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CASE REPORT

An unusual congenital heart disease, Pentalogy of Fallot

Una Cardiopatía Congénita inusual: Pentalogía de Fallot

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ABSTRACT

Introduction: Fallot's pentalogy is a cyanotic congenital heart disease, with right-to-left shunt and decreased pulmonary flow, which appears when adding a fifth cardiac anomaly to Fallot's tetralogy: atrial septal defect.
Objectives: the objective was to expose as clearly as possible the order of the clinical method applied during medical practice, the symptoms and signs that allowed diagnosing this entity and redirecting behavior.
Case presentation: we present the case of a 43-year-old patient, who from birth presented crises of respiratory distress, moderate mental retardation and musculoskeletal development; she was diagnosed with Tetralogy of Fallot and was considered by the surgical team as inoperable. She went to the clinic for a progressive worsening of her condition, physical examination detected acrocyanosis, Hippocratic fingers and holosystolic murmur of intensity IV-V/VI on the left sternal border. A series of imaging and electrocardiographic examinations were performed to establish the diagnosis, which included plain chest X-ray in posteroanterior projection and echocardiogram, the latter allowing the definitive diagnosis; Pentalogy of Fallot. The recommended treatment for this condition is heart surgery.
Conclusions: congenital heart disease has a diverse presentation around the world, as well as its severity. It was decided to present the case because it was uncommon in medical practice due to its low frequency. This made clear the relevance of an early diagnosis of these entities for the patient's quality of life.

Keywords: Congenital Heart Disease; Tetralogy of Fallot; Cardiac Surgery; Thoracic Surgery; Radiography.

RESUMEN

Introducción: la Pentalogía de Fallot es una cardiopatía congénita cianótica, con cortocircuito de derecha a izquierda y flujo pulmonar disminuido, que aparece al agregar una quinta anomalía cardiaca a la tetralogía de Fallot: la comunicación auricular.
Objetivos: se trazó como objetivo exponer lo más claramente posible el orden del método clínico aplicado durante la práctica médica, los síntomas y signos que permitieron diagnosticar esta entidad y redireccionar la conducta.
Presentación del caso: se presenta el caso de una paciente de 43 años de edad, quien desde su nacimiento presentó crisis de dificultad respiratoria, retraso mental moderado y del desarrollo musculoesquelético; la misma fue diagnosticada con una Tetralogía de Fallot y fue considerada por el equipo quirúrgico como no operable. Acudió a consulta, años después, por un empeoramiento progresivo de su cuadro, al examen físico se detectaron acrocianosis, dedos hipocráticos y soplo holosistólico de intensidad IV-V/VI en el borde

esternal izquierdo. Se realizó una serie de exámenes imagenológicos y electrocardiográficos para establecer el diagnóstico, la cual incluyó radiografía simple de tórax en proyección posteroanterior y ecocardiograma, esta última permitió el diagnóstico definitivo; Pentalogía de Fallot. El tratamiento recomendado para esta afección es la cirugía cardiaca.
Conclusiones: las cardiopatías congénitas tienen una presentación diversa en todo el mundo, así como su gravedad. Se decidió presentar el caso por resultar poco común en la práctica médica debido a su baja frecuencia. Con ello quedó claro la relevancia que posee un diagnóstico temprano de estas entidades para con la calidad de vida del paciente.

Palabras Clave: Cardiopatías Congénitas; Tetralogía de Fallot; Cirugía Cardíaca; Cirugía Torácica; Radiografía.

INTRODUCTION

Congenital heart disease (CHD) refers to structural malformations of the heart and great vessels resulting from an error in the embryogenesis of these structures.[1,2] They occur due to alterations in the embryonic development of the heart, especially between the third and tenth weeks of gestation.[3] However, the causes of many of these conditions are not yet fully understood. Approximately half of the most complex ones are fatal in the first years of life and are the focus of pediatric cardiology research. Heart malformations are mentioned in the Ebers Papyrus (2 000 years before our era) and defined by physicians such as Aesculapius and Hippocrates as "incomplete cavities." Although the first anatomical description of Tetralogy of Fallot was made in 1671, it was the French physician Étienne Fallot who, in 1888, described two other components of the tetralogy: right ventricular hypertrophy and dextroposition of the aorta over the septal defect.[4]

Worldwide, around eight out of every 1 000 live births have a congenital heart defect. In Cuba, around 300 children are born with congenital heart disease every year. The prevalence at birth ranges from two to three per 1 000 live births.[5]

