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Case Report Open Access

Uncommon Cardiac Malformation in a Rare Genetic Disease

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Objectives

The main reason for the presentation of this clinical case was to discover the hidden diagnosis of a young patient, who presented lipothymia during a sports class, while participating in the running trial.

Material and Methods

I am presenting the clinical case of a young man aged 24, who was participating in a sports class at his university, when he was participating in the running trial; he suddenly got dizzy, had an accelerated heart rate, chest pain and lipothymia, without losing consciousness. His colleagues intended to call the emergency services, but the young man suddenly got better and refused to be admitted at the hospital. After this incident he presented two other episodes of dizziness and an accelerated heart rate, during the sports class, he went to the family doctor and he sent him to a specialist. During the consultation, I founded at the objective examination rhythmic heart sounds, HR=75 beats/min, a mid-systolic click in the mitral area, proto-systolic murmur heart without irradiation with the character of a vapour ejection, BP=120/80 mmHg, a normal vesicular sound.

The Objective Examination

In the general objective examination the following were determined: the thorax shape of the chest was excavated and scoliosis was present (Figures 1a-1d), tall aspect (Figure 2), arachnodactyly (Figures 3 and 4), ligaments hyper flexibility (Figures 5, 6 and 7a-7c), blue sclera (Figures 8a-8d) and wearing glasses, oral cavity malformation (Figure 9) high palatal arch (Figure 10) and dental malformations (Figure 11).

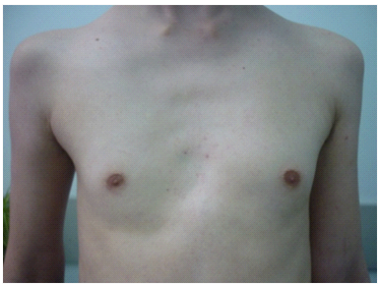

Figure 1a: The size of the thorax anterior-incidence of chest excavation.

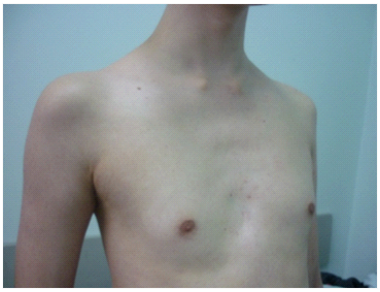

Figure 1b: The size of the thorax oblique–incidence of chest excavated .

The EKG shows a sinus rhythm, HR=78beats/min and a minor right bundle branch block (Figures 12a and 12b).

The chest X ray (Figure 13) shows aspect of the "drop heart" typical of this patient with ectomorph constitutional type and tall appearance.

The cardiac Doppler echocardiography (Figure 14) shows a prolapsed mitral valve, second degree mitral failure and unexpectedly was discovered an inter ventricular septum aneurysm and also an inter atrial septum aneurysm.

The abdominal CT (Figure 15) was within normal limits. The size of the abdominal aorta was normal, without aneurysms in other sites.

Therapy

It was concluded that the solution of the patient was referred to cardiovascular surgery. A procedure was performed to resected and

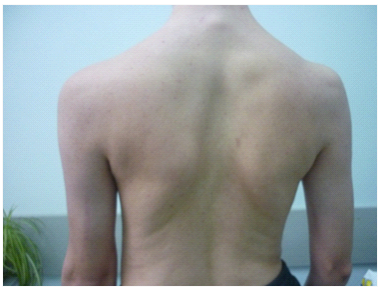

Figure1c: The size of the thorax–Posterior incidence-scoliosis.

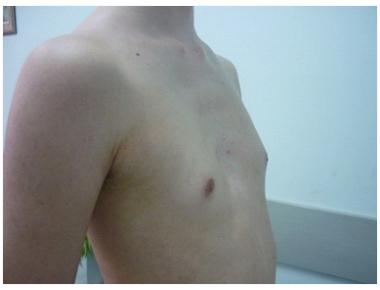

Figure 1d: The size of the thorax-Right lateral incidence–chest excavated.

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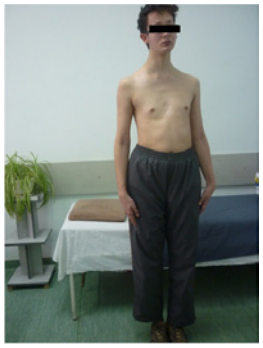

Figure 2: Ectomorph constitutional type-Tall Appearance.

