

DOWN SYNDROME AND CONGENITAL HEART PATHY: A LITERATURE REVIEW

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Abstract: The respective study is a literary review of articles, in which the main objective is to discuss knowledge about the correlation of Down Syndrome and Congenital Heart Disease. Both Down Syndrome and Congenital Heart Disease are prevalent diseases among children born and capable of generating countless consequences for these infants. The diagnosis is made clinically through the phenotypes of each child. If there is any doubt during the diagnosis, referral to a geneticist is made. The prognosis improves with each passing year, as advances in medicine allow for better treatments and better equipment capable of promoting better therapy for these children. Thus, aiming for greater knowledge on the topic, this study becomes effective.

Keywords: Down's syndrome; Congenital cardiopatics; Prognosis; Definition; Prevalence.

INTRODUCTION

Down Syndrome (DS) is a genetic determination that arises in the division of egg cells, in which three pairs appear on chromosome 21, known as trisomy 21. This disorder was recognized by the World Health Organization only in 1965, after the discovery, by Jérôme Lejeune, of the genetic cause of the pathology in question. (FREITAS et al, 2023)

Trisomy of chromosome 21 is what mainly defines DS disease, as well as other factors determining the phenotype of the child with the genetic disorder, for example, low ear implantation, elevated oblique eyelid folds, tongue protrusion, brachydactyly. Therefore, despite the external characteristics, children with DS can develop other pathologies, such as intellectual disability, congenital heart disease, speech and hearing disorders, among others that have an impact on the growth and development of affected children. (OLIVEIRA et al, 2023)

In particular, congenital heart disease consists of deformities related to the

cardiovascular system that appear during the gestation period, caused by numerous factors, such as environmental and genetic. Some of these pathologies can only be seen after birth, therefore, it is of great importance to carry out specific prenatal care, performing an obstetric ultrasound on a routine basis so that any changes can be diagnosed as quickly as possible (GOUVEIA, 2023).

Among the pathologies that affect children with DS, one of the most prevalent is congenital heart disease, which is a major determinant of both the prognosis and survival time of these patients. The main cardiac pathologies that affect this population are atrial septal defect, atrioventricular septal defect and interventricular septal defect. (CASTROPIL, 2023).

Therefore, given the prevalence and importance in the development of children with DS, it is important to discuss, evaluate and diagnose these pathologies early. Therefore, the objective of this review is to elucidate the key points that allow the association of Down Syndrome pathology with the factors involving Congenital Heart Disease.

METHODOLOGY

This article is a bibliographical review, which consulted databases with various literature, such as scientific articles published on the internet, documents made available by the Virtual Health Library, PubMed National Center for Biotechnology Information (NCBI). Research from 2009 to 2023 was used.

Thus, among the materials found, the data used came from scientific articles, books and other bibliographic sources that enabled the development of this document, with the aim of being critical and having an effective scientific basis.

The search was carried out using the keywords Down Syndrome; Congenital cardiopathy; Prognosis; Definition; Prevalence.

As a result, there was greater access to data networks in a more strategic way, since what would be searched was defined.

THEORETICAL FOUNDATION:

DEFINITION AND DIAGNOSIS OF DOWN SYNDROME

Down syndrome is a genetic disorder, caused by the presence of an extra pair on chromosome number 21, which is why it is called trisomy 21.

This dysfunction causes numerous dysfunctions in both psychomotor and neurological development, of which one of the most important consists in the intellectual delay of some part of this population, in which they respond to messages more slowly, which can generate complications in establishing and maintaining social relationships (FERREIRA, et al, 2019).

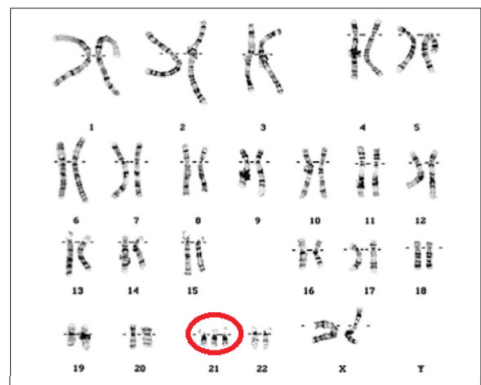


Figure 1: Trisomy 21.