At the end of the 1980s, a program aimed at prenatal diagnosis of CHD was launched in Cuba. Initially, it was centralized in the country's capital, but as specialists were trained for this task, it was extended to the rest of the provinces. Thanks to the establishment of this screening program and the work of professionals committed to their work and patient care, the number of prenatal diagnoses of this condition has increased.[6]

This group of diseases includes tetralogy of Fallot, which is cyanotic congenital heart disease with right-to-left shunting and decreased pulmonary flow, in which five anatomical defects are present: Pulmonary stenosis, right ventricular hypertrophy, ventricular septal defect, extraposition of the aorta over the septal defect, and atrial septal defect.[7] It differs from the tetralogy of Fallot in the presence of an atrial septal defect, which causes major complications or sequelae in the short, medium, or long term for this group of individuals.[7] The diagnosis is suspected based on symptoms and confirmed by echocardiography, which is usually sufficient to define the anatomy before surgery during infancy. Cardiac magnetic resonance imaging has become a tool for diagnosis and monitoring both before and after surgery, especially in patients with more complex pulmonary stenosis or pulmonary artery atresia with multiple collaterals.[8] It has been established as the method of choice for assessing the degree of pulmonary regurgitation and evaluating right ventricular hypertrophy.

Due to its low frequency and the importance of knowledge about it, this article aims to present a case of tetralogy of Fallot in a 43-year-old female patient.

CASE PRESENTATION

A 43-year-old female patient with white skin, according to her family, began to experience episodes of respiratory distress at birth; these symptoms started to attract the attention of her parents, who had no personal medical history at that time. At 3 months of age, as a result of these symptoms, but without any bluish discoloration of the skin or mucous membranes, she was admitted to the Manuel Fajardo Hospital for evaluation due to the possibility of a congenital heart defect. During her admission, a complex heart condition was diagnosed, whose clinical, anatomical, and hemodynamic characteristics led the cardiovascular surgery team treating her to conclude that palliative surgery was not possible.

At the time of consultation, it was found that she currently has severe mental retardation and muscular and skeletal development. Family members report that she has experienced dyspnea on exertion and orthopnea, so it was decided to admit her to the health center for study and treatment.

Physical examination revealed acrocyanosis, clubbing of the fingers, and an IV-V/VI holosystolic murmur at the left sternal border.

Several imaging tests were performed, which are shown below. Figure 1 shows the electrocardiogram, which reveals right ventricular hypertrophy (high R waves in V1) and early transition zones from V2 to V3. The left precordial leads show QRS complexes.

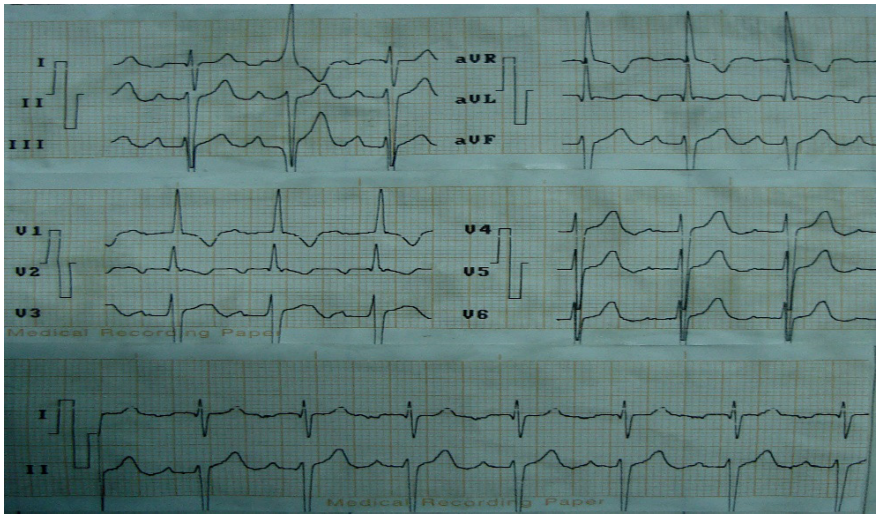

Figure 1. Taken from the patient's medical record

Figure 2 shows a simple chest X-ray in the posteroanterior view, showing an increased cardiothoracic index, widened aortic shadow with aortic arch to the left, and decreased pulmonary vascular network.

