Case Study

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A Case Report of Type 1 Brugada Syndrome with Scn5a Mutation Augmented by Fever

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Abstract

Brugada syndrome is an inherited disorder characterized by a channelopathy of cardiac sodium, potassium, and calcium channel. The pathophysiology of this disorder is not completely elucidated yet, however, most of the reported cases are caused by a pathogenic alteration in the SCN5A gene, leading to the malfunction of cardiac sodium channels. Three ECG patterns are frequently described in the literature, type 1, type 2, and type 3. However, only the type 1 pattern is considered diagnostic of Brugada syndrome in the appropriate clinical context. Therapeutic strategies can range from conservative medical management with antiarrhythmic medications to Implantable Cardioverter Defibrillator placement. Prompt recognition is of utmost importance since this pathology can rapidly evolve into life-threatening arrhythmias and sudden cardiac death. Here we present a case of a 20-year-old male who presented after a surgery and with clinical findings of Brugada syndrome in which the patient suffered from brain damage post-resuscitation due to cardiac arrest.

Keywords: Brugada syndrome, SCN5A mutation, fever

Background

Brugada syndrome is an inherited arrhythmia caused by genetic mutations that disrupt the cardiac sodium channel which results in risk of ventricular fibrillation and subsequent sudden cardiac death (Kapplinger et al., 2010), (Priori et al., 2013). The prevelence of Brugada syndrome is higher in young male Southeast Asians who often have no prior structural heart abnormality and a mutation in the SCN5A gene can be found in approximately 15 – 30% of individuals affected by this syndrome (Al-Khatib et al., 2018), (Priori et al., 2015). The essential tool to discover and diagnose this disease is the ECG. The typical pattern on the ECG is coved-type ST elevation in V1 –V3 followed by a negative T wave (also known as type 1 Brugada pattern) (Kapplinger et al., 2010). Here we report a case with clinical findings of Brugada syndrome in which the patient suffered from brain damage post-resuscitation due to cardiac arrest. Through this case, we advocate early diagnosis and prompt management to prevent the aggravation of the patient's condition.

Case Summary

A 20 year-old male with no personal or family history presented with fever and abdominal pain. The patient was admitted to the surgical department of a hospital where he was diagnosed of appendicitis. An apendicectomia was performed a few hours later. After the surgery, his condition deteriorated rapidly. An urgent ECG was recorded and revealed that the patient had the type 1 Brugada pattern as seen below.

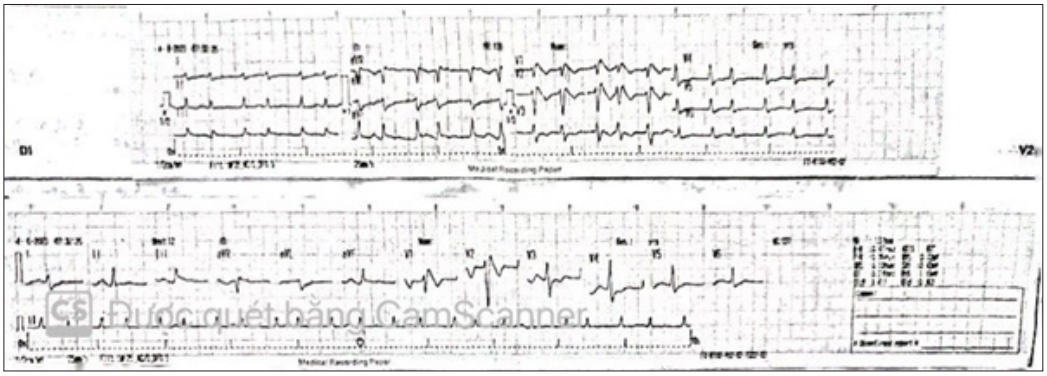

Figure 1: Electrocardiography after surgery

He then developed ventricular fibrillation and subsequent cardiac - respiratory arrest. The patient was successfully resuscitated although he was in coma and intubated. He later suffered from multi-organ failure and ventilator pneumonia. After being treated aggressively and underwent hemodialysis for 7 days, the patient was referred to a more advanced institution. There he was medically stablilized and weaned from mechanical ventilation. He was then transferred to our hospital for rehabilitation. We performed a cardiac ultrasound on the patient and no structural abnormality was found, and the Brugada pattern can no longer be seen on the ECG.

