



## Evolution of a child with Treacher Collins syndrome undergoing physiotherapeutic treatment

### *Evolução de uma criança com síndrome de Treacher Collins em tratamento fisioterapêutico*

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#### Abstract

**Introduction:** Treacher Collins syndrome, or mandibulofacial dysostosis, is a hereditary disorder and is manifested by craniofacial malformations. The incidence is close to one case per 40,000 live births, without relation to gender or race. The infant carrier may present neurological development. This rare syndrome requires documentation of its main clinical and kinetic-functional consequences. **Objectives:** The purpose of this study was to describe the clinical and kinetic-functional findings for a child who has Treacher Collins syndrome and receives treatment in the Physiotherapy Department at the Pestalozzi Association in Goiania, and to present the evolution of motor function and psychomotor development during rehabilitation.

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**Materials and methods:** The information was obtained through interviews with the mother, and evaluation of the child at nine and eleven months old, using the infant neurological assessment sheet, Inventory Operational Portage (IPO) and Gross Motor Function Measure (GMFM). **Results:** The GMFM showed that the child had a higher trend in the items for lying and rolling, sitting, crawling and kneeling and minor changes in the items for standing, walking, running and jumping. In the IPO, the child progressed in all of the assessed areas: infant stimulation, motor development, socialization, self-care, cognition and language. **Conclusion:** The child showed progress in psychomotor development in accordance with that expected for their age and initial assessment. It is suggested that children with this syndrome be treated by multidisciplinary teams in the first years of life, preventing delays and deviations in development.

**Keywords:** Mandibulofacial Dysostosis. Child development. Developmental disabilities.

### Resumo

**Introdução:** A síndrome de Treacher Collins ou Disostose Mandibulofacial é um distúrbio hereditário e manifesta-se por malformações craniofaciais. A incidência aproxima-se de um caso a cada 40.000 nascidos vivos, sem interferência de sexo ou raça. A criança com essa síndrome pode apresentar atraso no desenvolvimento neuropsicomotor. A síndrome possui rara documentação, sendo necessário o esclarecimento de suas principais consequências, achados clínicos e cinético-funcionais. **Objetivos:** Descrever os achados clínicos e cinético-funcionais de uma criança com síndrome de Treacher Collins que frequenta o setor de Fisioterapia da Associação Pestalozzi de Goiânia, bem como apresentar a evolução da função motora e desenvolvimento neuropsicomotor durante seu tratamento de reabilitação. **Materiais e métodos:** As informações foram obtidas por meio de entrevistas com a mãe, por avaliações realizadas aos nove e onze meses de idade, pela ficha de avaliação neurológica infantil, Inventário Portage Operacionalizado (IPO) e Gross Motor Function Measure (GMFM). **Resultados:** O GMFM demonstrou que a criança obteve maior evolução nas dimensões deitar e rolar, sentar, engatinhar e ajoelhar e menor evolução nos itens das dimensões em pé, andar, correr e pular. No IPO a criança evoluiu em todos os itens avaliados nas áreas estimulação infantil, desenvolvimento motor, socialização, autocuidados, cognição e linguagem. **Conclusão:** A criança apresentou evolução no desenvolvimento neuropsicomotor de acordo com o esperado para a sua idade e avaliação inicial. Sugere-se que crianças com esta síndrome sejam acompanhadas por uma equipe multidisciplinar nos primeiros anos de vida, visando à prevenção de atrasos e desvios no desenvolvimento infantil.

**Palavras-chave:** Disostose mandibulofacial. Desenvolvimento infantil. Deficiências do desenvolvimento.

## Introduction

Treacher Collins syndrome, or mandibulofacial dysostosis, is a hereditary disorder characterized by craniofacial abnormalities and appears with various clinical variables: deformities in the conformation of the 1<sup>st</sup> branchial arch of the mouth; enlargement of the buccal cavity towards the ear; deformities in the conformation of the 1<sup>st</sup> and 2<sup>nd</sup> branchial arches, consisting of congenital absence of half of the lower jaw; Crouzon syndrome, which is part of a group of congenital malformations characterized by premature closure of cranial sutures; hypertelorism, congenital deformation of the cranium and the face, manifesting

in excessive deviation of the eyes with enlargement of the nostrils. All these pathologies must be included in the differential diagnosis of the syndrome (1, 2).

