

# EARLY TREATMENT OF OROFACIAL ALTERATIONS WITH PHYSICAL THERAPY AND PALATAL PLATE IN CHILDREN WITH DOWN SYNDROME

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## ABSTRACT

Down syndrome (DS) or trisomy 21 is the most frequent autosomal congenital anomaly, with a worldwide prevalence of 10/10,000; in Chile, the prevalence is 2.5/1,000 live births. People with DS present an open mouth with increased salivation, an everted lower lip, elevated and inactive upper lip, and lowering of the angle of the mouth on account of growth and development disorders, and muscular hypotonia.

**Objective:** To evaluate the literature on the early treatment of orofacial alterations in children with DS to prevent or minimize them. **Method:** Search in PubMed and Scielo databases, regardless of the year of publication or language; 26 papers were selected. **Results:** There are positive effects on the orofacial motor function, observing significant changes in the most severe cases. **Conclusions** A child with DS can benefit from early treatment, but there are insufficient comparable studies in terms of duration, age and type of therapy. **Keywords:** Down Syndrome, malocclusion, physical therapy modalities.

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## RESUMEN

El Síndrome de Down (SD) o trisomía 21, es la anomalía congénita autosómica más frecuente cuya prevalencia mundial es de 10/10.000, en Chile de 2,5/1.000 nacidos vivos. Por trastornos de crecimiento, desarrollo e hipotonía muscular, las personas con SD presentan boca abierta con gran salivación, labio inferior evertido, elevación del labio superior en inactividad y descenso del ángulo de la boca.

**Objetivo:** Evaluar la literatura sobre el tratamiento temprano de alteraciones orofaciales en niños con SD para prevenirlas o minimizarlas. **Método:** Búsqueda en base de datos PubMed y Scielo, sin discriminar año de publicación, idioma: inglés-español, seleccionando 26 artículos. **Resultados:** Existen cambios positivos en la función motora orofacial, observando mayores cambios en los casos más severos. **Conclusiones:** Un niño con SD se beneficia del tratamiento temprano, pero faltan estudios comparables en cuanto a duración, edad y tipo de terapia. **PALABRAS CLAVE:** Síndrome de Down, Maloclusión, Modalidades de terapia física.

RESUMO: A síndrome de Down (SD) ou a trissomia do cromossomo 21, é a anomalia congênita autossômica mais frequente, com prevalência global de 1 / 1.000, no Chile de 2.5 / 1.000 nascidos vivos. Devido aos distúrbios de crescimento, desenvolvimento e hipotonia muscular, as pessoas com SD apresentam boca aberta com grande salivação, lábio inferior evertido, elevação do lábio superior inativo e ângulo da boca mais baixo.

Objetivo: Avaliar a literatura sobre o tratamento precoce dos distúrbios orofaciais em crianças com SD para preveni-las ou minimizá-las. Método: Pesquisa na base de dados PubMed e Scielo, sem discriminar ano de publicação, idioma: inglês-espanhol, selecionando 26 artigos. Resultados: Existem mudanças positivas na função motora orofacial, observando-se grandes alterações nos casos mais graves. Conclusões: Uma criança com SD se beneficia do tratamento precoce, mas faltam estudos comparáveis em termos de duração, idade e tipo de terapia. PALAVRAS-CHAVE: Síndrome de Down, má oclusão, modalidades fisioterapêuticas.

#### Authorship contribution

- a) Conception and design of study.
- b) Acquisition of data.
- c) Data analysis.
- d) Discussion of results.
- e) Drafting of the manuscript.
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## **INTRODUCTION**

Down syndrome (DS), also known as trisomy 21, trisomy G or mongolism, is the most frequent autosomal congenital abnormality. Approximately 95% of cases with DS are due to an extra chromosome 21, i.e., they have 47 chromosomes instead of 46. The other 5% is caused by other chromosomal abnormalities: 3% due to translocation and 2% due to mosaicism or partial trisomy.

