

PHYSIOTHERAPEUTIC INTERVENTION IN EARLY STIMULATION IN PATIENTS WITH DOWN SYNDROME WITH ATRIOVENTRICULAR SEPTAL DEFECT

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Abstract: Introduction: Down syndrome is the most recurrent chromosomal abnormality in humans and the most predominant cause is intellectual disability. However, people with DS have a great ability to develop through extensive neuroplasticity that can be stimulated from the first months of life by family, health and education professionals. Furthermore, it may be associated with congenital heart disease, visual changes, hormonal changes and changes in typical neuropsychomotor development. They must be monitored from birth by a pediatrician who will be responsible for requesting exams, in accordance with current clinical assessment, in order to receive appropriate treatment and develop within their own time. **Methods:** This study presents itself as a literature review based on publications indexed in the VHL, PubMed, LILACS and SciELO Brazil databases. **Results:** Regarding the quantity of the research, 488 articles were found, they were screened according to exclusion and inclusion aspects, and finally 20 articles were used in this study. **Final Considerations:** In the research used to construct this article, it was observed that physiotherapeutic intervention in early stimulation in patients with down syndrome and atrioventricular septal defect was beneficial for motor and cognitive development when stimulated during and after physiotherapy with parents and family members.

Keywords: Down syndrome, early stimulation, physiotherapy, atrioventricular septal defect, neuropsychomotor development, congenital heart disease.

INTRODUCTION

Down Syndrome (DS) is a way of living life and is characterized by a genetic alteration, the presence of an extra chromosome, trisomy 21. It is a syndrome that presents signs of muscular hypotonia, ligament laxity and muscle weakness. Furthermore, DS allows for an increase in the prevalence of other diseases, including changes in the heart, vestibular system, gastrointestinal system, endocrine system, musculoskeletal system, respiratory system and nervous system. (SANTOS et al., 2022).

PEREIRA et al. (2021), claims that according to the World Health Organization (WHO), down syndrome is the most common chromosomal deformity in live births, with a prevalence of 1:1000 worldwide. In Brazil, there is a birth of a child with DS every 600 and 800 births, regardless of ethnicity, gender or social class. Furthermore, the MINISTRY OF HEALTH. (2013), the life expectancy of children with DS increased considerably from the second half of the 20th century, due to advances in medicine and progress in the health sector due to heart surgery.

In agreement with Santos et al. (2020), babies with down syndrome present a severe motor delay that may be related to their large body restrictions, such as joint hypermobility and muscular hypotonia that is apparent soon after birth. These characteristics prevent movements from being carried out and consequently harm the entire development of the body scheme. The body schema is the global, scientific and differentiated image of one's own body, and is essential for the baby's psychomotor development. These restrictions are responsible for slow movement, low postural control, and changes in the vestibular, somatosensory, visual and proprioceptive systems are responsible for motor delay, as hypotonia makes it difficult to explore the environment and gain experiences.

According to Sotoriva and Segura (2013), they state that one of the possible causes of motor deficits in individuals with DS can be understood as defects in the morphology of synapses and in the recycling of vesicles at the neuromuscular junction. Therefore, the sooner a child's motor development delay is diagnosed, the better their chances of receiving effective treatment will be, which will offer less risk of developing disabilities throughout their lives.

According to Dias et al. (2017), mentions that those with DS who have congenital heart disease, half have atrioventricular septal defects (AVSD), which causes high mortality in these patients and only 2% have it in isolation. It is usually associated with atrial septal defect (ASD), ventricular septal defect (VSD) and patent ductus arteriosus (PDA). Furthermore, studies show that individuals with Down syndrome may present early pulmonary structural changes (microvascular changes) and pulmonary hypertension, due to blood flow from left to right, which leads to intrapulmonary hyperflow and worsening of pulmonary hypertension.

