CASE REPORT

A Mistake that has Hurt No One: Sinus Mistakus

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ABSTRACT

There are times when we, the health care providers make a diagnosis and plan to treat that condition accordingly. In the mean time, because of a second opinion or another specialist consult might change the diagnosis completely and therefore the mode of management could change drastically. Here we present a similar case scenario for work-up of chest pains changing the diagnosis and therefore the mode of treatment. However in this process the patient did not get hurt ("Sinus Mistakus").

Key words: Arrhythmogenic right ventricular dysplasia, Atrial septal defect, Abnormal ECG and Echocardiography

CASE REPORT

A 15-year-old teenage boy was seen in my office for cardiac consultation after having chest injury during his school sports. He was initially seen the emergency room locally for his evaluation and was sent home requesting him to come to my office for a cardiac consult. During wrestling, he was hurt physically to his chest and needed an urgent evaluation.

By the time he came to my office, his chest pains had resolved and his vitals were stable. There is no history of any heart disease in the family and no history of congenital heart disease.

His baseline resting electrocardiogram (ECG) was abnormal: Normal sinus rhythm, prominent R-waves in the right precordial leads suggestive of pulmonary hypertension or right ventricular (RV) hypertrophy Figure 1.

His two-dimensional (2D) echocardiographic study revealed: Preserved the left ventricular (LV) systolic function with LV ejection fraction of 60–65%, normal left atrial size, dilated right ventricle, and mildly dilated right atrium. No evidence of any thrombus and no significant valvular dysfunction noted. No evidence of pericardial effusion Figures 2 and 3.

Question

Why were his ECG and 2D-echocardiographic studies abnormal? Were these studies abnormal due to his recent chest injury or these abnormal findings were already there and the history of chest injury was simply a "distraction" in his diagnosis?

Based on these abnormal findings, a cardiac magnetic resonance imaging (MRI) was requested which reported following pertinent findings:

1. Severely increased RV volume with RV apical hypokinesis
2. Normal global LV and RV function
3. Increased RV volume and hypokinesis suggestive of arrhythmogenic RV cardiomyopathy (ARVC)
4. Genetic testing recommended.

Bigger Question

After all these tests, this teenager (wishes and) wants to know if he could continue to play the sports of his choice!

ARVC/DYSPLASIA (D)

Introduction

ARVC/D is characterized by progressive fibrofatty replacement of the RV myocardium. It represents an

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underdiagnosed cardiac condition leading to recurrent ventricular tachycardia, heart failure, syncope, and occasionally sudden cardiac death in younger people.[1] It is a genetic cardiomyopathy (mostly autosomal dominant) with an estimated prevalence of 1 in 1000 to 1 in 5000.[2] Familial occurrence can be as much as 50%, and it can account for 3%–4% of deaths in sports with sudden cardiac deaths in population younger than 65 being 5%.[3] It is the second most common cause of death after hypertrophic cardiomyopathy causing up to 20% of sudden cardiac deaths in patients under the age of 35 years. It is more common in men than women (3:1) and people of Italian and Greek descent. Some patients might have an autosomal recessive expression with what is called Naxos disease which is associated with woolly hair and skin changes.

Etiology

This condition is considered a part of idiopathic cardiomyopathies as per its nature of progressive disease with unclear pathogenesis and etiology. Basso *et al*.[4] proposed four hypotheses for a possible explanation of this disease status. The first hypothesis: Apoptosis that is programmed cell death leading to progressive myocardial muscle damage and loss followed by fibrofatty replacement which increases the electrical vulnerability of the right ventricle leading to life-threatening arrhythmias.[5] The second hypothesis: The dysontogenetic theory: This disease should be considered a congenital heart disease, thereby leading to the abnormal development of right ventricle and thus leading to dysplasia. The third hypothesis: The degenerative theory: A metabolic disorder may affect the right ventricle and result in the progressive replacement of myocardium by fat and fibrous tissue. The fourth hypothesis: The inflammatory theory: The fibrofatty replacement of the right ventricle is considered as a healing process in the setting of idiopathic myocarditis.[6]