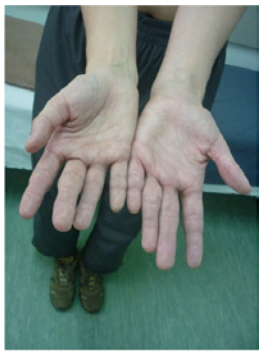

Figure 3: Long and thin fingers-Arachnodactyly.

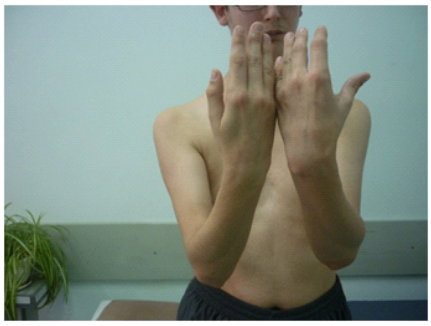

Figure 4: Long and thin fingers–Arachnodactyly.

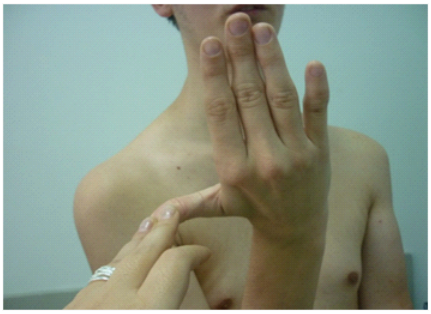

Figure 5: Ligaments hyper flexibility-Arachnodactyly.

repaired the aneurysms (Figure 16). In time of surgical intervention was discovered also a sinus of Valsalva aneurysms (SVAs). SVAs are a rare anomaly. The site of the aneurysm was the right coronary sinus. Surgery was the treatment of choice. The concomitant surgical repair of

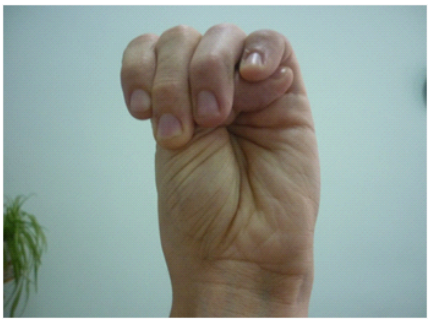

Figure 6: The sign of thumb-hyper flexibility.

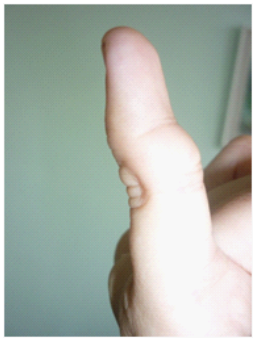

Figure 7a: Ligament and joint hyper flexibility .

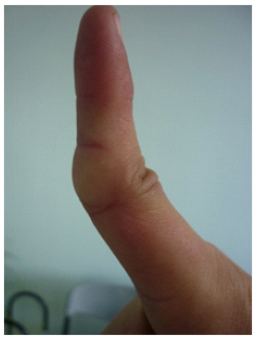

Figure 7b: Ligament and joint hyper flexibility.

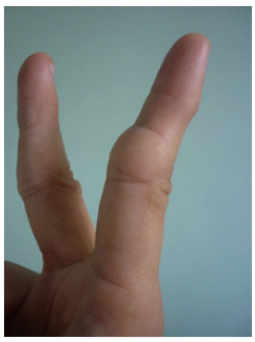

Figure 7c: Ligaments and joints hyper flexibility.

associated ventricular septal aneurysm, atrial septal aneurysm and the sinus of Valsalva aneurysm. Elective surgical repair was performed with low risk. The aneurysms was resected and repaired in this patient. The

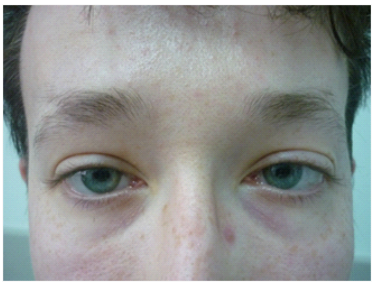

Figure 8a: Blue sclerotics and strabismus.

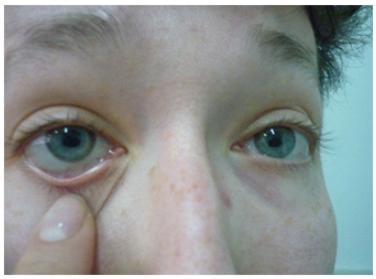

Figure 8b: Blue sclerotics and strabismus.