According to Rezende (2014), atrioventricular septal defect is a coronary disease associated with intracardiac shunting of blood, which can often lead to irreversible changes in pulmonary vascular resistance and develop pulmonary arterial hypertension if not repaired. Patients with trisomy 21 are at high risk of early development of pulmonary arterial hypertension if it is not corrected early.

According to the Newborn Health Care Manual (2014), septal defects have the main characteristic of left-right shunt and pulmonary hyper flow. The clinical picture is characterized by tachydyspnea and heart failure that occur due to the large left-right shunt and pulmonary hyperflow, without any degree of cyanosis. Symptoms become more abundant after the 2nd week of life, when

pulmonary vascular resistance is lower and the magnitude of pulmonary flow increases. On physical examination, hyperphosis of the 2nd bulba in the pulmonary focus and a heart murmur are normally observed. It presents normal peripheral saturation (greater than 95%) and, in general, does not present signs of low systemic output. The chest X-ray image shows an enlarged cardiac area and an increase in the pulmonary vascular network.

Research shows that babies with Down syndrome have considerable motor delay when compared to typical babies. This delay directly affects independence, the child's exploration of the environment and the achievement of motor milestones in the corresponding periods. The acquisition of these skills in DS follows the same order as typical babies, however these skills occur later. (SANTOS et al., 2020).

There is evidence that early perceptions about changes in neuropsychomotor development, as well as early intervention, benefit the child and family, including developing their motor skills that can prevent cognitive, behavioral, educational and social disorders. As therapeutic programs are essential in this intervention, and states RIGONI et al. (2022), which must be started as early as possible to be functionally effective. Still, the process becomes more successful when there is a support network that involves training the parents with the therapist to instruct them and thus develop early stimulation daily at home with the infant.

According to Torquato et al., (2013) mentions that, physiotherapeutic treatment for patients with down syndrome is more focused on developing proposals that are in accordance with their needs and problems related to postural adjustments and delays. motors in which sitting and standing are mainly affected. Therefore, physiotherapy is recommended to carry out gait training,

transpostural changes, static and dynamic balance using techniques and resources that are suitable for being carried out on the ground.

Therefore, the present work aims to explore the main evidence in the literature on the physiotherapeutic intervention of early stimulation in patients with Down Syndrome with atrioventricular septal defect.

METHODS

This is a literature review of a qualitative, quantitative and exploratory nature. A search was carried out in sources of information in health and multidisciplinary areas, namely: SCIELO Brasil, VHL - LILAC's and PUBMED, using the keywords (which were selected through the Health Sciences Descriptors (DeCS)), being them: down syndrome, early stimulation, physiotherapy, atrioventricular septal defect, neuropsychomotor development and congenital heart disease; with the combination of Boolean operators (AND, OR and NOT) and truncation (*); The search process took place according to figure 1. The following inclusion criteria were considered: availability of text, studies available in Portuguese, Spanish and English and that were published in the period between 2013 and September 2023 and as criteria for exclusion, the following were used: texts that presented information that was not related to physiotherapeutic interventions in early stimulation in patients with down syndrome with atrioventricular septal defect and duplicate articles in the databases.

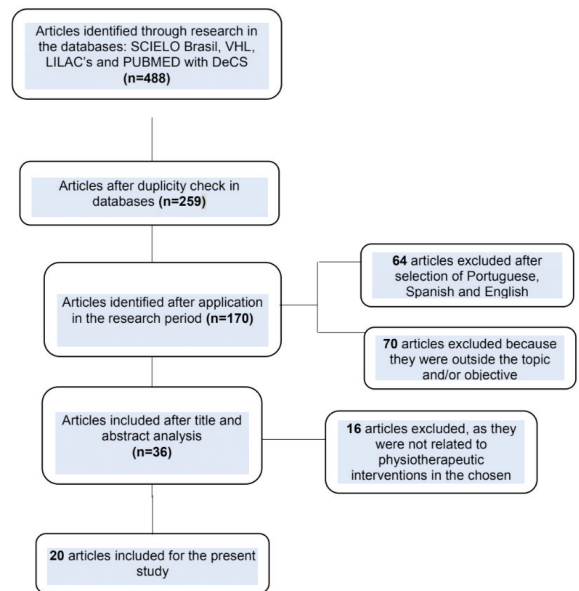


Figure 1: Research flowchart

SOURCE: Elaborated by the author, 2023

RESULTS

The search resulted in 488 articles, which were screened according to exclusion and inclusion aspects, and finally 24 articles present in this study were selected.

