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CASE REPORT

Oligosymptomatic Eisenmenger syndrome in adolescent

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Abstract

Eisenmenger syndrome is characterized by high pulmonary pressure, caused by a high pulmonary flow state, with inverted shunt (right-to-left) or bidirectional. The condition is a complication of a congenital heart defect not repaired and the diagnosis is based on clinic, echocardiogram and cardiac catheterization. The case reported is a 13 years old teenager that had lost the follow-up a year after the diagnosis with 5 years old. She returned with dyspnea, worsening of echocardiographic patterns, pulmonary hypertension and under weight. Without surgical indication, the patient were followed up clinically for a year and evolved with a similar dyspnea pattern, healthy weight and normal neurological development. ES is an extreme condition in which, due to a previous cardiac defect, pulmonary vascular resistance becomes greater than systemic resistance and the flow reverses irreversibly, leading to high morbidity, mainly due to hypoxemia. Besides the surgical treatment, some drug options may be used, such as: bosentan, PDE-5i, prostanoids and sildenafil associated with bosentan. Bosentan is the first choice drug, the others should be evaluated individually. Despite the aforementioned therapeutic guidelines, it is essential to avoid congenital heart defects from reaching this dramatic picture. The oligosymptomatic evolution of the case describes a possibility of the natural history of this entity.

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INTRODUCTION

Eisenmenger Syndrome (ES) is characterized by the elevation of pulmonary pressure to systemic levels, caused by increased pulmonary vascular resistance, with reverse (right-left) or bidirectional shunt. Such elevation is caused by a large unrepaired congenital defect, intra (ventricular septal defects) or extracardiac (large vessel interposition)[1,2]. The main clinical manifestations of this entity are: dyspnea on exertion, cyanosis, syncope, chest pain and arrhythmias. The diagnosis is made by the clinic in addition to the performance of tests, such as the electrocardiogram and cardiac catheterization, which closes the diagnosis[3]. SE, with an exact prevalence still unknown, is a progressive, irreversible condition associated with high mortality and morbidity when not diagnosed early[1]. For this reason, it is very important to prevent the condition, with corrective surgery before the pulmonary changes appear. Once the syndrome has developed, there is no specific treatment other than transplantation. Drug treatment is based on clinical support[3]. The treatment of SE is complex and has limited results. It is essential to avoid any factor that could decompensate the balanced physiology.

This paper aims to describe a case of Eisenmenger Syndrome with an atypical presentation (oligosymptomatic) diagnosed in a Brazilian university hospital.

CASE REPORT

Patient GVS, female, 13 years old, born in Goiânia-GO, coming from Abadia de Goiás, was referred to the Pediatrics outpatient clinic at 4 years and 2 months due to agenesis of the left kidney, diagnosed since pregnancy. As comorbidities, she had VSD, esophageal atresia (corrected at 9 days of life) and asymmetry of the extremities (scanometry showed a left lower limb 3.5 cm longer than the right), resulting in the diagnosis of an association with VACTREL. He had normal MPN and normal renal function. In genetic investigation, any type of chromosomal syndrome was excluded.

The echocardiogram showed VSD of moderate size with bidirectional shunt, right ventricle (RV) with moderate increase in volume and significant pulmonary artery hypertension (PAH), with indication for surgery for VSD. Despite the diagnosis of Eisenmenger Syndrome in the period, he did not undergo surgery and lost a segment after one year.

After 7 years, he returned to the service with weight and height deficit and dyspnea on great exertion and strong emotions . It was evaluated by the pediatric pneumopediatrics, cardiology and pediatric endocrinology teams. She had borderline growth velocity, which was associated with Eisenmenger Syndrome. A new echocardiogram was performed, which showed an atrioventricular septal defect with large VSD, bidirectional shunt plus incompetent mitral cleft with moderate regurgitation, EF: 71%, PSVD: 98 mmHg, significant increase in RV, mild biatrial dilatation. He underwent catheterization that showed precapillary PAH with vasoreactivity test with a 32% drop in pulmonary vascular resistance, Qp/Qps<1 and no surgical indication.

The patient had thelarche at age 10 and menarche at age 11, being referred to the gynecology outpatient clinic to start high-efficacy contraception due to the impossibility of being able to get pregnant, news that emotionally affected GVS. At the time of this report, the patient is 13 years old, eutrophic, M3P3, dyspneic on strenuous exertion, not taking medication and not using a contraceptive method. She is under multiprofessional follow-up, with no further complaints.