Source: Brazilian Society of Pediatrics (2020)

According to the Brazilian Society of Pediatrics (2020), approximately every 800 births, 1 child has DS, regardless of other factors that may influence this genetic mutation. Therefore, it is considered the most common chromosome disorder among the human race, which can generate numerous dysfunctions in the growth and development of the affected child.

The diagnosis of this genetic syndrome is mainly clinical through observation of the stigmata of the child's phenotypes, in which the signs are often clear. The main characteristics are a flat face, increased flexion in the joints, protrusion of the tongue, reduced tone, oblique palpebral fissures, small ears and low implantation, single palmar crease (BRAZILIAN SOCIETY OF PEDIATRICS, 2020).

Therefore, the diagnosis, when based on the patient's physical characteristics, is capable of being carried out early, right after the birth of the child, the more signs the child presents, the more effective and precise, the certainty of the pathology is obtained. Referral to a Genetics specialist is only indicated if there is clinical doubt about the disorder (BRAZILIAN SOCIETY OF PEDIATRICS, 2020).

DEFINITION AND DIAGNOSIS OF CONGENITAL HEART DISEASE

According to the Brazilian Society of Pediatrics, Congenital Heart Disease (CHD) is an alteration present in the child's cardiovascular structure. There are different prognoses and types of CHD that range from fatal to simple heart diseases. Therefore, some of these pathologies can lead to death within a few hours of the child's life, due to the lack of correct blood circulation in the affected child's body (BRAZILIAN SOCIETY OF PEDIATRICS, 2022).

The diagnosis of this pathology can be carried out early, even in the child's fetal life, as long as prenatal care is carried out correctly during pregnancy and, based on evidence of changes, the tests requested are ultrasound and fetal echocardiography, among other tests capable of carrying out early diagnosis and efficient programming for this child in the postpartum period, therefore reduces the possibility of death of the newborn (BRAZILIAN SOCIETY OF PEDIATRICS, 2022).

EPIDEMIOLOGICAL PROFILE OF CHILDREN WITH DOWN SYNDROME WITH HEART DISEASE

The association of the pathologies Down Syndrome and Congenital Heart Disease is extremely important, since these diseases when correlated in the same child can cause major health problems in a relevant way, as it can reduce life expectancy and cause more consequences to the child. child development (BOAS, et al, 2009).

In a particular study carried out in Brazil, it was noted that there is a high prevalence of congenital heart disease in children with Down Syndrome. The main heart diseases visualized were Interatrial Communication and Atrioventricular Septal Defect, both, with approximately 37% prevalence in the population of children with DS (BOAS, et al, 2009).

Therefore, it is essential that all children who have the genetic disorder DS must undergo a cardiac evaluation immediately after birth, to carry out early diagnosis and implement measures that can prevent avoidable consequences for the child (BOAS, et al. al, 2009).

PROGNOSIS OF CHILDREN WITH DOWN SYNDROME WITH CONGENITAL HEART DISEASE

Children with DS already have a reduced life expectancy in relation to atypical children of their same age group, whereas children who, in addition to the genetic disorder, have associated pathologies, such as congenital heart disease, have this rate even lower, since in addition to the consequences syndrome, also have complications generated by problems with the cardiovascular system (BOAS, et al, 2009).

However, over time and with the evolution of knowledge and medical equipment, there is an improvement in this life expectancy, since the diagnosis can be made early and the stipulated treatments are more effective,

such as surgical treatment. for heart disease and measures of children's development in relation to DS (BOAS, et al, 2009).

Advances in medicine, at the current time, are mainly responsible for making changes in the lifestyle of this population, as it allows the inclusion of these patients in social environments, allows the more effective development of patients and may be able to eliminate consequences preventable for people with the syndrome (LACERDA, 2014).

FINAL CONSIDERATIONS

This article is a bibliographic review of the literature with the main objective of listing the prevalence and prognosis of patients with Down Syndrome and Congenital Heart Disease at the same time.

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