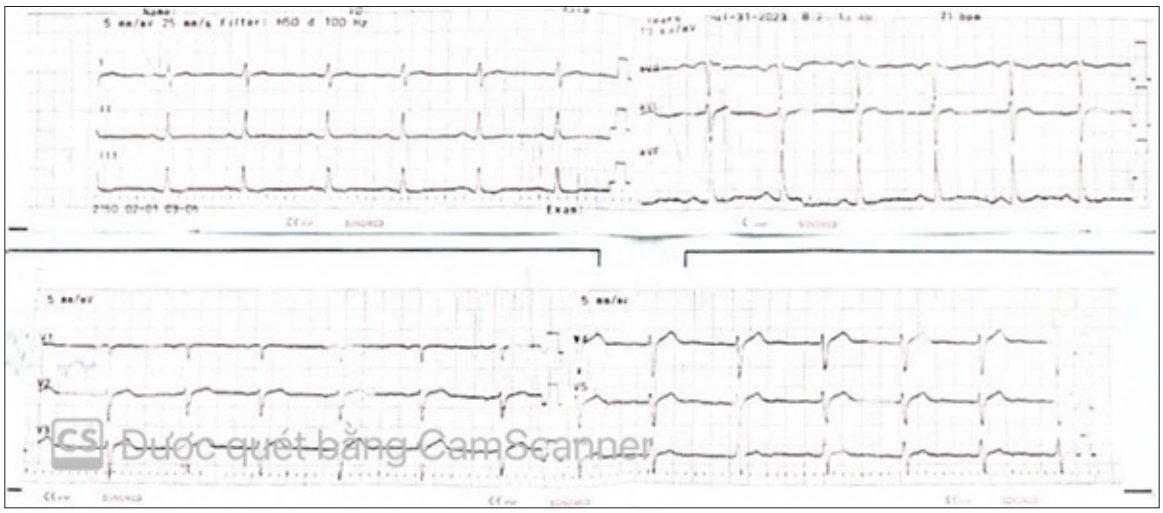

Figure 2: Electrocardiography after 7 days

After a long course of supportive treatment and rehabilitation, the patient was finally able to resume some basic life functions such as mouth feeding, defecating, urinating and walking with supportive equipments. A genetic analysis was indicated and the result showed that there is a non-clinvar heterozygous variant in SCN5A gene (autosomal dominant/autosomal recessive). Heterozygous mutations of this gene is consistent with Brugada syndrome, linking to the clinical features.

Discussion

As mentioned above, Brugada syndrome often affects young individuals with no history of heart disease, which makes it very challenging to discover this dangerous disease. If left undiagnosed, Brugada syndrome can cause devasating consequences as seen with the patient in the reported case. Therefore, the key point in preventing this is to regconize the ECG pattern to make the diagnosis early. The baseline ECG can be normal but the Brugada pattern can be umasked by some factors, particularly fever in this case report. Other factors include ischemia, drugs (sodium channel blockers, calcium channel blockers, beta blockers, nitrates, alpha agonists, cocaine, alcohol), hypo/hyperkalemia, hypothermia, etc. Moreover, the placement of the right precordial leads in the 3rd or 2nd intercostal spaces increases the sensitivity of discovering the Brugada pattern in some patients. Since patients present with wide range of complains and contexts, all physicians, escpecially those whose specialty not cardiology, should be able to regconize the ECG pattern so that no Brugada syndrome diagnosis is missed (Al-Khatib et al., 2018), (Priori et al., 2015).

Once the diagnosis is made, the only defenitive treatment is implantable cardioverter defibrillator (ICD). However, this method of treatment might be unsuitable for many patients due to high cost and co-morbidities. Therefore, the general management also plays a vital role in preventing the deterioration of the patient. This includes:

- Avoiding large meals and excessive alcohol intake
- Avoiding drugs that can cause Brugada syndrome ECG changes or are associated with arrhythmias
- Proper hydration and electrolyte replenishment
- Treating fever immediately with antipyretic medications
- Quinidne may reduce ventricular fibrillation and may be useful in some situations
- Isoproterenol may be useful in suppressing arrhythmic storms and preventing further episodes of arrhythmia
- Avoiding increased vagal tone perioperative with slow peritoneal insufflation and deflation
- Because Brugada syndrome is an inherited disease, ECG screening and genetic testing should also be conducted on the patient's family members (Priori et al., 2015).

Conclusion

This case is a typical example for Brugada syndrome, a rare yet lethal disease. Early diagnosis with ECG pattern and genetic testing on the patient and the patient's family members is crucial in preventing and mitigating the burden of this disease.

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