DISCUSSION

In the study by Danielli et al. (2016), it was observed that the intervention program favored prone, sitting and standing postures as babies are encouraged and encouraged to handle the objects around them. The prone position is essential in this development as it forms the anti-gravity muscles for sitting and standing positions and consequently for rotations and coordination of movements. The authors, Ramos and Muller et al. (2020) state that there is a delay in reaching motor milestones after 12 months of age in children with DS, as during this period there is a greater number of tasks that are required of the nervous system in this age group, in addition to motor and cognitive skills, language maturation also occurs, which demands greater speed in the progress of these milestones.

The study by Almeida, Moreira and Tempski. (2013), highlighted the role of physiotherapy in early stimulation in the different stages of the life of an individual with DS, ranging from zero to eighteen years old. However, it is of great importance to recommend that the sooner family members seek to undergo physiotherapy, the greater independence and functionality will bring to this patient. However, the study by Gonçalves (2016) demonstrates in its results that parents attach great importance to early intervention, but state that public institutions do not have available and adequate services to offer, making it necessary to resort to private therapies, reducing the possibility of parents with low resources to have dignity in these services and compromising the future of their children.

According to Hoepers, Schenkel and Schivinski. (2013), the process of growth and development are products of internal and external factors. Among these, external aspects deserve to be highlighted, related to the quality of the environment where the infant is located, including basic nutrition, body hygiene and the care offered by family members, in addition to socioeconomic, affective and psychological conditions. Furthermore, Amaral, Corrêa and Aita (2019), add that the development of congenital heart disease patients is preceded by low cardiopulmonary performance to perform daily tasks, hypoxemic crises are common and may be accompanied by crying and everyday situations that require physical effort such as playing and expressing oneself.

According to Mourato, Villachan and Mattos. (2014), confirm in their study that only a minority of patients were referred for investigation of possible congenital heart disease before six months of age, preventing early diagnosis. This can make cardiac surgery difficult and evolve into

pulmonary hypertension, as it can lead to surgical intervention contraindicated due to pulmonary hyper resistance, increasing mortality in these individuals. The work of Trevisan et al. (2013) reiterates, regarding the relationship between chromosomal disorders and the types of heart defects, there is a combination with atrioventricular septal defect, especially in patients with DS (a frequency of 66.7%). This is consistent with literature findings that describe rates of 40-50%.

As Souza (2016) demonstrates in his sample that motor milestones such as (crawling and walking) that were not achieved by the participants, both for children with heart disease (CHD) and non-cardiac disease, ended up not being an impediment as the participants found ways to move independently of these mentioned. Although, specific motor milestones were not taken into consideration. Melo (2020), details in his research that mothers who received prenatal care in the private sector, many of them received a diagnosis of their child with DS shortly after birth. However, it was not possible to state that the reason for the late diagnosis was the lack of access to more complex exams, however, it is understood that the late diagnosis could be due to the lack of follow-up with the obstetrician or fear of undergoing invasive exams that could jeopardize risk to the baby's health.

