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CASE REPORT

Coronary disease with fatal disclosure in 9 year old child: a case report

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Coronary Artery Disease, Child, Hyperlipoproteinemia Type II, Hypercholesterolemia.

Abstract

This article is about a 9-year-old male child with chest pain and clinical signs of hypercholesterolemia. During medical investigation we confirmed primary hypercholesterolemia and diagnosed bicuspid aortic valve and severe coronary disease, this being the cause of chest pain. Unfortunately, the patient evolved to death during the procedure, despite a successful coronary stent implantation and cardiac resuscitation. Atherosclerotic disease is rare among children, being commonly associated to familial forms of hypercholesterolemia. Bicuspid aortic valve, on the other hand, is a common congenital heart disease, but the association between both is rarely described. Chest pain is a common cause of complaint, and although in the vast majority of cases it is not associated with cardiac etiology, in the setup of hypercholesterolemia, especially familial forms, it should prompt the pediatrician attention to threatening etiology.

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INTRODUCTION

Chest pain in pediatric patients, although common[1]. should be investigated in detail for the cardiac cause when in the context of familial hypercholesterolemia[2]. This condition can cause rare aggravations in children, such as obstructive coronary atherosclerosis[3]. Bicuspid aortic valve is a common congenital abnormality, however the association with hypercholesterolemia is rarely described in the literature[4]. The aim of this article is to report the case of a pediatric patient with obstructive coronary atherosclerosis associated with a bicuspid aortic valve.

CASE REPORT

In May 2018, patient KSM, from Novo Repartimento, Pará, was referred to Fundação Hospital de Clínicas Gaspar Vianna (FHCGV) in Belém, Pará, due to chest pain. At the consultation, the mother reported that five months earlier the patient had had an episode of chest pain (non-radiating retrosternal pain) followed by fainting while playing. On the same day, he sought medical care at a health center in his municipality, about five hundred kilometers from the capital, where he underwent an electrocardiogram, which had a normal result (SIC). At the time of the examination, the attending physician heard a heart murmur and requested a transthoracic echocardiogram.

The prescribed exam was performed in his hometown two months after the first episode of chest pain followed by syncope and the patient was referred to FHCGV, a referral service in cardiology in the State of Pará, for evaluation by a pediatric cardiologist. The mother also reported that the child persisted with episodes of chest pain on exertion, which improved at rest, but without further fainting.

As for the history, the mother reported that the patient had been hospitalized for pneumonia at four years of age. No noteworthy family history was denied. The food diagnosis was a qualitative error (little vegetable intake), while the nutritional assessment showed that the patient was in a thin state with weight = 21.5kg; Height = 125cm; BMI of 13.76kg/m² (between percentile 15 and percentile 3 for age). Assessment of neuro-psychomotor development showed appropriateness for age.

On physical examination, the patient was in good general condition, eupneic, acyanotic, normocardic, with xanthomas on the extensor face of the elbows and knees (Fig 1). Cardiac auscultation showed an ejective systolic murmur (4+/6+), with the presence of thrill, in the high right sternal border. Pulmonary auscultation showed no changes and the pulses were median, symmetrical and palpable in the upper and lower limbs. The abdomen showed no changes.

A chest X-ray showed a slight left ventricular enlargement. The electrocardiogram showed T wave repolarization changes from V1 to V3 and signs of left ventricular overload. The echocardiogram revealed an *ostium secundum atrial septal defect* of 4mm, a bicuspid aortic valve with leaflet calcification



Figure 1. Xanthomas on the knee (A) and elbow (B).

and limited mobility with a maximum systolic gradient of 77mmHg and a mean of 47mmHg. and normal ejection fraction with a slight increase in left cavities. Laboratory examination showed total cholesterol of 720mg/dl (HDL: 34mg/dl; LDL: 644.7 mg/dl; VLDL: 41.3mg/dl; Triglycerides: 219 mg/dl).

In view of the diagnosis of primary hypercholesterolemia, simvastatin 10mg/day was started and, due to the association with bicuspid aortic valve and severe aortic stenosis, a hemodynamic study was indicated. The study showed parietal irregularities in the thoracic aorta, a thickened aortic valve with reduced mobility, and a marked obstructive lesion in the ostia of the right coronary artery and the left main coronary artery. During the procedure, the patient presented refractory bradycardia and hypotension, a *stent was implanted* in the left main coronary artery to improve local blood circulation, but the patient died (Fig 2).

DISCUSSION

In pediatrics, chest pain is a common complaint[1]. Although in many cases the cause is not a cardiovascular problem, an accurate diagnosis must be sought and problems that can generate potentially fatal situations[1] such as coronary artery disease, presented in this case, whether or not associated with congenital heart disease.

Changes in lipid metabolism are important risk factors for coronary artery disease[5]. Although the process of atheromatous plaque formation begins in childhood, the clinical manifestations of acute or stable coronary artery disease usually occur in adulthood[5]. In most cases, the increase in circulating lipids is due to secondary causes such as increased dietary intake, sedentary lifestyle, diabetes and chronic liver disease[6]. However, primary hypercholesterolemia, of genetic origin, can cause clinical atherosclerosis at a very early age[7].

