

Prevalence and types of congenital heart disease in patients with Down syndrome in a hospital in Ecuador

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Abstract

Down syndrome, also known as trisomy 21, is a common chromosomal condition associated with congenital heart disease in 40-60% of cases, leading to serious medical complications. In a descriptive, retrospective, cross-sectional study conducted at the Hospital General Puyo in Ecuador over four years, 30 patients with Down syndrome were examined to determine the frequency of each type of congenital heart disease in this population. The results showed that non-cyanotic heart defects were the most common, with atrial and ventricular septal defects being the most prevalent, followed by the atrioventricular canal. The prevalence of congenital heart disease was higher in men and patients from Pastaza. Most cases were found in preschoolers, with a higher frequency in patients older than two years. This study provides essential information on the prevalence and types of congenital heart disease in patients with Down syndrome, which may help to improve early diagnosis and treatment of these conditions in this population.

Keywords: Down Syndrome, Cardiac Malformations, Atrial Septal Defect, Ventricular Septal Defect.

Prevalencia y tipos de cardiopatías congénitas en pacientes con Síndrome de Down en un hospital de Ecuador

Resumen

El Síndrome de Down, también conocido como trisomía 21, es una afección cromosómica común que se asocia con cardiopatías congénitas en un 40 a 60% de los casos, lo que puede llevar a complicaciones médicas graves. En un estudio descriptivo, retrospectivo y de corte transversal realizado en el Hospital General Puyo, en Ecuador, durante un período de cuatro años, se examinaron 30 pacientes con Síndrome de Down para determinar la frecuencia de cada tipo de cardiopatía congénita en esta población. Los resultados mostraron que las cardiopatías no cianóticas eran las más comunes, siendo las comunicaciones interauriculares y las comunicaciones interventriculares las más prevalentes, seguidas del canal auriculoventricular. La prevalencia de las cardiopatías congénitas fue mayor en hombres y en pacientes de la provincia de Pastaza. La mayoría de los casos se encontraron en preescolares, con una mayor frecuencia en pacientes mayores de dos años. Este estudio proporciona información importante sobre la prevalencia y los tipos de cardiopatías congénitas en pacientes con Síndrome de Down, lo que puede ayudar a mejorar el diagnóstico y tratamiento tempranos de estas afecciones en esta población.

Palabras clave: Síndrome de Down, Malformaciones Cardíacas, Comunicación Interauricular, Comunicación Interventricular.



Prevalência e tipos de doenças cardíacas congênicas em pacientes com Síndrome de Down em um hospital no Equador

Resumo

A síndrome de Down, também conhecida como trissomia do cromossomo 21, é uma condição cromossômica comum que está associada a doenças cardíacas congênicas em 40-60% dos casos, o que pode levar a sérias complicações médicas. Em um estudo descritivo, retrospectivo e transversal realizado no Hospital General Puyo no Equador durante um período de quatro anos, 30 pacientes com síndrome de Down foram examinados para determinar a frequência de cada tipo de cardiopatia congênita nesta população. Os resultados mostraram que as cardiopatias não cianóticas eram as mais comuns, sendo os defeitos do septo atrial e os defeitos do septo ventricular os mais prevalentes, seguidos pelo canal atrioventricular. A prevalência de cardiopatias congênicas foi maior em homens e em pacientes da província de Pastaza. A maioria dos casos foi encontrada em pré-escolares, com uma frequência maior em pacientes com mais de dois anos. Este estudo fornece informações importantes sobre a prevalência e os tipos de cardiopatias congênicas em pacientes com síndrome de Down, o que pode ajudar a melhorar o diagnóstico precoce e o tratamento destas condições nesta população.

Palavras-chave: Síndrome de Down, Malformações Cardíacas, Defeito Septal Atrial, Defeito Septal Ventricular.

1. Introduction

Down syndrome or trisomy 21, one of the best-known chromosomal alterations and the most prevalent worldwide, has an estimated incidence of 1 in 600 to 1 in 1000 live newborns [1]. It contains features that make it a characteristic syndrome; it has been established that it has a 40 to 60% relationship with congenital heart disease. Moreover, it has been found that congenital heart disease along with respiratory infections is among the two leading causes of mortality in patients with Down syndrome in the corresponding age range of 0 to 2 years [2].