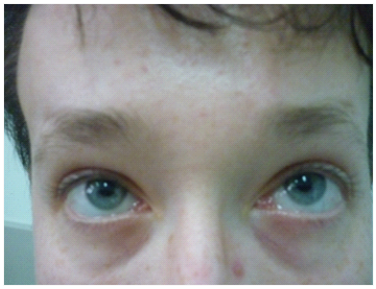

Figure 8c: Blue sclerotics.

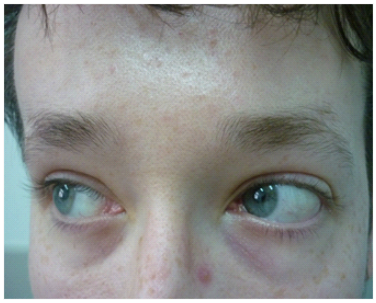

Figure 8d: Blue sclerotics.

development of dissecting aneurysm of the interventricular septum is rapid. Its prognosis is poor. Early diagnosis and operation are necessary. The wall of the aneurysm was resected and the interventricular septum was reconstructed. Good exposure was obtained from the incision of the ascending aorta and right ventricular outflow tract.

Results and Discussions

Mitral valve prolapse and mitral regurgitation are diseases that are frequently observed in Marfan syndrome, but the association with an inter atrial and inter ventricular septum aneurysm is very uncommon and rare. The final diagnosis was: Marfan syndrome, prolapsed mitral valve, mitral insufficiency grade II, aneurysm of inter atrial septum and aneurysm of inter ventricular septum, minor right bundle branch block. After surgical intervention the postoperative evolution of the patient was favorable after the inter atrial and inter ventricular septum have recast.

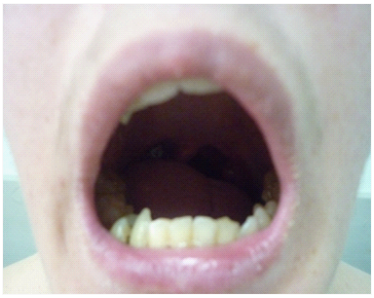

Figure 9: Malformation of oral cavity deviation of uvula at the right tongue.

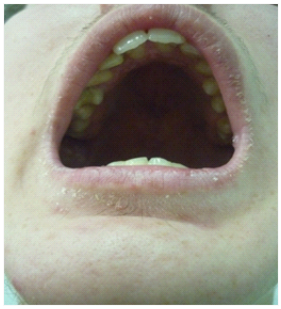

Figure 10: High palatal arch asymmetry and side and elevation of the right side of the tongue.

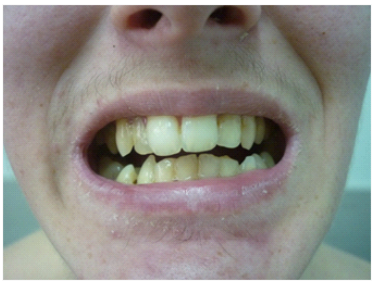

Figure 11: Dental malformation.

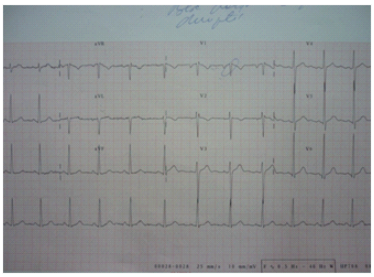

Figure 12a: EKG–sinus rhythm, minor right rate heart 75 beats/min, minor right bundle branch block.

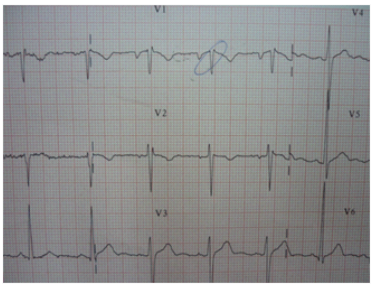

Figure 12b: View image - rate heart 75 beats/min, bundle branch block.

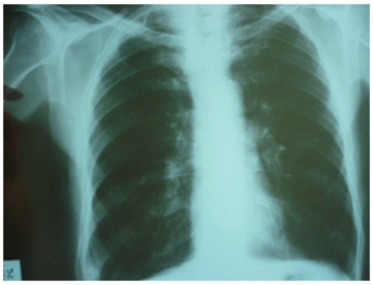

Figure 13: The chest X ray shows "drop heart".