In the pediatric population, the recommendation for assessing serum levels of cholesterol and triglycerides is between nine and eleven years and between seventeen and

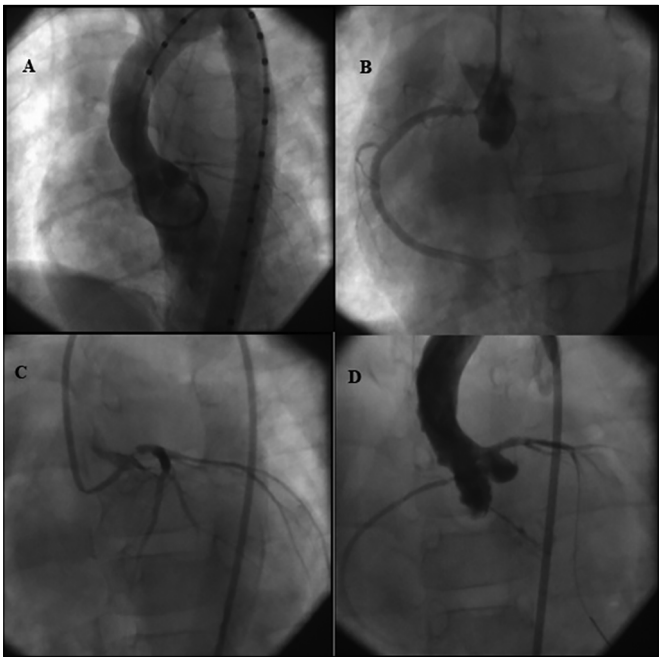


Figure 2. Cardiac catheterization showing : (A) irregularities in the aortic wall, obstructive lesions in the right (B) and left (C) main coronary arteries; result after implantation of a "stent" device in the left main coronary artery. (D).

twenty-one years[6]. Already between two and ten years of age, this monitoring is carried out in situations where there are first-time relatives grade with, mainly, severe forms of dyslipidemia; dyslipidemia clinic; other diseases such as hypothyroidism, nephrotic syndrome, immunodeficiency and systemic arterial hypertension, as well as the use of drugs that can increase cholesterol levels[2,6]. Likewise, careful monitoring of children with familial dyslipidemia is mandatory[2]. Despite being in an age group for laboratory screening and presenting clinical signs of dyslipidemia (xanthomas), the patient had no previous investigation report, which delayed the diagnosis, allowing the evolution of the condition.

Familial hypercholesterolemia (FH) is a genetic disease with an autosomal dominant pattern, in which there is a reduction in the hepatic *clearance* of LDL-cholesterol (LDL-c) due to a series of defects in the gene encoding the receptor for this substance[6]. There is then the accumulation of LDL-c in the systemic circulation, which causes all the deleterious effects associated with the condition. It can occur in the heterozygous form, with an estimated incidence between 1:250-1:500 individuals, and in the homozygous form, which is rarer and has more severe clinical manifestations, with an incidence ranging from 1:160,000 to 1:1,000,000[6,8].

The diagnosis of FH should be suspected in the presence of clinical signs of extravascular cholesterol deposits, such as xanthomas, xanthelasmas and corneal arches, as well as reports of first-degree relatives with an early history of coronary artery disease and high cholesterol[8]. Elevated serum levels of total cholesterol and LDL fraction, the latter with values above 150-190mg/dl, on two successive occasions and in the absence of secondary causes, increase the possibility of a genetic cause[6,8]. In the case in question, even though the patient had no family history worth noting, given the clinical picture strongly suggestive of the presence of xanthomas, the absence of factors that raise the possibility of secondary causes (obesity, use of medication, other underlying disease) associated with abnormal laboratory tests, the diagnosis of primary hypercholesterolemia was made.

Coronary artery disease in children is also rare and is mainly related to congenital anomalies or Kawasaki disease[1,9]. Cases related to atheromatous plaque are even rarer, and in the pediatric population they are related to familial hypercholesterolemia[3]. Symptoms such as chest pain, fatigue and syncope point to myocardial ischemia and the presence of xanthomas in the limbs, in addition to high serum levels of LDL cholesterol and triglycerides, increase the likelihood of coronary artery disease[3,7,10]. The definitive diagnosis of coronary artery disease is made by coronary angiography, to define the lesions and therapeutic intervention[1].

Treatment is based on lifestyle changes and the use of oral statins[2,8]. Non-drug treatment consists of physical exercises, a diet low in saturated fat and greater intake of vegetables, fish, etc.[8]. Although there is controversy regarding the age at which drug treatment begins[2,8], there is consensus on the use of statins, ezetimibe and bile acid sequestrants[2,8]. The therapeutic objective is to reduce LDL-c to levels as low as 110-130mg/dl, in order to reduce cardiovascular risk in the long term[2,8]. Unfortunately, in the case described, the young man had a complication that prevented an evolution in the treatment.

The bicuspid aortic valve is strongly associated with diseases of the aorta[10], such as aortic stenosis presented by the patient, in addition to having a recognized role in the early onset of coronary artery disease[11]. The earlier appearance of atherosclerotic plaques in the aorta and coronary arteries of patients with familial hypercholesterolemia has already been described in the literature[12]. The unusual and rare association between these two risk factors for coronary artery disease in the patient may have been fundamental for the early appearance of the clinical picture presented.

CONCLUSION

Chest pain in pediatric patients is a common symptom and should not be taken lightly by the pediatrician. Although the cause is rarely a cardiovascular problem, in some situations, such as those presented in this case, they can signal the existence of potentially fatal conditions. Attention to the patient's history and physical examination, aided by laboratory and imaging tests, should guide the pediatrician in order to obtain an accurate diagnosis, in order to avoid tragic outcomes and ensure adequate treatment for patients.

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