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RESEARCH ARTICLE

Tetralogy of Fallot: Early Diagnosis and Treatment and its Impact on Survival Rates

Fernanda Abade Lemos*, Keyla Bispo Silva, Camila Caldeira de Campos, Marcio Cerqueira de Almeida, Sérgio Roberto Molfi de Lima Filho, Taíse Santos Rocha, Emerson Pinheiro Ferreira, Wesley Alves de Souza, Talita Souza Rocha, Sabrina Andrade dos Santos, Antonio de Jesus do Nascimento, Mariana Calazans Alves, Fernanda Leopoldo Uva, Scarleth Barbosa da Silva, Jaine de Sousa Oliveira

***Corresponding author:** janeteabade01@outlook.com

ABSTRACT

Introduction: Tetralogy of Fallot affects about 3 out of 10 thousand live births, occupying a rate of 10% of all congenital cardiac malformations.

Method: this is a systematic retrospective study of bibliographic review articles, of quantitative approach and descriptive and exploratory nature, with the timeframe from 2013 to 2023.

Results: It was possible to verify that Tetralogy of Fallot is a serious congenital pathology and therefore requires surgical intervention to correct the defects present in the cardiac structure. Most bibliographic studies show that early intervention, around the first year of life, is the best treatment option and is associated with an increase in the survival rate of these individuals.

Discussion: the importance of preventive strategies during prenatal care is evident, with counseling and crucial information for mothers, aiming to provide quality care to the child at birth and to proceed with the best form of treatment, in order to reduce the mortality rate caused by this disease and proportionally increase the survival rate and life expectancy of this population.

Conclusion: given the importance of early diagnosis and treatment, it is essential that pregnant women are attended by qualified professionals who know how to identify the risk factors and clinical signs of this pathology, in addition to having easy access when necessary to perform fetal echocardiography, or more specific tests, in favor of the quality and life expectancy of these individuals.

1. Introduction

Tetralogy of Fallot (TF) is a congenital alteration caused by four main features: interventricular septal defect, obstruction of blood flow from the right ventricle, aorta in dextroposition and hypertrophy of the right ventricle. This pathology is known as the most common cyanotic congenital heart disease in newborns, resulting in a decrease in the amount of oxygen in the bloodstream, causing cyanosis, polycythemia and hypoxia as the main symptoms.[1]

According to Oliveira[2], TF affects approximately 3 in 10,000 live births, representing 10% of all congenital heart defects. In Brazil, this pathology represents an overwhelming mortality, accounting for 60 to 70% of total infant mortality, with 25% of these deaths occurring on the first day of life, highlighting the need for early diagnosis and treatment.[3]

Since it is a congenital pathology, the diagnosis of TF can be made in the prenatal period by means of fetal ultrasound and confirmation by fetal echocardiography. Although the diagnosis in the fetal period is the most indicated, because it increases the possibilities of early intervention, the discovery of this pathology in most cases is made only after birth, through clinical signs.[2]

It is noteworthy that the clinical signs of the pathology vary according to the number of cardiac anomalies, with ectopic cyanosis being the primary one, caused by right-to-left shunt, as a result of ventricular septal defect, which is reflected in the deviation of venous blood flow to the left ventricle, leading to the circulation of oxygen-poor blood.[2]

Thus, in addition to the cyanotic appearance, the cardiac alterations caused by this pathology generate more serious symptoms that can affect the life of the newborn, namely dyspnea with small exertions, causing impairment of daily activities such as breastfeeding, leading to dehydration, hypoxia, respiratory changes such as hyperpnea, lethargy, metabolic acidosis and loss of consciousness.[4]

The main intervention to treat TF is surgery. However, although it is the only effective treatment option, it has some contraindications, such as insufficient weight of the newborn, prematurity, small size of the pulmonary artery, neurological impairment or associated defects.[5]

Although medical science has evolved significantly in recent years, the ideal age for surgical repair is still controversial. In this sense, Oliveira[2], states that surgical intervention up to 12 months of age is known to be more effective than late intervention, being responsible for the reduction in mortality, but although it is associated with improvements, early treatment, in addition to the contraindications described above, can only be performed in asymptomatic patients, which limits the use of this therapy.[4]

Thus, in view of the implications described and being aware of the high level of mortality caused by this pathology, the present study aims to observe, through a systematic study, the incidence of mortality caused by this pathology, revealing how much early intervention, based on diagnosis and treatment, can influence the increase in survival rate.

2. Method

This is a retrospective review of scientific articles, of quantitative approach and descriptive-exploratory characteristic, with the timeframe from 2013 to 2023. According to Galvão[6], through this method it is possible to collect scientific articles whose researchers consider important for the description of the theme, without precisely specifying the criteria used in the review, so that it can be reproduced by other researchers. Articles indexed in PubMed and SciElo databases were used, as well as articles from electronic journals. The platform with the largest number of articles related to the central topic of the study was PubMed when searching for the term "Tetralogy of Fallot AND treatment", with 264 publications. After narrowing the search and selecting those articles that, in addition to this topic, included aspects related to early diagnosis and treatment, 20 articles remained and were used in the present study.