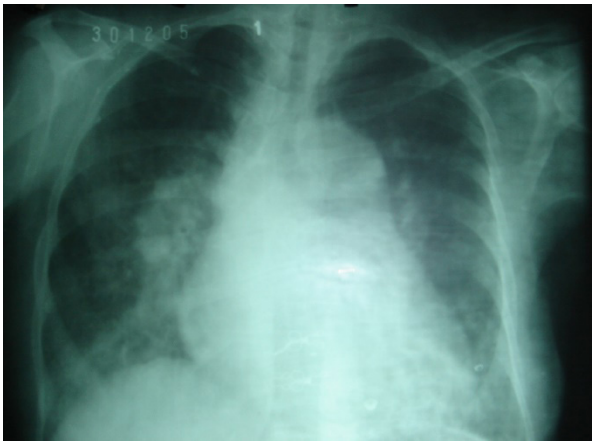

Figure 2. Taken from the patient's medical record

Figure 3 shows ventricular septal defect and severe infundibular and valvular pulmonary stenosis. Absence of the interatrial septum is also observed.

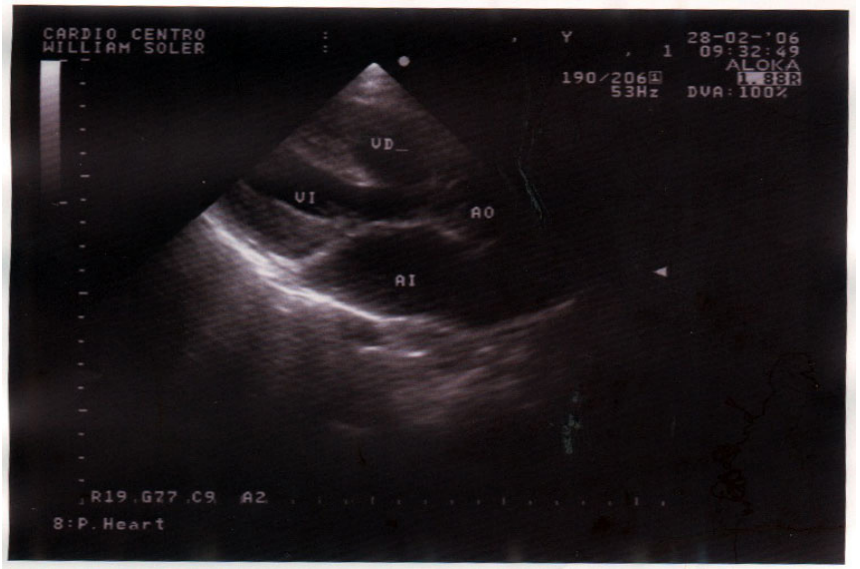

Figure 3. Taken from the patient's medical record

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Figure 4 shows aortic override, mitral-aortic continuity, and a left and patent aortic arch. Figure 6 shows the inverted location of the liver and spleen. Based on the above findings, we concluded that this case was an unoperated tetralogy of Fallot accompanied by situs inversus with levocardia.

