Hemochromatosis as junctional tachycardia, a rare presentation

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Abstract

We present here a 45-year-old male with no past medical problem who presented with palpitations. He was found to have supraventricular tachycardia intractable to medical therapy. Later his rhythm converted to junctional tachycardia. Further workup revealed hemochromatosis to be primary etiology causing the arrhythmia. The low index of suspicion for additional workup is key to diagnosis and successful outcome.

Introduction

The primary electrophysiological function of the atrioventricular (AV) node is conduction of cardiac impulse generated in the sino-atrial (SA node) area. It also functions as a secondary pacemaker for cardiac muscle and at rare occasions, it can take control over the normal sinus rhythm and act as a primary locus of cardiac impulse generation.[1]

Arrhythmias arising from the atrioventricular (AV) junction occur as automatic tachycardia or as an escape mechanism when sinus node is unable to work well.[2] Junctional tachycardias are usually manifested with digoxin toxicity, surgical involvement or manipulation of the AV-node, ischemia, and tumors involving the AV-node.[3]

Infiltrative diseases of myocardium causing junctional tachycardia has never been reported so far. We present here a case of hemochromatosis presenting as intractable supraventricular tachycardia (SVT) and a brief literature review of SVT.

Case Report

A 45-year-old male with no previously reported medical and surgical history presented to emergency room with shortness of breath and palpitations for 4 days. He felt better in between but his symptoms recurred a night before prompting him to seek medical attention. He had no fever, chest pain, dizziness, vision symptoms or a recent change in bowel or urinary habits. Family history was significant for coronary artery disease in father and uncle had a history of atrial fibrillation.

Initial Vitals were pertinent for blood pressure 139/92 mmHg, pulse 149 beats/min, respiratory rate of 20/min and temp 98.6F, O_2 saturation of 98% on room air. Electrocardiogram showed supraventricular tachycardia to 144 beats/min which initially converted to normal sinus rhythm after Cardizem and later reverted to SVT.

On physical examination, heart sounds were normal without murmur, gallop or a rub. Lungs were clear to auscultation without wheezing and there was no pedal edema. Laboratory work showed normal blood count, metabolic profile except AST of 67 U/L, normal electrolytes and TSH.

The patient was admitted for continuous telemetry, was started on intravenous Cardizem with Sotalol and plan to do an echocardiogram. Next morning, his heart rate did slow down but rhythm did not break even being on Sotalol and Cardizem drip overnight. Echocardiography showed mild concentric left ventricular hypertrophy with normal ejection fraction of 55-60%, impaired left ventricular relaxation and no regional wall motion abnormalities. Electrophysiology team was involved and the patient was given 12 mg of adenosine with subsequent slowing of rhythm showing underlying junctional tachycardia.

Diltiazem and sotalol were discontinued and the patient was started on flecainide. Blood work was ordered to rule out infiltrative diseases including iron studies and ACE level. Iron studies indicated serum iron of 161 mcg/dL, ferritin 973 ng/ml, TIBC 230 ug/dL and iron saturation of 70%.

Cardiac MRI was done which showed 2 to 3 mm focus of potential mid-myocardial enhancement in the interventricular septum and hepatic iron deposition with calculated iron concentration of 3.25 mg/gram corresponding to mild iron overload. Hematology/Oncology team was involved and the patient was advised phlebotomy for hemochromatosis and outpatient follow-up. Outpatient work up revealed hereditary hemochromatosis (HH) positive mutations for C282Y. Two copies of the C282Y mutation were identified. Results for H63D and S65C were negative.

Discussion

Infiltrative cardiomyopathies are characterized by the deposition of abnormal substances within the heart tissue and have a well-established association with heart failure and arrhythmias.[4] The deposition of the abnormal substances *e.g.*, amyloid and iron in the myocardial tissue initiate the inflammatory response which leads to the accumulation of fluid, cellular debris along with the substance causes an increased wall thickness.[5] Wall thickness does not essentially mean myocyte hypertrophy, typically in the case of infiltrative disease as some cause chamber enlargement with secondary wall thinning. Ultimately the wall thickness, myocyte hypertrophy and reduction in chamber volume lead to diastolic dysfunction.[4] The conduction pathway can also be affected by the infiltrative process leading to the rhythm disturbances. Both heart failure and arrhythmias stand as the leading cause of the death in infiltrative cardiomyopathies.[6]