Esquirol provided the first description of a child who allegedly had trisomy 21 in 1838; eight years later, Seguin described a patient with characteristics suggesting an abnormality, which would later be known as DS. In 1866, English physician John Langdon Down published an article that accurately describes some of the characteristics of this syndrome, which today bears his name <sup>(1)</sup>.

DS or trisomy 21 is the most common chromosomal disorder associated with intellectual disability, accounting for 10% of all cases of this disability. The degree of intellectual disability can vary widely: the average IQ is 50, ranging between 20 and 80 <sup>(2)</sup>.

The global prevalence of DS is 10 per 10,000 live births. However, there are major differences between countries, and these depend mainly on sociocultural variables. In countries where abortion is illegal, such as Ireland or the United Arab Emirates, the prevalence is higher: between 17 and 31 per 10,000 live births; in the United States 1 in 732 babies, while the prevalence rate is 1.08 per 1,000 live births in the United Kingdom. In France, on the other hand, prevalence drops to 7.5 per 10,000, perhaps due to the high abortion rate (77%) <sup>(3)</sup>.

In Latin America, DS prevalence rates have increased from 1974 to 2005. The International Clearinghouse for Birth Defects Monitoring Systems recorded a rate of 1.46 per 1,000 births in 1974-1979, with a peak of 1.85 in 1998. Besides, the Latin American Collaborative Study of Congenital Malformations reported a global rate of 1.88 for 1998-2005. Below this average, we find Brazil (1.72), Colombia (1.72), Bolivia (1.55), Venezuela (1.49), Ecuador (1.48) and Uruguay (1.32). Above this average, we find Argentina (2.01), Paraguay (1.98) and Chile, which has the highest rate in South America, with 2.47 per 1,000 live births <sup>(3)</sup>.

Prevalence has no racial, socioeconomic or gender predominance, but increased maternal age is associated with a greater likelihood of having a child with trisomy 21. At age 35, the risk is already higher, and it becomes considerably higher after age 45, reaching a rate of 1 in 25 live births <sup>(4)</sup>.

DS has various characteristics, alterations and/or clinical manifestations, both systemic and craniofacial, expressed individually in different degrees. The systemic abnormalities that appear are cardiovascular, hematopoietic, nervous, and musculoskeletal, among others. This is why from birth, all children with DS must undergo multiple examinations and assessments to detect certain alterations in time, and throughout their growth, they must attend frequent medical and dental appointments <sup>(5-6)</sup>.

People with DS have a distinctive phenotype, which is partly due to growth disorders, bone development, and generalized muscle hypotonia <sup>(6-7)</sup>. They have multiple genetically determined orofacial disorders, as well as varying degrees of dysfunction of the stomatognathic system <sup>(4)</sup>. Some of these features are small skull facies, midface and nasal bone depression, flat malar processes and upward slanting eyes. They also have a short, flat cranial base, reduction of maxillary length or maxillary hypoplasia and midface retrusion. An Angle Class III skeletal pattern <sup>(8-9)</sup> is generally described; however, many aspects of craniofacial morphology remain unclear, such as detailed anatomy of the mandible (body, ramus, and chin) and alveolar dimensions. Therefore, some authors have described the mandible as small, while others have found that it is similar to that of the population not affected by the syndrome <sup>(10-12)</sup>.

Castillo Morales classifies the pathological disorders suffered by patients with DS into primary and secondary. Primary disorders involve hypotonia of the orofacial muscles, decreased tone of the ligaments of the temporomandibular joint (TMJ), protruded hypotonic tongue with midline diastasis, and weak lingual frenulum. Midface decreased by maxillary hypoplasia, reduced palate height, hypotonic velum, delayed dentition, microdontia and agenesis. Secondary pathologies occur due to malfunction of the oral and respiratory structures and include subluxation of the mandible, mouth breathing, respiratory tract infections, malocclusions, and phonation problems. These pathologies include a characteristic open mouth with increased salivation, an everted lower lip, elevated and inactive upper lip, and lowering of the angle of the mouth. In addition, the constant tongue

protrusion does not allow for proper mouth closure and causes a dry, cracked lingual surface, and the upper and lower incisors protrude, sometimes causing an open dentoalveolar bite <sup>(13-15)</sup>.