Different studies have observed that children with Down syndrome present congenital heart disease, with atrioventricular wall defects being more frequent. The early approach to these defects has given better results when they are surgically resolved. For this reason, it is essential to diagnose and treat congenital malformations promptly [2].

Congenital heart disease associated with Down syndrome is related to the genetic alteration that carries this pathology. It is an alteration that often requires surgical correction to improve prognosis and increase life expectancy in this patient. Hence the importance of early diagnosis [3]. The preferred method for diagnosing new cases of heart disease in Down syndrome is echocardiography to determine its hemodynamic impact on the patient [4].

It has been established that 8 to 10 of every 1000 live newborns have some congenital heart disease. Therefore, it has been selected as one of the most frequent defects in Down syndrome [5]. Based on the data described above, it is evident that this type of defect requires an exhaustive study to obtain real data in our environment, which will allow us to know the reality of this type of malformation. According to the study developed by the 'Manuela Espejo Mission', it has been detailed that, in Ecuador, around 7,457 people have Down Syndrome or Trisomy 215. Of these, 3597 (48.24%) are women, and 3860 (51.76%) are men [6].

Down syndrome is a frequent chromosomal alteration; this chromosomal alteration, together with cleft lip, is considered one of the leading causes of morbidity in Latin America. It can be seen that congenital heart disease in Down syndrome has a high frequency; in several studies carried out in Mexico and Colombia, a frequency of 40 to 60% has been obtained. In fact, in similar studies in different parts of Colombia, a slight tendency towards the male sex can be seen, and it is the fourth most frequent type of chromosomal alteration [7].

In Ecuador, after several studies, it has been shown that Down syndrome is present in 1 per 550 live births. This represents a prevalence rate in our country of 0.06 per 100 inhabitants. It has been established that the provinces of Manabi, Sucumbios, and Santo Domingo have the highest prevalence, with 0.09 per 100 inhabitants. In contrast, in the provinces of Carchi, Chimborazo, Imbabura, and Pichincha, it is 0.03%. No studies have been conducted in Pastaza [4]. The absence of primary data on the frequency of heart disease associated with Down syndrome in this region of the country is the reason for conducting this important study that can serve as a starting point for further research on the subject. Therefore, this study seeks to be a reference point, providing primary data for further analysis.

The methodology is presented in section 2, and the results in section 3. The discussion is shown in section 4.

2. Methodology

2.1. Type and design of research

This is a descriptive, cross-sectional, retrospective study that seeks to determine the frequent congenital heart diseases in patients with a previous diagnosis of Down Syndrome at the General Hospital Puyo, Pastaza Province, from January 2014 to May 2018.

2.2. Population under study

All patients diagnosed with congenital heart disease with Down syndrome at the Hospital General Puyo will be analyzed. This project was carried out at the Hospital General Puyo of the Ministry of Health, which has the Internal Medicine Service and, within this, the Cardiology specialty. The period to be analyzed is from January 1, 2014, to May 31, 2018.

2.3. Inclusion criteria

The entire population with a diagnosis of congenital heart disease with Down syndrome was taken.

2.4. Population size

The cardiology specialist reported the number of cases diagnosed with congenital heart disease in patients with Down syndrome at the Hospital General Puyo, Pastaza Province, from January 2014 to May 2018. No sample calculation was required because it was a small universe.

2.5. Data collection and synthesis technique

For the collection, the database provided by the Hospital Statistics Center was reviewed from January 01, 2014, to May 31, 2018. In the case of identifying the record of a patient with a diagnosis of Congenital Heart Disease and Down Syndrome, it was corroborated by reviewing medical records or by the echocardiographic diagnosis expressed in the medical history.

2.6. Hypotheses

The most frequently diagnosed congenital heart diseases in patients with Down syndrome at Hospital General Puyo are due to defects of the atrioventricular wall.