First, there was a selective and skimming reading of the texts to extract the information compatible with the objective of the study, tabulating the main findings in digital media. Then, a table of the articles used was created, with relevant abstracts related to the topic in question.

Detailing the methodological process better, the same occurred as follows: for the process of reading and bibliographic review was used the theory of Laurence Bardin characterized by a set of techniques of analysis of communications that aims to obtain indicators that have the knowledge relative to the conditions of production of the

message. The theory of content analysis proposed by Bardin is divided into 3 phases, namely, pre-analysis, exploration of the material and treatment of the results.

In the first phase, the organization of the material was made, proceeded with the floating reading with the initial intention of recruiting only the texts that covered the proposed objective, consequently the material was coded and explored for better understanding and later the categorization was made. Regarding categorization, all articles are attached in a folder via computer, randomly. The main facts were grouped in a separate document and then compared and analyzed. Finally, following the third phase, with the results obtained with regard to the scientific evidence on the results of early diagnosis and treatments in ET, a relationship was established between the object of analysis and the proposed content, reaching new paradigms, and it is possible from there to formulate concrete evidence based on the studies already carried out and found in the literature on the theme in question.[7]

Inclusion criteria were articles published since 2013, regardless of language, that addressed mortality and life expectancy, as well as information on the types of diagnoses and interventions performed in the population with TF. Abstracts, technical documents, books, studies that dealt with the pathology studied in a secondary way, articles accessible only to subscription, and studies published earlier than 2013 were excluded.

3. Results and Discussion

This systematic review confirmed that TF is a severe congenital pathology that requires surgical intervention to correct the structural defects of the heart. Most bibliographic studies show that early intervention, around the first year of life, is the best treatment option and is associated with an increase in the survival rate of these individuals.

Although there are some discrepancies regarding the exact age for surgical correction and some limitations, most of the articles reviewed indicate that delaying this procedure can negatively affect the health of children with TF, culminating in the appearance of complications. Thus, both the diagnosis by fetal echocardiography and the early treatment are reflected in a good prognosis.

Next, a chart has been prepared to show the main studies used to arrive at the present result (Chart 1).

Chart 1 - Summary of the articles used to construct the results. Bahia, BA, Brazil, 2023.

AUTHORS	PUBLICATIONYEAR	METHOD	HIGHLIGHTS
Campos	2014	Retrospective Clinical Data Analysis	In children with 28 months of age who underwent surgery for definitive correction, a survival rate of 84.6% was demonstrated.
Donofrio *et al.*	2014	Literature Review	One of the points emphasized was the importance of early diagnosis, still in the prenatal period, by means of fetal echocardiography.
Heinisch *et al.*	2019	Retrospective observational analysis	Data analysis of 25 children between 3.5 and 10 years of age showed that late repair of TF can be performed with a low mortality rate, but there may be complications related to growth retardation.
Kim *et al.*	2013	Retrospective chart review	It showed that definitive repair performed in infants older than 1 year was associated with an increase in mortality, and when performed before 6 months improved the recovery of right ventricular hypertrophy.
Pereira	2015	Observational study	The mean age for surgery was considered late (72 months) when compared to other studies that indicate the procedure is performed up to 12 months of age.
Silva *et al.*	2022	Descriptive cross-sectional study	The review of 104 medical records showed that most children underwent 1st cardiac surgery late (3 years). Subsequent complications (ischemic stroke, heart failure, endocarditis) are thought to be related to the late surgical approach.

Source: The authors, 2023

According to the World Health Organization (WHO), on a global scale, 6% of all births have some congenital anomaly, and every year about 295 thousand children die in the first 4 months of life due to these changes. In Brazil, approximately 80% of children diagnosed with heart disease will require surgery over time, and half of them, approximately 12,000, will require surgery in the first year of life.[8] Among the congenital heart diseases, TF is gaining prominence and depends on surgical intervention as it is considered potentially fatal.[4] Early diagnosis as well as therapeutic intervention is crucial to reduce mortality and increase life expectancy in this community.

The early diagnosis of TF can be carried out during pregnancy, based, among other methods, on fetal echocardiography, which is considered essential in the evaluation of fetal cardiac anomalies[9-10], allowing the identification of 40% to 50% of cases.[11] It is a test that should be performed in pregnant women at high and low risk for congenital heart disease. On the other hand, late diagnosis is associated with several complications, such as seizures, cardiac arrest and death, because defects involving TF directly affect the amount of oxygenated red blood cells circulating in the bloodstream[12,13].