Castillo Morales' orofacial regulation therapy (ORT), developed in 1978 in Munich, Germany, seeks to improve several of the primary and secondary pathologies common to children with DS. The therapy includes two components: the first is manual physical stimulation of the orofacial muscles, generally provided by a physical therapist, and the second is the insertion of a stimulating, removable palatal plate, as needed. The palatal plate is designed to modify the resting position of the tongue, causing specific tongue movements, increasing upper lip mobility as well as the tone of the facial muscles <sup>(16)</sup>, leading to improvements in mouth closure, suction, phonation, swallowing, and nasal breathing <sup>(4-15)</sup>.

Given the above, many factors favor malocclusion in children with trisomy 21. Therefore, since they are young and especially during growth and development, they should receive multidisciplinary dental care to evaluate the sequence of expected abnormalities that can be prevented or to intervene in each case. Orthopaedic and orthodontic treatment from early life to adulthood can correct and control functional abnormalities, as well as the development of dental and facial structures <sup>(4-17)</sup>.

This review aims to evaluate the literature on early treatment with physical therapy and palatal plate in children with DS to prevent or minimize orofacial disorders.

## **METHODOLOGY**

A scientific literature review was conducted following a structured search methodology in PubMed and Scielo. The terms used for the search were "Malocclusion down syndrome", "Orthodontic treatment down syndrome", "Early orthodontic down syndrome", "Stimulating plate down syndrome", "prevention malocclusion down syndrome"; which were combined; 180 papers were obtained. Articles in English and Spanish were included, regardless of the

year of publication. In addition, the following papers were excluded: repeated papers, those referring to treatments for adults and those of little relevance to the subject matter.

Thus, 26 papers were considered, of which 10 include the evaluation of early treatments in children with DS in order to prevent or minimize orofacial alterations, especially orofacial regulation therapy with palatal plate. In the following stage, the papers were read critically, and the information was summarized and analyzed.

## **DEVELOPMENT**

Castillo Morales Orofacial Regulation Therapy (ORT) is the basis of studies that seek to evaluate early treatment alternatives in children with DS to prevent and/or decrease orofacial alterations, such as abnormal tongue and lip position. Later studies developed and evaluated treatments inspired by ORT, but with modifications. They had in common the use of physical therapy, and the main variant was the type of plate used (as indicated). Therefore, it is more accurate to call it early treatment with physical therapy (ETPT) or early treatment with physical therapy and palatal plate (ETPTP).

The indication of the use of a stimulating palatal plate focuses on patients with: a) a broad hypotonic tongue and which is in an interdental or interlabial position for several hours a day; b) a tongue with diastasis and additional protrusion; c) an inactive hypotonic upper lip with narrow sides <sup>(15-16,18)</sup>.

As early as 1991, a study involving 67 children with trisomy 21 evaluated the results of ETPTP, reporting positive changes in the tongue and lip position, with improvements in motor function in actions such as suction and saliva control. All this was observed after one year of treatment, using the plate 1 hour a day in the beginning, and then progressing to 3 or 4 hours a day <sup>(15)</sup>. With the same treatment but only after 10 months, improvements in mouth closure were observed. The clinical examination and the parent survey showed that the mouth remains open for less time compared to the beginning of the therapy, that there were

noticeable improvements in tongue position, which was no longer outside the mouth or between the teeth most of the time <sup>(18)</sup>.