3. Results

3.1. Distribution of patients by age at diagnosis of heart disease

Congenital heart disease was more frequently diagnosed in preschoolers, with 9 cases (30%), the same number as in younger infants (Figure 1). In addition, if we add the cases reported at the school stage, 5 cases (16.66%) to the preschoolers, we have 14 cases (46.6%); that is, almost half of the cases diagnosed with congenital heart disease were described at an advanced age.



Figure 1. Distribution of patients by age at diagnosis.

3.2. Distribution of patients by sex

There is a slight inclination towards the male sex, 53.3%, compared to 46.7% in the female sex (Figure 2), so no differences could be established in this study.

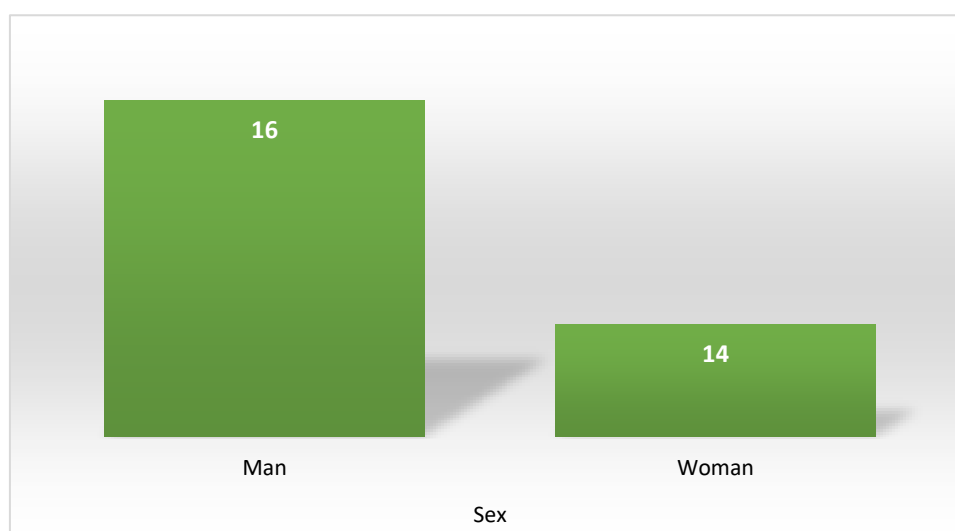


Figure 2. Distribution of patients by sex.

3.3. Distribution of patient by origin

Pastaza is the province with the highest number of cases of congenital heart disease in patients with Down syndrome, with 15 patients, i.e., 50% of the cases, due to being the location of the research center, as shown in Figure 3. In addition, Hospital General Puyo has Neonatology, Pediatrics, Genetics, and Cardiology services. In addition, it can be seen that Tungurahua is the province with the second highest number of cases, with nine patients (30%), due to its proximity to the study site. While the provinces farther away from the research site had the lowest number of cases, that is, Napo and Morona Santiago, in addition to these provinces having second-level hospitals.