One of the most common autosomal recessive genetic disorders of iron metabolism in white populations is Hereditary hemochromatosis (HH).[7] This leads to inappropriately high iron absorption. Three major missense mutations of the hemochromatosis gene (HFE) are C282Y, H63D, and S65C.[7] Hemochromatosis causes the deposition of iron within the myocytes and dilated left ventricle with global systolic dysfunction.[8] Typically, in the iron-overload state along with lower ejection fraction, the atrial and ventricular arrhythmias and heart blocks are common presentations. Symptoms range from shortness of breath, palpitations and fainting spells due to the sudden decrement of cardiac output. The average survival of patients with untreated hemochromatosis is less than a year. If treated appropriately, survival is comparable to the normal population.

The case presented is of sustained SVT in an iron-overloaded patient not responsive to therapeutic measures. The conversion of SVT into a junctional rhythm in absence of coronary artery disease, digoxin toxicity or adult age is very uncommon. SVT causing palpitations led to early diagnosis and timely intervention to pursue further diagnostic work up and management of our patient.

Incidence and prevalance of SVT

The incidence of SVT is around 35 cases per 100,000 persons per year, with the prevalence of about 2.25 per 1000 (excluding atrial fibrillation, atrial flutter, and multifocal atrial tachycardia).[9]

Classification of SVT: SVT is usually classified based on the mechanism of ventricular activation *i.e.* AVNRT (atrioventricular nodal reentrant tachycardia), AVRT (atrioventricular reciprocating tachycardia) or presence of an additional pathway. For AVNRT, usually, no p-wave is seen after a QRS complex. On the contrary, for narrow QRS complex AVRT, p wave after a QRS complex is seen.

Classification of AVNRT

A reentrant tachycardia involving 2 functionally distinct pathways is generally referred to as *fast* and *slow* pathways. Most commonly, the fast pathway is located near the apex of Koch's triangle, and the slow pathway inferoposterior to the compact AV node tissue. Variant pathways have been described, allowing for *slow-slow* AVNRT.[10] i) *Typical AVNRT:* AVNRT in which a slow pathway serves as the antero-grade limb of the circuit and the fast pathway serves as the retrograde limb (also called *slow-fast AVNRT*). ii) *Atypical AVNRT:* AVNRT in which the fast pathway serves as the anterograde limb of the circuit and a slow pathway serves as the retrograde limb (also called *fast-slow AV node reentry*) or a slow pathway serves as the anterograde limb and a second slow pathway serves as the retrograde limb (also called *slow slow AVNRT*).

Classification of AVRT

Atrioventricular reentrant tachycardia (AVRT): A reentrant tachycardia in which impulse travels reciprocally from ventricles to atria via an accessory pathway and causes atrial activation. i) *Orthodromic AVRT:* An AVRT in which the reentrant impulse uses the accessory pathway in the retrograde direction from the ventricle to the atrium, and the AV node in the anterograde direction. The QRS complex is generally narrow or may be wide because of pre-existing bundle-branch block or aberrant conduction. ii) *Antidromic AVRT:* An AVRT in which the reentrant impulse uses the accessory pathway in the anterograde direction from the atrium to the ventricle, and the AV node for the retrograde direction. Occasionally, instead of the AV node, another accessory pathway can be used in the retrograde direction, which is referred to as pre-excited AVRT. The QRS complex is wide (maximally pre-excited).

Junctional tachycardia

A non-reentrant SVT that arises from the AV junction (including the bundle of his).

Management of SVT's

No matter what the mechanism of arrhythmia is, every SVT is treated on a similar algorithm. The first step is to determine hemodynamic status. If the patient is hemodynamically unstable (*i.e.* hypotension, shortness of breath, chest pain, shock
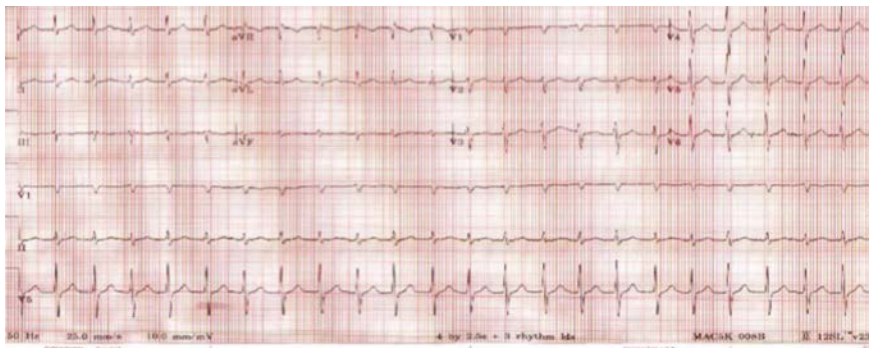

Figure 1. Initial tracing showing Narrow complex tachycardia.