A 2000 study attempted to increase the evidence having a control group, which allowed researchers to evaluate and analyze the results in a different way than what the literature showed up to that time. The study recognized this lack as a justified weakness in ethical issues <sup>(15)</sup>. They showed that after 2 years of treatment, there were considerable improvements in lip closure in 65% of the individuals under study, versus 8% in the control group that only underwent physical therapy. Something similar occurs with tongue position, which improved in 55% of children in the study group compared to 30% in the control group <sup>(19)</sup>. Carlstedt published similar results from a study of the same type, but with treatment that lasted for 4 years. They observed positive effects on oral motor function and significant improvement in the following variables: lip activity, tongue position and facial expression <sup>(20-21)</sup>. These results are very similar to those obtained with shorter treatments, such as those conducted by the same author 9 and 12 months after the initial examination <sup>(22)</sup>. In agreement with these results, a 2004 study followed the same protocol as previous research (physical therapy plus palatal plate), finding that after one year of treatment there are changes in mouth position were observed in 75% of a total of 20 children under study, and also that 65% showed improvements in tongue position <sup>(23)</sup>.

The above studies repeatedly evaluate changes in lip position, mouth closure and tongue position. However, other studies further evaluate other relevant aspects, such as habit persistence, tooth eruption, occlusion and speech development <sup>(20-21,24)</sup>. Regarding tooth eruption, Bäckman et al. showed that patients treated without palatal plate had fewer erupted teeth than children treated with this device and than children without DS. This might be due to stimulation of the oral mucosa with the use of palatal plates, thus accelerating tooth eruption <sup>(24)</sup>. The use of ETPTP from 6 to 48 months of age (modifying the plate depending on the child's development) may prevent alterations in tooth occlusion. The posterior crossbite is significantly lower in subjects undergoing early treatment than in individuals in

the control group <sup>(24)</sup>. Likewise, one study reports changes in the speech of children undergoing this treatment: their abilities are three times better than in untreated children <sup>(24)</sup>, in contrast to no favorable results shown in studies of the same type <sup>(20-21)</sup>.

In Chile, Padró et al. conducted a study in 2010 after applying plate treatment for two months. This term was very limited in relation to what was suggested by some authors <sup>(22)</sup>. Padró et al. conducted weekly monitoring and observed minor and slightly favorable changes in tongue tone, mouth closure, tongue retraction and lip position, in a sample of 5 children with DS aged 15 months on average <sup>(25)</sup>.

The age at which it is advisable to start treatment must also be considered, as experiences vary widely in this regard. Treatments starting at only 1, 2, or 3 months of age have shown good results in tongue position, lip contact, and mimetic expressions <sup>(4,20,23)</sup>, even though the cooperation of such a young infant may seem complex. At the other end, there are cases where treatment started as late as age 5 <sup>(22-23)</sup>. It is essential to consider that some studies included highly uneven ages, the differences among them reaching 30 months <sup>(21)</sup> or 59 months <sup>(23)</sup>. An analysis of variance determined that the results show no age-related effects <sup>(15)</sup>.

Castillo Morales states that tooth eruption hinders the therapy because the device retention methods are harder to implement. Therefore, interrupting the treatment during that period until total tooth eruption is achieved seems to be an alternative with good results when treatment is started before this stage of development <sup>(18)</sup>.

Long-term studies that evaluate and appraise the stability of the results after several years of therapy are even scarcer. The positive effects found at the end of treatment improved regarding the usual mouth position, increasing the time it was kept closed, and the usual tongue position, increasing the time it was kept inside the mouth <sup>(18)</sup>. Korbmacher found that 12 years after the end of the treatment, mouth position improved in 30% of children, it remained unchanged in 10% of cases, and it deteriorated in 45% of them. However, changes

since the start of treatment include 55% improvements in this item, and 40% of cases remain unchanged. Regarding tongue position, changes from the end of therapy to the check-up after 12 years show a positive effect in 20% of children, same result in 60% of cases and 5% of cases with deterioration. There were positive changes from the beginning of the treatment until the check-up after 12 years in 75% of individuals, and the condition remained the same in 25% of cases, with no adverse effects <sup>(23)</sup>.