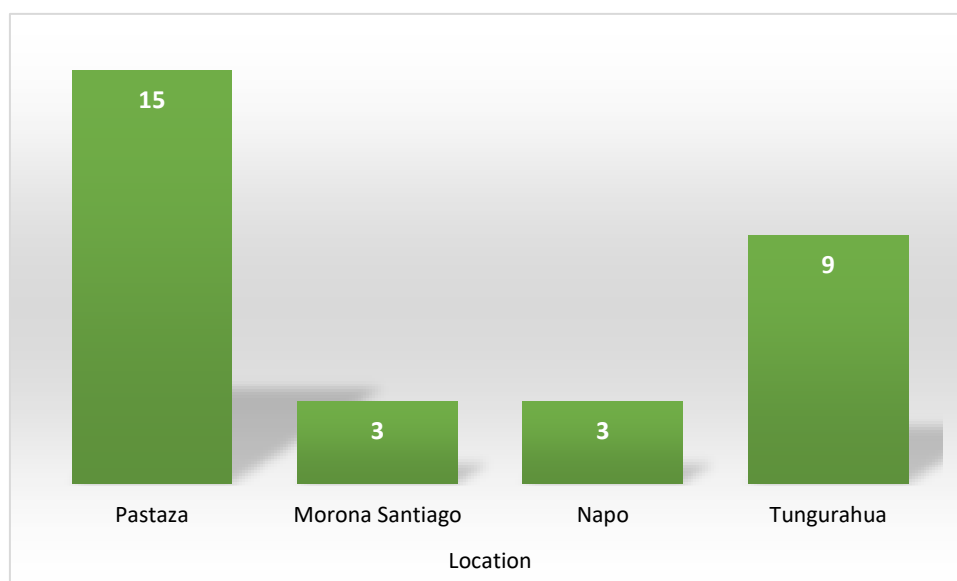


Figure 3. Distribution of patients by origin.

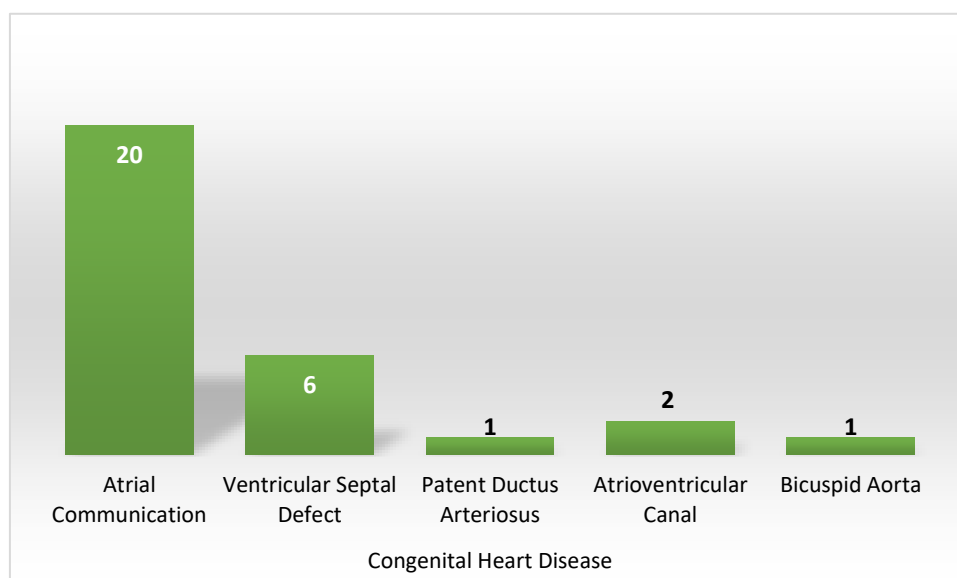


Figure 4. Distribution of patients by the variable congenital heart disease.

3.4. Distribution of patients by type of congenital heart disease

In this study, all the cases were non-cyanotic congenital heart disease. The most frequent congenital heart disease was an atrial septal defect (66.7%), followed by a ventricular septal defect (20%). This is followed by a ventricular septal defect (20%), as shown in Figure 4. Three cases of the atrioventricular canal were observed, two of which were accompanied by a ventricular septal defect and one by an atrial septal defect. In addition, there was one case of persistent ductus arteriosus and one of the bicuspid aorta.

4. Discussion

Our study had several limitations, among which the most important was the need for more specific data since the Genetics service at the Hospital General Puyo was recently incorporated. The main limitation was the data collected from all patients with Down syndrome who have attended the Hospital, which is why it was impossible to calculate the frequency of cardiopathies in children with Down syndrome. Still, obtaining the children with Down syndrome and cardiopathy from the cardiology service was possible, thus establishing the most frequent. The atrial septal defect was found first, with a relative frequency of 66.7%. A correlation can be seen with studies where it is present in 51.8%, as [5]. The non-presence of cyanotic congenital heart disease may be due to the small sample size presented in this study.

Other studies found that the most frequent non-cyanotic congenital heart disease associated with Down syndrome is a ventricular septal defect (61%), as reported [8]. Similar studies show ventricular septal defect as the most frequent cardiopathy, such as the study by [7], where 61.74% is obtained. The present study showed ventricular septal defect as the second most frequent congenital heart disease associated with Down syndrome, with a relative frequency of 20%. This was followed by 3 cases of the atrioventricular canal, one of patent ductus arteriosus and one of the bicuspid aorta. All the pathologies found in this study belong to non-cyanotic congenital heart disease. According to the bibliography consulted, we can demonstrate that in the Hospital General Puyo, we have not found concordance of the most frequent congenital cardiopathies with the rest of the studies since the atrioventricular canal is described as the most frequent, representing up to 80% of the cardiopathies, as it is mentioned [2].