Pathophysiology

In this condition, the most common location for fibrofatty replacement of the normal myocardium is between

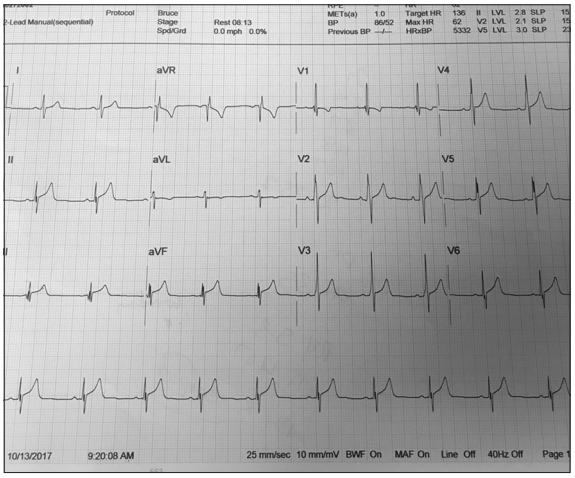

Figure 1: Sinus rhythm. Rightward axis deviation. Prominent R waves in right precordial leads. Likely early repolarization changes

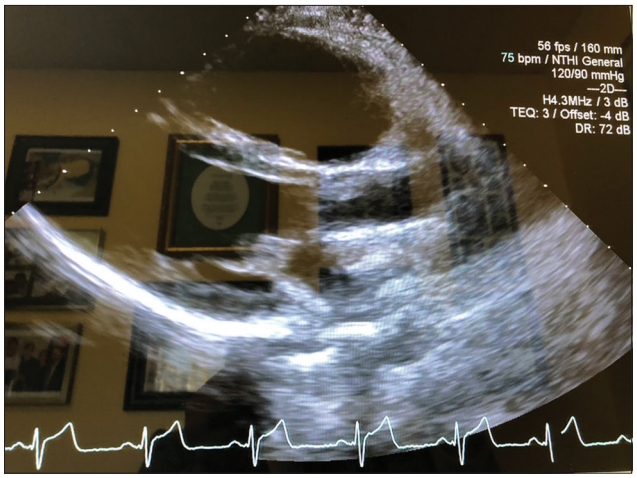

Figure 2: Parasternal long axis showing dilated right ventricle

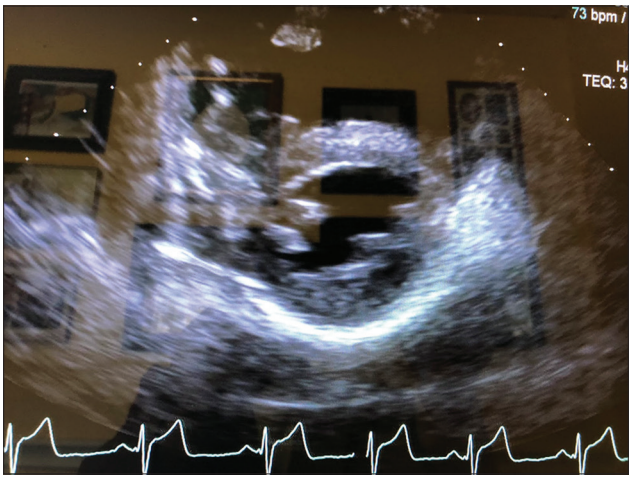

Figure 3: Parasternal short - axis showing dilated right ventricle

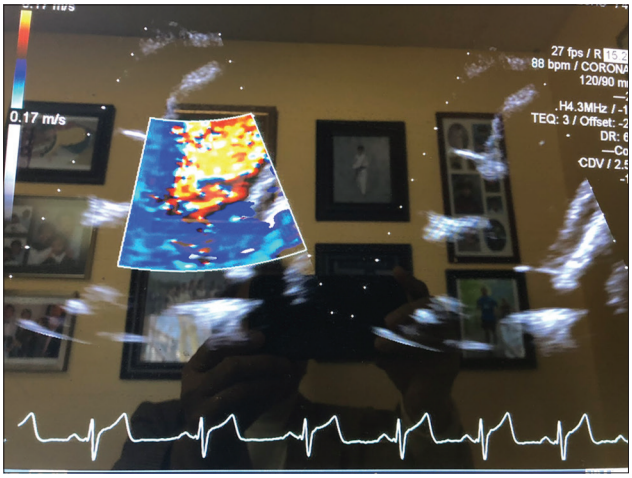

Figure 4: Color flow suggestive of sinus venosus atrial septal defect

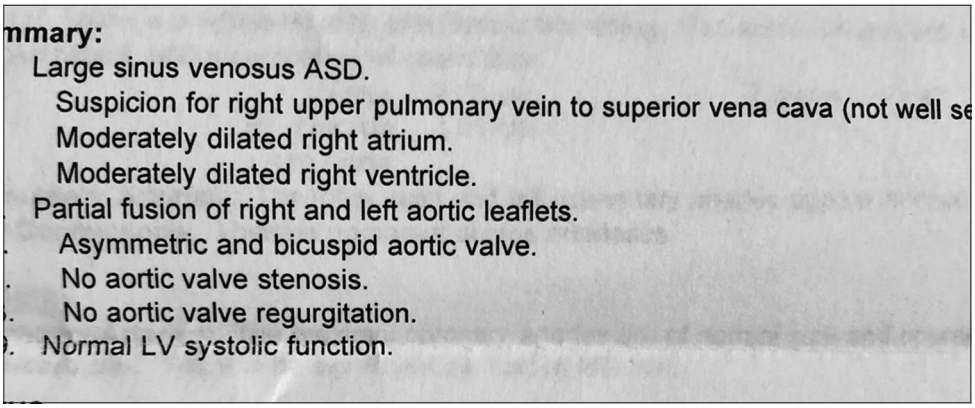

Figure 5: Summary of his two-dimensional echocardiography[11,12]

the anterior infundibulum, the RV apex, and inferior/diaphragmatic aspect of the right ventricle, the so-called "the triangle of dysplasia"[7] which can lead to dilation and aneurysm formation with paradoxical motion. LV and the interventricular septum can be affected at times.

There are two variants of this condition: The fatty variant mostly affects the right ventricle: Almost complete replacement of myocardium without thinning of the ventricular wall and the fibrofatty variant: With significant thinning of the RV wall and LV may be involved in this variant as well.[8]

Clinical features and diagnosis

The classic presentation of ARVC is usually arrhythmia, in the form of ventricular tachycardia, frequently from the RV outflow tract (RVOT) also known as adenosine-sensitive ventricular tachycardia. The range of manifestations varies from asymptomatic to premature ventricular contractions, biventricular heart failure, arrhythmia, and sudden cardiac death, usually in young and the athletes.[9]

ECG changes seen in this condition may include:[10]

1. An epsilon wave (most specific finding, seen in 30% of patients)
2. T-wave inversions in V1-3 (85% of patients)
3. Prolonged S-wave upstroke of 55 ms in V1-3 (95%)
4. Localized QRS widening of 110 ms in V1-3
5. Paroxysmal episodes of ventricular tachycardia with LBBB + left bundle branch block morphology (RVOT).

Plan of management

Based on this diagnosis, he was advised to see an electrophysiologist for a consult and possibly a defibrillator placement. I spoke to his mother to see if she was interested in a second opinion. She liked the idea. I called up Cleveland clinic and spoke to an electrophysiologist. As he was only 15 years old, they wanted him to be seen by a pediatric electrophysiologist to whom his mother eventually spoke; however, she could not afford to take him in person to Cleveland for consultation Figures 4 and 5.