## **DISCUSSION**

Given the high variability of the elements that make up the ETPTP, it is impossible to attribute specific effects to each component. However, the isolated insertion of a stimulating plate without additional physical therapy is not indicated. In fact, none of the papers reviewed, in which early treatment was performed, reported the exclusive use of palatal plate. Manual exercises contribute to strengthening orofacial muscles and support the plate's mechanical stimulation effect; however, no study accurately describes this physical training <sup>(23)</sup>.

The severity of the signs and symptoms of children with DS may differ greatly from one to the other, which is why the insertion of a stimulating palatal plate is required only in a quarter or a third of all patients. Children with mild orofacial dysfunction show a more harmonious development of the orofacial region per se than children with more severe orofacial dysfunction <sup>(23)</sup>, which agrees with the results obtained from the papers reviewed.

The goal of palatal plate therapy is to create a positional shift of the tongue up and backwards, combined with automatic muscle training and inactive upper lip stimulation. The scientific evidence reviewed <sup>(4,15,18-25)</sup> agrees that inserting a palatal plate leads to tongue retraction to the back of the mouth, strongly justifying the use of ETPTP. However, objective quantification methods are needed to evaluate real changes in muscle tone.

A critical point of the methodology of the studies is a large number of types of palatal plates, based mostly on the design of Castillo Morales, to which various modifications are made. During tooth eruption, in some cases, the plate was modified using accessory retention systems, and in others, the treatment was interrupted for a few months until the oral condition was stable. The use of different designs varied not only among participants in different studies but also within the same study and even in the same patient, leading to greater bias. Therefore, it is not possible to determine the best alternative, which becomes more complex when conducting a standardized comparison of the technique <sup>(22)</sup>.

Regarding treatment stability, there are few studies with long-term evaluations. A treatment period of one year has been estimated to be too short to draw definitive conclusions as to whether the treatment has lasting effects on orofacial function <sup>(22-23)</sup>. Korbmacher H. et al. follow up on patients for over 12 years, which offers a greater predictive potential on the long-term stability of the effects achieved during treatment. This study concluded that long-term orofacial development depends on the severity of orofacial features in early childhood, before plate insertion. Children with customary open-mouth position—greater than 10 mm—and extraoral tongue position (severe orofacial features) showed better development during the follow-up period than those with customary interlabial space smaller than 10 mm and interdental tongue position (moderate orofacial features) at the beginning of ETPTP <sup>(23)</sup>. This demonstrates that changes in orofacial appearance remain stable in most cases, although the mechanical stimulation of the palatal plate is not present in the follow-up period <sup>(23)</sup>. This agrees with Cudzilo et al., who reported that increased muscle tension also continued after plate removal <sup>(4)</sup>.

Although there is still little scientific evidence on how stable the results obtained with combined physical therapy and palatal plate are over time, the benefits directly observed in the muscle function of the tongue and lips also have an impact on the child's better development, observed, for example, in functions such as: swallowing, occlusion, tooth eruption, chewing, speech and language <sup>(20,23-24)</sup>. Non-treatment and no improvement of

abnormal orofacial muscle forces perpetuate or exacerbate a class III skeletal pattern or incisive alveolar biprotrusion <sup>(23)</sup>, since malocclusions in children with trisomy 21 increase or worsen with age due to craniofacial growth delay and characteristic oral motor dysfunction <sup>(20)</sup>. However, the consequences of the growth pattern towards a skeletal class III with a negative anterior overjet cannot be avoided with ETPTP <sup>(24)</sup>.