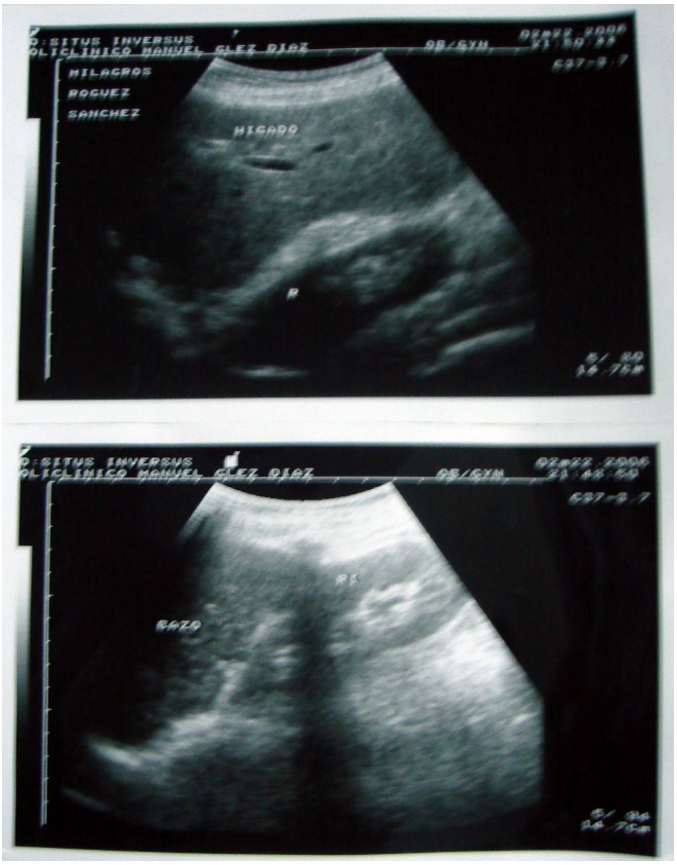

Figure 4. Taken from the patient's medical record

Given its rarity in the scientific community, the patient's family gave their consent for this clinical case to be shared.

DISCUSSION

Tetralogy of Fallot is the most common cyanotic congenital heart disease in adults, with an incidence of 0,08 % and accounting for 5-8 % of all congenital heart diseases. It affects approximately 1 in 8 500 live births, with a slight predominance in males over females. Comparative literature provides data indicating that genetic factors play a decisive role in the onset of this disease. There are not many cases reported or information available on pentalogy, but it is suggested that it is more prevalent in males,[9] which is not consistent with the present study, as the patient was female. This manuscript corresponds to the results found in various studies on the tetralogy of Fallot.[12,13,15,16] Still, however, it was found that there is little literature on this unusual presentation called pentalogy. Although Cuba does not have much-published content on the subject, several studies are related to this condition. It also coincides with the studies by Santos et al.[10], who suggest that atrial septal defect was the cardiac malformation most commonly associated with tetralogy of Fallot, turning it into pentalogy.[10] Right aortic arch can be found in up to 25 % of cases. Cyanosis appears most frequently between the third and seventh month of life, except when pulmonary stenosis is very severe, in which case it appears at birth.[11] Diagnosis is suspected based on symptoms and physical examination and confirmed by echocardiography or magnetic resonance imaging. In the case presented, echocardiography was of exceptional diagnostic value.

Cardiac surgery is the recommended treatment, with at least one-third of patients requiring some intervention before one year of age.[12,13] Survival beyond the third decade is exceptional (2 %) without surgical intervention, according to epidemiological studies and autopsy data. With corrective surgery, it is > 90 %. If anatomical surgical reconstruction is performed, the quality of life is optimal, and the only possible sequela is pulmonary insufficiency, which in some cases requires pulmonary valve replacement. Late mortality is related to reoperations, arrhythmias, and bacterial endocarditis. Infectious endocarditis is a serious complication that can have a significant impact on the prognosis and evolution of patients with congenital heart disease. We must

also make it clear that cyanotic heart disease predominates in the highest-risk group.[14,15]

It should be noted that since 1986, the William Soler Cardiocenter has been recognized as the country's leading institution for the care of patients with congenital heart disease. The efforts of surgeons, interventional cardiologists, and clinicians, focused on improving and providing the best possible care to heart patients, have increased short- and long-term survival. Most reach adulthood, a complex process that guarantees the continuous improvement of the quality of life of a vulnerable population that requires timely diagnosis, medical treatment, surgery, interventional care, and rehabilitation throughout their entire life cycle.[7]

In the present case, the patient was diagnosed with tetralogy of Fallot at the age of 43, a disease that had been overlooked in all her previous consultations. Her evolution was followed by cardiology and genetics specialists. It is essential to highlight the need for early diagnosis of this type of condition, which greatly ensures the survival of the individual and prevents the development of complications. Its clinical presentation has a broad context, and the cardinal signs of cyanosis, shock, and heart failure can be confused with pulmonary or infectious problems, which delays diagnosis. Therefore, physicians caring for these patients must establish a wide range of differential diagnoses and consider CC when managing a critical patient.[13]

Congenital heart malformations are currently significant, as they cause high mortality due to their level of complexity.[14,15] It is therefore important to highlight the relevance of determining fetal and maternal risk factors that could be involved in their development, to exercise some control over these factors, and, failing that, recognize them early so that they can be treated appropriately, ideally in the postpartum period.[14,15,16]

CONCLUSIONS

Tetralogy of Fallot is a malformation that occurs infrequently in clinical practice. Its diagnosis, through imaging studies, requires interest and extensive training from medical personnel. The treatment of choice is surgery; however, this procedure is not always possible in patients, so new therapeutic measures must be implemented

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CONFLICTS OF INTEREST

There are no conflicts of interest.

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