NUMBER	AUTHORS/ YEARS	TITLE	MAIN RESULTS
1	Danielli et al.(2016).	Effects of an early motor intervention program on the development of babies in a residential shelter	The studies showed that the intervention program had better post-intervention results in the prone and seated positions as babies are stimulated and encouraged to handle objects. The prone position was fundamental in development, as it prepares the anti-gravity muscles for sitting and orthostasis for waist rotation and coordination of movements.
2	Ramos e Müller. (2020).	Motor and Social Milestones of Children with Down Syndrome in early stimulation	In this study, the motor milestones that were most delayed according to the child's age group were characterized. It was observed that children with DS can acquire motor skills at the same rate or very close to what is expected for typical development. However, this process can take up to twice the estimated time for the skills to progress properly.
3	Hoepers, Schenkel e Schivinski. (2013).	Heart disease and motor development in Down Syndrome: case series	Children with DS and congenital heart disease showed motor development similar to that of children without heart disease, with factors such as the father's education, regular financial status and housing conditions influencing this result.
4	Almeida, Moreira e Tempiski. (2013).	The Contributions of Physiotherapy in Down Syndrome in children: An integrative review	The research reports the use of stimulation in various stages of the life of an individual with Down Syndrome and shows how physiotherapy, together with the multidisciplinary team, is important to reestablish functionality in these individuals.
5	Amaral, Corrêa e Aita.(2019).	Profile of independence in self-care of children with Down syndrome and congenital heart disease	In this research, it was found that the functional performance in self-care activities of children with DS and CC are lower than that of children with typical development.
6	Mourato, Villachan e Mattos. (2014).	Prevalence and profile of congenital heart disease and pulmonary hypertension in Down syndrome in a pediatric cardiology service	The prevalence of congenital heart disease in DS patients was higher in the studied service compared to other studies, which can be explained by the service being a reference. Still, the low percentage of referral up to six months reinforces the need for better tracking of DS patients. This conduct becomes imperative when considering the high frequency and progression to PH in these patients.
7	Trevisan et al.(2013)	Chromosomal Abnormalities among Patients with Congenital Heart Disease	Regarding the relationship between chromosomal disorders and the types of heart defects, there is a very significant association with atrioventricular septal defect, especially in patients with Down Syndrome. Indices in the literature describe 40-50%, therefore there is a high probability that when we are faced with a patient with an atrioventricular septal defect that he will have DS (about one in two).
8	Souza. (2016).	Comparison of Motor Behavior between children with Down syndrome with heart disease and those without heart disease after specific physiotherapeutic intervention	The study allowed the comparison of motor behavior between children of the same chronological age with DS and CHD and children with DS without CHD. After specific and individualized physiotherapeutic intervention in these children, there was a distinction between the gain of new motor skills, being greater in the group of individuals with heart disease, showing that these children, despite showing an even greater motor delay when compared to participants without CHD in the pre-intervention period, may improve their motor skills gradually when they receive adequate physiotherapeutic intervention.
9	Melo.(2020).	Congenital heart disease in children with down syndrome	The results of the present study were similar to those found in the literature, as 60% of the sample had congenital heart disease, the most common being ASD, PDA, AVSD and murmur.
10	Gonçalves. (2016).	The importance of early intervention in children with Down Syndrome	The levels of early intervention as resources are scarce and not all children have free access, having to resort to private institutions.

TABLE 1: Articles related to the role of physiotherapy in early stimulation in children with Down Syndrome with atrioventricular septal defect

SOURCE: Elaborated by the author, 2023.

FINAL CONSIDERATIONS

In this study, the importance of physiotherapeutic intervention in early stimulation in individuals with DS and atrioventricular septal defect was observed in being assisted and undergoing therapy and heart disease correction as soon as possible so as not to progress to a more serious condition. Furthermore, the recommendation and lack of assistance in public hospitals leads less fortunate parents to endanger the future health of their children and delay neuropsychomotor development.

However, in this study it was noted how significant early stimulation and family participation are in the evolution of the DS patient's condition in reaching motor milestones and leading a healthy life with the highest possible quality of life. Likewise, their interaction with internal and external environments was observed to be potentially influential in their way of living. Therefore, early diagnosis and medical monitoring from prenatal care are essential, as the sooner it is started, the greater its neuronal maturation will be through neuroplasticity.

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