexpected nocturnal death syndrome of patient on the same night of admission.

DISCUSSION

Brugada syndrome constitutes a deadly threat that often remains latent for many years, only to manifest itself in a lethal arrhythmia in persons considered to be otherwise healthy. Main reason for delayed diagnosis of Brugada syndrome is the periodic normalization of electrocardiographic features of the syndrome,¹ which is related to incomplete penetration of the gene responsible for the syndrome. Moreover, changes in ECG may be subject to influences of factors, such as body temperature² or autonomic system tone.^{3,4} It has an autosomal dominant pattern of inheritance with incomplete penetrance. Yearly screening of ECG and genetic testing is recommended for family members. Implantable cardiac defibrillator is

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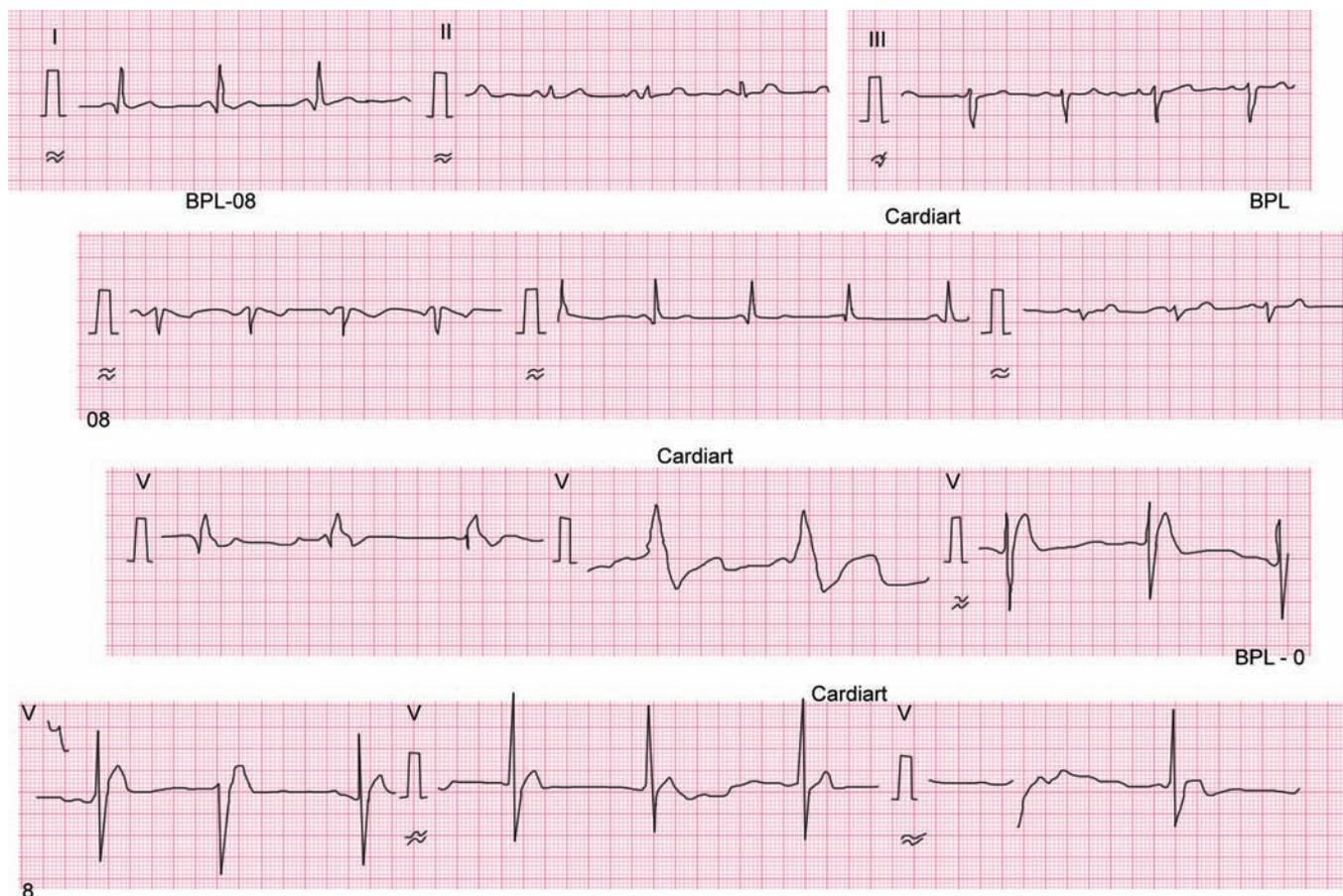


Fig. 1: Electrocardiogram with saddleback ST segment elevation in V1–V2

the choice of treatment and should be advised for those having ECG changes.

CONCLUSION

All ST elevations are not due to myocardial infarction and may have different etiology with risk of sudden cardiac death.

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