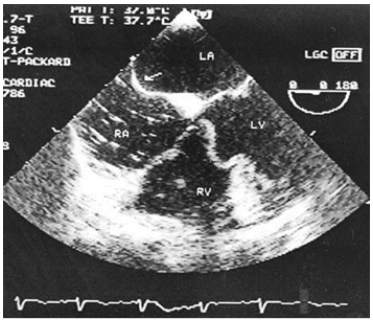

Figure 14: Findings of an Aneurysm of the Muscular Interventricular septum associated with aneurysm of the Interatrial Septum.

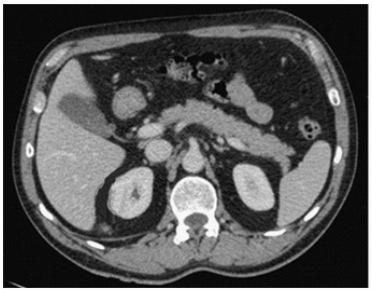

Figure 15: The abdominal CT normal-without aneurysms in other sites of the abdominal aorta.

As mentioned above the both aneurysms of the membranous interventricular and interatrial septum are an uncommon cardiac abnormality with no incidence statistic reported in the literature. It has been documented in isolation but is seen in association with congenital heart disease in 0.3% of cases and in association with VSDS in 19%

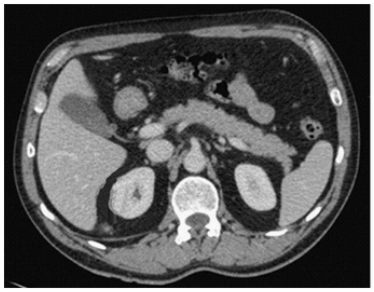

Figure 16: Intraoperative photograph showing a thin aneurysm of interventricular septum. Note that the aspirator (white cylinder in the upper part of the photograph) is introduced through the interventricular septum.

of cases [1]. Etiologies of the condition include idiopathic formation, trauma, infection, or as the result of spontaneous closure of a pre-existing VSD. The majority of cases are thought to be congenital in origin.

The rarity and obscurity of its clinical symptoms and the potential sequelae of membranous VSAs attest to the importance of awareness among clinicians and radiologists [2]. Clinical manifestations are variable. Some patients are asymptomatic with normal physical examination whereas others can present with hemodynamic compromise. Possible complications include conduction abnormalities, aneurysm rupture, bacterial endocarditis, right ventricular outflow tract obstruction, significant intracardiac shunting, and thromboembolism.

Atrial septal aneurysm may be isolated or associated to another anomaly. Commonest association is patent foramen ovale (PFO). Silver and Dorsy found patent foramen ovale in 8 out of 16 patients [3]. Other associations are atrial septal defect [2], mitral valve prolapsed [4,5] tricuspid valve prolapse, Marfans syndrome, sinus of valsalva aneurysm and aortic dissection [1]. Shunt across ASA is more frequently detected with transesophageal echocardiography than with transthoracic echocardiography [1]. Association with mitral and tricuspid valve prolapse and other abnormalities such as Marfans syndrome, sinus of valsalva aneurysm may point to common inherent connective tissue deficiency. Familial clustering of ASA has also been reported.

The diagnosis of the Marfan syndrome was late because the patient didn't presented to the physician before this events.

Conclusions

1. The Marfan syndrome is a known but, very rare a genetic disease.

2. The mitral valve prolapse diagnosis may be associated with, or not with mitral failure, it is also usual.

3. The inter atrial and inter ventricular septum aneurysms are very uncommon.

4. The diagnosis is possible if the genetic sub layer of the disease is taken into account and especially the presence of the lax connective tissue in a large amount and ligaments hyper flexibility, which could be valid in the case of inter-atrial and inter-ventricular septum, which are usually more lax and with an exaggerated mobility that can produce these kinds of changes.

5. The thoracic abdominal CT has not shown any other aneurysm in the aortic artery level.

6. The postoperative evolution has been favorable with the recast of the inter atrial septum and inter ventricular septum.

7. This patient with atrial and ventricular aneurysm of the septum has an increase risk of an embolism at different sides because this malformation develops a risk of clots at these levels and after that arterial embolism with the risk of a stroke attack or acute arterial ischemia in different arterial sides.

8. When we find in our medical practice a clinical case with Marfan syndrome we must take into account the possibility of the existence of this uncommon malformation: aneurysm of inter atrial or inter ventricular septum is very dangerous because there exists possibilities to develop clots at these levels and with an increased risk of acute arterial embolism anywhere in the arterial system and the development of an acute ischemia at the arterial level caused by an obstruction.

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