Twenty-five cases were found with single heart disease; in 5, more than two heart diseases were found in the same patient. Regarding sociodemographic factors, the male sex was slightly predominant with an absolute frequency of 16 cases (53.3% relative frequency), compared to 14 cases in the female sex, corresponding to a relative frequency of 46.7%, so there was no evidence of a predominance of one sex over the other. These data do not agree with several studies where 52.5% were observed for the female sex. Therefore, it has not been possible to establish differences between one sex over the other [9].

The ages at which these heart diseases were diagnosed were divided into age groups in which preschoolers and young infants were among the most frequent, with 9 cases corresponding to 30% of relative frequency for each. There is evidence of a problem in the echocardiographic screening of the different congenital heart diseases associated with Down syndrome; if we add the cases found in the preschool and school stages, obtaining about 14 cases, corresponding to a relative frequency of 46.6%, This means that almost half of the diagnosed cases of congenital heart disease were described at an advanced age, showing a late diagnosis of this type of heart disease since it is recommended that children with Down syndrome be followed up by cardiology before the age of one month and timely surgical correction before six months of life [10].

Concerning the place of origin of the patients, there have been no previous studies on this variable in our environment; however, with the present study, we found that in the province of Pastaza, there is a more significant number of cases of congenital heart disease in patients with Down syndrome with a relative frequency of 50%, due to the better structural organization of the General Hospital Puyo, which has a direct interrelation between the Neonatology, Pediatrics, Genetics, and Cardiology services. As for the other

provinces of residence, it can be seen that the province of Tungurahua is the second with the second highest number of cases; we could explain this data due to the geographical proximity that exists between these two provinces and the fact that the hospital becomes a point of reference in terms of echocardiographic screening of this type of malformations in pediatric patients. An attempt was made to perform bivariate analysis considering the variable heart disease versus sex, age, place of origin, and type of heart disease. However, statistical significance was not found in the results, probably due to the study's small sample size. The present study can guide further research to estimate the incidence and prevalence of patients with congenital heart disease and Down syndrome in this country's region [11], [12].

5. Conclusions

Congenital heart disease associated with Down syndrome is highly frequent in our environment, among which the most frequent type is non-cyanotic. No cases of cyanotic congenital heart disease were recorded due to the small sample of our study. Among the sociodemographic variables of the patients with congenital heart disease diagnosed with Down syndrome, the majority were male, without denoting a marked difference; more studies have been found that support the higher frequency in the female sex. The most frequent age range in which the diagnosis was made was in preschoolers, the diagnosis and surgical correction of these pathologies are recommended before six months of age, so we think these patients should have been approached earlier. Of all the patients diagnosed, 50% were from Pastaza, while the rest came from other provinces; this variable has not been studied in other studies.

The most frequent congenital heart disease in patients diagnosed with Down syndrome is an atrial septal defect, followed by a ventricular septal defect; this is associated with the bibliography consulted in Latin American studies but is not related to the American studies name atrioventricular canal as the most frequent defect. Three cases of the atrioventricular canal have been found, all of which are non-cyanotic congenital heart defects. Only 5 cases presented two concomitant heart diseases, while the majority (25 cases) showed a single heart disease. Our study had several limitations, including the lack of specific data collection. The main limitation was the data collected from all the patients with Down syndrome who have attended the Hospital, which is why it was impossible to calculate the frequency of heart disease in children with Down syndrome. Still, obtaining the children with Down syndrome and heart disease from the cardiology service was possible, thus establishing the frequency of each heart disease and determining the most frequent ones.

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Institutional Review Board Statement

Not applicable.

Informed Consent Statement

Informed consent was obtained from all subjects involved in the study.

Conflicts of Interest

The authors declare no conflict of interest.

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