Several studies agree that the results are not attributable to therapy and that the actual effect of muscle function can only be assessed in the long term. In short-term studies, changes in the position of specific muscles are observed, while in the long term, the effect on muscle tone and stability over time can be evaluated. Therefore, it can be ruled out that it has been a transient improvement due to the recent application of a mechanical element <sup>(21-25)</sup>. After 4 years of therapy, Carlstedt et al., conclude that lip contraction during speech may imply that the palatal plate has improved muscle tone in the lips <sup>(20)</sup>. Regarding the results of the questionnaire applied to parents, they show differences between the groups of patients treated and untreated in relation to open mouth habits, sleeping habits, snoring and sleep apnea, with less snoring detected in patients treated with palatal plate. This may depend on an improved tongue position during sleep. However, to verify that the plate has positive effects on breathing patterns during sleep, thorough, long-term research is needed <sup>(20)</sup>.

It is questionable that none of the assessment methods used in the reviewed studies is standardized (clinical examination, video recording/recording, photographs and parent questionnaire). The outcome of ETPTP is difficult to measure objectively <sup>(19)</sup>, and visual evaluation remains one of the main methods of measuring early treatment progress, i.e., recording changes in mimetic muscle tension, tongue position, and lip closure, during and after treatment <sup>(19)</sup>. Haberfellner and Richter (1980) and Stratton (1981) agree that the accepted method to document treatment effects remains clinical observation <sup>(15)</sup>. A video recording of the child's face is considered a suitable method for evaluating orofacial muscle function as it allows us to repeat the analysis, although the recording reflects oral function for a very limited period and can, therefore, be influenced by fatigue, the child's mood or

environmental conditions such as camera position and lighting, time of day, etc. <sup>(19-22)</sup>. Other studies include, as a complement, the application of a questionnaire to parents in each monitoring session, considering that this procedure may be influenced by parental expectations with an inaccurately positive result <sup>(4,16,18-19)</sup>. Additionally, it is challenging to evaluate front photographs of infants and young children because it is not possible to obtain a standardized position <sup>(19)</sup>. Clinical examination and video recording appear to be more reliable methods of evaluating therapeutic progress, in contrast to the parent interview, which is based on the subjective estimation of guardians. These methods would be significantly more reliable if examiners were trained, calibrated and blinded. In addition, a standardized protocol for interviewing parents could be developed. It is therefore essential to develop precise, reproducible and reliable techniques for the examination and evaluation of oral function <sup>(17)</sup>.

It is essential to have full patient and parent cooperation in every stage for treatment success since the result of the therapy always depends on their commitment. ETPTP is beneficial but imposes additional demands on children and caregivers already overburdened, which is why the practitioner must motivate them to participate and carefully monitor the effects of treatment <sup>(4)</sup>. In the literature, the different interindividual developments observed between one child with DS and another are attributed to different degrees of compliance with treatment by parents and patients <sup>(23)</sup>.

In general, heterogeneity is observed in the duration of treatment and the average age of participants and the time elapsed after the intervention. In the studies included in this review, at the start of treatment, the mean age of participants ranged from 2 months to 12 years. However, it is well known that Castillo Morales therapy emphasizes early treatment to achieve normal oral motor function in children with DS <sup>(17)</sup>. Because the first year of life is the period of greatest development of the central nervous system and mouth, treatment should be started as early as possible <sup>(26)</sup>. On the other hand, the duration of use of palatal plate was between 2 and 48 months. Given the differences outlined above, it is difficult to interpret

the findings for the ideal age and treatment duration. It is suggested that future studies standardize the duration of treatment and age of the children studied, to verify the optimal timing of intervention <sup>(17)</sup>.

## **CONCLUSIONS**

Evidence suggests that a child with DS effectively benefits from ETPTP, as improvements in oral motor function and facial expression, increased mouth closure and decreased lingual protrusion are observed, with more significant changes in more severe cases. However, it cannot be said that with this therapy there is a real increase in muscle tone, as this requires the use of objective quantification methods.

We need further studies with more representative samples, standard and objective assessment methods, and with limited parameters concerning the age of beginning of treatment and its duration.

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