However, before performing fetal echocardiography, it is necessary that the health care professionals do justice to the physical examination and anamnesis that should be performed at all visits during the gestational period.[14] All these approaches are considered indispensable preventive measures in the detection of congenital heart disease, and from that moment it is possible to guide the mother to maintain a healthy lifestyle, the importance of vaccinations for the baby, as well as instruct about the diagnosed pathology and the approach to future treatment.[3]

Although there are studies that address the importance of early diagnosis, Barreira[4] points out that among congenital heart disease, TF is the least diagnosed in the prenatal period, which may be related to poor visualization of cardiac structures. It is worth mentioning that, in addition to some cardiac anomalies manifesting only in the 3rd trimester, the absence of an identifiable risk factor may affect the diagnosis.[4]

Nevertheless, it is obvious the importance of preventive strategies during prenatal care, with counseling and crucial information for mothers, with the aim of providing quality care to the child at birth and proceeding with the best form of treatment to reduce the mortality rate caused by this disease and proportionally increase the survival rate and life expectancy of this population.

Regarding the best treatment option for TF, this is still a widely debated issue and early total correction or 2-stage palliation can be performed using the Blalock-Taussig technique.[15,16] Total correction is usually performed before the age of 12 months and has a low mortality rate, but is associated with late right ventricular dilatation and prolonged mechanical ventilation.[2-17]

Still, it is believed that the advantages of total surgery outweigh the disadvantages, since such an approach allows the correction of the problem in a single surgical time, shortens the right-left shunt time, reduces the length of stay of cyanosis and polycythemia, and the left ventricular volume[18]. It is considered a promising treatment as the perioperative mortality rate in uncomplicated ET is less than 5%.[19]

The goal of the Blalock-Taussig technique is to reduce hypoxemia, improve pulmonary flow, and allow time for the pulmonary artery to develop.[20] As with any procedure, some disadvantages may occur, such as pulmonary artery distortion, ischemia, pseudoaneurysm, and thrombus formation.[2] However, it is the severity of TF in relation to the clinical picture of the baby and other factors that will determine the best surgical approach.

In this sense, studies have shown that the total correction of this pathology should be performed as early as possible, since the late approach is associated with the appearance of complications and, therefore, with increased morbidity and mortality[21], whose issues were found in the review of medical records of children and adolescents aged 0 to 19 years made by Silva.[22]

A study conducted in 2013, whose technique was the retrospective review of the medical records of 326 patients with TF who underwent total correction, showed that the 15-year survival rate was 95.4% and the absence of cardiac death was 98.8%. It was also concluded that when this surgery is performed before 6 months of age, the recovery of right ventricular hypertrophy is considered better, and when performed in children older than 1 year of age, it is associated with increased mortality.[23]

Consistent with the previous study, another retrospective analysis study conducted in a neonatal service analyzed the medical records of neonates with TF who underwent surgical treatment and showed an 84% higher survival rate. This finding is consistent with most studies that suggest early treatment, as there is no benefit to delaying total repair beyond 12 months of age.[24]

Thus, as years have passed and surgical approaches to TF have improved, the life expectancy of these patients has also increased, with a 30-year survival rate of 90%, and consequently the number of adults with this pathology has increased worldwide, demonstrating the importance of health services offering this treatment.[25,26]

One study was found in the literature that analyzed data from a small group of children with TF who underwent surgery around the age of 6 years and, contrary to other studies, showed a low mortality rate. However, late treatment was associated with chronic malnutrition, hypoxia, and growth retardation.[27]

From the analysis of the mentioned facts, the importance of early diagnosis and treatment in individuals with ET was confirmed, and total correction performed in the first year of life is the most used approach and therefore considered the best strategy, since it reduces the overload of the right ventricle and consequently promotes the extinction of hypoxia, cyanosis and secondary organ damage, culminating in a reduction in mortality and an increase in survival rate.[28-30]

5. Conclusion

Among the congenital heart diseases, TF is one of the most common cyanogenic diseases in neonates and has a high mortality rate worldwide. Therefore, studies show the importance of early diagnosis and treatment in these cases to increase the survival of this population. Although there are still controversies in the literature regarding the ideal age for surgery, this systematic review concludes that early treatment is the best option, especially if it is performed in the first year of life, since the bibliographic data indicate a decrease in mortality and an improvement in survival.

The long-term effects, especially after the third decade of life, should be better studied in order to highlight the real potential problems, not only in the physical but also in the mental health of adults with TF. Given the importance of early diagnosis and treatment, it is essential that pregnant women are attended by qualified professionals who know how to identify the risk factors and clinical signs of this pathology, in addition to having easy access when needed to perform fetal echocardiography, or even more specific tests, in favor of the quality and life expectancy of these individuals.

- Declaration of competing interests

The authors have no competing interests to declare.

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