Thereafter, I arranged his consultation with an electrophysiologist in Buffalo. Eventually, he ended up seeing a pediatric cardiologist/electrophysiologist at the University of Rochester. His tests were reviewed and more tests were requested including another 2D-echocardiography which was performed based on a pediatric protocol/guidelines at University of Rochester and the following pertinent findings were noted:

So now what?

His diagnosis has been completely changed from arrhythmogenic RV dysplasia to sinus venosus atrial septal defect!

Instead of being considered for a defibrillator placement, he is now preparing himself to undergo cardiac surgery to have his atrial septal defect closed off.

Lesson learned

In hindsight, based on my 2D-echocardiographic report showing dilated right ventricle and right atrium, I should have requested a transesophageal echocardiographic study (TEE) first before requesting a cardiac MRI. However, I did not see any clear-cut atrial septal defect (primum or secundum), and therefore, I could not think of any obvious cardiac pathology. Second, TEE is somewhat a semi-invasive procedure, and therefore, MRI came to my mind first. In addition, cardiac MRI report was very suggestive of arrhythmogenic RV cardiomyopathy!

The best part is that the patient is going to get the right kind of treatment, and so thankfully, this "mistake" did not hurt him.

Based on this never-to-forget experience, I am giving it a new name in my cardiology experience: "Sinus Mistakus!"

REFERENCES

1. Corrado D, Thiene G, Nava A, Rossi L, Pennelli N. Sudden death in young competitive athletes: Clinicopathologic correlations in 22 cases. Am J Med 1990;89:588-96.
2. Basso C, Corrado D, Marcus FI, Nava A, Thiene G. Arrhythmogenic right ventricular cardiomyopathy. Lancet 2009;373:1289-300.
3. Murray B. Arrhythmogenic right ventricular dysplasia/ cardiomyopathy (ARVD/C): A review of molecular and clinical literature. J Genet Couns 2012;21:494-504.
4. Basso C, Corrado D, Thiene G. Cardiovascular causes of sudden death in young individuals including athletes. Cardiol Rev 1999;7:127-35.
5. Valente M, Calabrese F, Angelini A, Basso C, Nava A, Rossi L, *et al. In vivo* evidence of apoptosis in arrhythmogenic right ventricular cardiomyopathy. Am J Pathol 1998;152:479-84.
6. Thiene G, Nava A, Corrado D, Rossi L, Pennelli N. Right ventricular cardiomyopathy and sudden death in young people. N Engl J Med 1988;318:129-33.
7. Marcus FI, Fontaine GH, Guiraudon G, Frank R, Laurenceau JL, Malergue C, *et al.* Right ventricular dysplasia: A report of 24 adult cases. Circulation 1982;65:384-98.
8. Pinamonti B, Sinagra G, Salvi A, Di Lenarda A, Morgera T, Silvestri F, *et al.* Left ventricular involvement in right ventricular dysplasia. Am Heart J 1992;123:711-24.
9. Foale RA, Nihoyannopoulos P, McKenna WJ, Oakley CM, Krikler DM, Rowland E, *et al*. Right ventricular abnormalities in ventricular tachycardia of right ventricular origin: Relation to electrophysiological abnormalities. Br Heart J 1986;56:45-54.
10. Jain R, Dalal D, Daly A, Tichnell C, James C, Evenson A, *et al.* Electrocardiographic features of arrhythmogenic right ventricular dysplasia. Circulation 2009;120:477-87.
11. Bayar N, Arslan S, Koklu E, Cagirci G, Cay S, Erkal Z, *et al*. The importance of ECG findings in the diagnosis of atrial septal defect. Kardiol Pol 2015;73:331-6.
12. Rojas CA, El-Sherief A, Medina H. Embryology and developmental defects of the interatrial septum. Am J Roentgenol 2010;195:1100-4.

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