Figure 2. Strip showing accelerated Junctional rhythm.

Figure 3. Junctional rhythm in beginning of EKG followed by normal sinus rhythm. EKG prior to discharge from hospital.
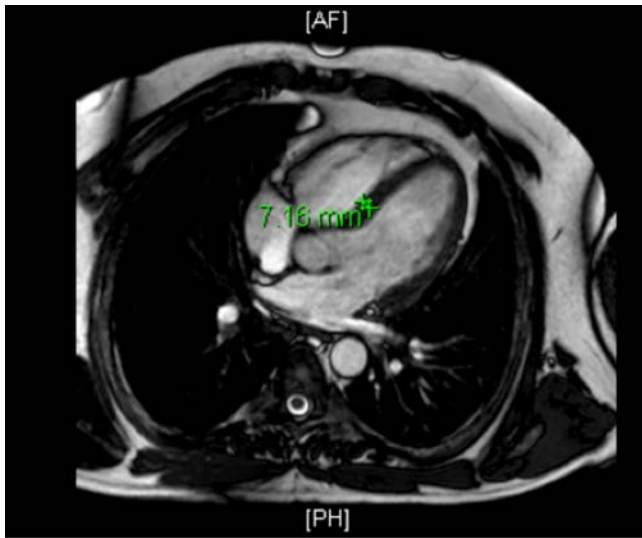
Figure 4. Cardiac MRI showing 2 to 3-mm focus of potential mid myocardial enhancement in the interventricular septum towards the base.
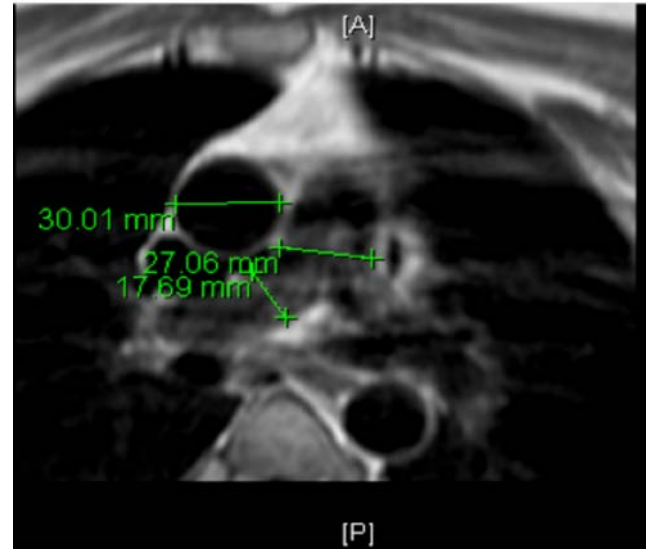

Figure 5. MRI evidence of iron deposition within the imaged liver with a calculated liver iron concentration of 3.25 mg/gram.

or decreased level of consciousness) then cardioversion is advised. If the patient is hemodynamically stable then try vagal maneuvers, especially if the patient is hemodynamically stable. The next best step is adenosine 6mg initially followed by a repeat dose of 12 mg. If an arrhythmia is not breaking, other AV nodal blockers like beta blockers and calcium channel blockers can be used. Anti-arrhythmic drugs like IV Procainamide, propafenone, flecainide and ibutilide are other alternatives if AV nodal blockers are not working.[11] However, if the arrhythmia is arising from pre-excitation from an accessory pathway, then use of AV node blockers can be harmful rather than of being use. They can exacerbate the syndrome by blocking the heart's normal electrical pathway and ultimately lead to degeneration into ventricular fibrillation and ultimately cardiac arrest.

Conclusions

It is very uncommon to have an SVT as the initial presentation of hemochromatosis. It may be slowed down with AV nodal blockers to a junctional tachycardia but the addition of antiarrhythmic may be required for conversion to normal sinus rhythm.

High degree of suspicion, early diagnosis and getting rid of excess iron is the key to the successful outcome.

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