COMMENTS

SE is an extreme condition in which, due to a previous heart defect, pulmonary vascular resistance becomes greater than the systemic one and the flow is irreversibly reversed, leading to a clinical picture of high morbidity, mainly due to hypoxemia[1,2].

Several congenital heart defects are associated with an increased risk of developing this syndrome, the main ones being interventricular communication (IVC), patent ductus arteriosus and interventricular septal defects[1,4]. Showing the importance of this condition, it is estimated that 10% of patients with IVC (as in the case reported here), after 2 years of age, may progress to SE. However, in recent years, with the advancement of cardiac surgery and improvement in the care of these patients, the cases of SE have reduced considerably[5].

The diagnosis is initially based on clinical features. Dyspnea on exertion is common, and was the only symptom reported by GVS when seeking medical attention again. Other signs and symptoms are usually present, such as cyanosis, syncope, chest pain, arrhythmias and, more rarely, right heart failure. Blood count, electrocardiogram and echocardiogram are also part of the investigation. Cyanosis and erythrocytosis are marked in SE and rarely occur in patients with other causes of PAH[2]. In spite of all the others, cardiac catheterization, with the measurement of pulmonary artery pressure, is the test used to confirm the diagnosis[3].

The treatment of SE is complex and with limited results. It is essential to avoid any factor that could decompensate the balanced physiology. Dehydration, high altitude and moderate to severe isometric exercise should be avoided. For the same reason, pregnancy is strictly contraindicated for these patients, leading to a mortality of 30% to 50%[6]. Such information can greatly affect patients, as occurred in this case described, and, for this reason, the risks must be explained in a careful and detailed manner.

As supportive measures, drug treatment can be tried. The use of endothelin receptor antagonists (ERA) such as Bosentan was the first advanced therapy to be studied in SE. The Bosentan Randomized Trial of Endothelin Agonist Therapy-5 (BREATHE-5) has shown that its use improves symptoms and the ability to perform physical activity by reducing PVR and increasing pulmonary blood flow. Currently, European

guidelines support the use of Bosentan as the first-choice treatment for patients with functional class (FC) III-IV[7-9].

Despite few randomized clinical trials, there is also evidence of the beneficial effects of PDE-5i and prostanoids in patients with SE in FC III. However, as the levels of evidence are lower than with the use of ERAs, PDE-5i are recommended as second-line therapy[7-9].

The combined use of different types of advanced therapies to maximize clinical benefit can improve treatment outcomes and prognosis. A randomized double-blind study evaluated the effect of the combination of sildenafil and bosentan in 21 patients with SE (NYHA II and III). In this study, it was shown that the only benefit of combined therapy was in relation to peripheral oxygen saturation[10,11]. A retrospective study of 121 patients showed that the use of supplemental sildenafil after failure of the single therapy with Bosentan improved patient performance, with a significant difference in 1 to 3 year survival in the advanced therapy group compared to the treatment group. non-advanced therapy[4,12].

According to some studies, patients with right ventricular dysfunction may benefit from home oxygen supplementation, which would also help to reduce the progression of erythrocytosis, reducing the need for hemodilutions[12].

Anticoagulation and antiplatelet aggregation is still a controversial subject that needs further studies. The decision on the establishment of anticoagulant therapy in children and adolescents depends on the analysis of each case individually. There is no evidence that antiplatelet drugs used alone can have a significant impact on the outcome of these patients. Combined use, however, may be justifiable[12].

In addition to drug treatment, interventional procedures such as balloon atrial septostomy and transplants can also be used in cases that are refractory to other treatments. [5] In patients without left ventricular dysfunction, in which the correction of the heart defect is still possible, lung transplantation concomitant with the repair of the anatomic defect can be performed[12]. Heart-lung transplantation is another option, being a treatment recognized as successful in ES. It is the only procedure capable of improving patients' quality of life and potentially also the only one to increase survival. However, the ideal time for the procedure is a challenge, especially due to the long lines for the transplant, which impacts the survival outcomes of patients undergoing the procedure[13].

As seen, there is still great difficulty in managing SE, and its consequences greatly affect the lives of patients. Preventing congenital heart defects from reaching this dramatic picture is essential. Pediatricians, in particular, are of great importance in these cases, with the possibility of early diagnosis and indication of immediate correction of the cases. The oligosymptomatic evolution of the reported case describes a possibility of the natural history of this entity.

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