Treacher Collins syndrome was described in 1846 by Thomson and is known by the name of the British ophthalmologist Treacher Collins, who reported two cases of the syndrome in 1900. Later, in 1944, Franceschetti Klein wrote an extensive review and proposed the term “mandibulofacial dysostosis”, by which it is described in the current literature (3). The incidence approaches one case per 40,000 live births, without relation to gender or race. The syndrome is a dominant autosomal disease of craniofacial development resulting from mutations and loss

of the function of the TCOF1 gene, which encodes nucleolar phosphoprotein. There is a 50% chance of a child inheriting the condition when one parent has the syndrome, and transmission is random (4, 5, 6).

The genetic alteration is present in the distal portion of the long arm of chromosome 5 (5q31.3-q33.3) (7). The phenotypic expression of this disease probably results from a congenital malformation involving the first and second branchial arches. The basic characteristic of this disease refers to a disability during the seventh week of gestation, while the facial bones are forming (8, 9).

The syndrome is characterized by an antimongoloid inclination of the palpebral fissures, lower eyelid coloboma, micrognathia and hypoplasia of the zygomatic arches and microtia (2, 10, 11). Malformations affecting other parts of the body may also occur, such as congenital heart disease; cryptorchidism or the non-descent of one or both testicles into the scrotum, usually caused by hernias, lack of hormonal stimulation or changes in the abdomen; and mental deficiency (2).

The malformations diagnosed at birth are stable, that is, they are not progressive with age (12). Obstruction of the upper airways due to structural factors often generates obstructive apnea in children with Treacher Collins syndrome, as in the case of the angle from the base of the reduced skull, where it modifies the position of the pharynx from the posterior to the anterior; from the narrow nasopharynx and reduced maxillary with anterior projection, reducing the antero-posterior dimension of the nasopharynx (2). The reduction in the size of the jaw is associated with the tongue in the posterior position, generating alterations resulting from improper breathing patterns (13).

Changes in the dentition are more intense and lead to facial changes, such as anteversion of the nostrils, elongated face, protruding cheeks, open bite beforehand, and contraction of the orbicularis and mentonian muscles when trying to close the mouth — this difficulty leads patients to adopt a change in breathing pattern in order to facilitate the exchange of gases, thus becoming mouth breathers (14, 15, 16). To facilitate the passage of air through the mouth, it is necessary to increase the functional interocclusal space. For this purpose, mouth breathers perform mandibular lowering and extension of the head, damaging the visual field, and cervical flexion occurs as compensation (14, 15, 16).

Mouth breathers present changes in the stomatognathic system. This system is comprised of the bones, nerves, muscles, joints and teeth, and has an impact on the whole of the tonic postural system and can trigger changes in cascade and modifications with respect to the position of the head. The main basis of its operation is in correct breathing, chewing and swallowing that manifest in the structural formation of the organs, causing the body to deviate from its axis. When these changes are not detected early, they can turn into degenerative skeletal deformities and cause severe consequences (17).

In the sensory-motor aspect, these children present delayed physical development resulting from the difference in the acquisition of levels of integration in the central nervous system. The following characteristics may also present: weight gain, resulting from a motor deficit; slowness in the development of grinding and protection reactions; asymmetric phase lasting longer than usual, coarse, slow and heavy movements; functional development of scoliosis; delay in the ability to roll over, drag themselves, crawl and walk; stiffness in the distal extremities; flaccidity in the abdomen and in the thorax muscles, hindering breathing capacity; flat feet and valgus; limited fine motor skills and presenting a need to develop activities that include circular movements, especially in locomotion. Furthermore, one can observe delays in walking, present at about month 18 or even up to month 24 (18).

Almeida and Formiga (19) applied the IPO evaluation form with a child with Williams syndrome, in order to evaluate their performance in relation to the activities of extensive and fine motor development. They found that the child presented difficulties in fine motor skills. The instrument proved to be very useful for children with genetic syndromes.

The care of children with Treacher Collins syndrome is multidisciplinary, covering areas such as otorhinolaryngology, ophthalmology, neurology, pediatrics, dentistry, psychology, social work, occupational therapy and physiotherapy. This is a syndrome that has rarely been documented in the literature; thus, it is necessary to clarify the main consequences of this disease and the important clinical and kinetic-functional findings for patients.

Physiotherapists, as part of the multidisciplinary team, must present knowledge about the process of normal acquisition of motor development, which is composed of muscle tone, posture, primitive reflexes,

postural reactions, sensory-motor coordination and spontaneous movements (20).

From the above-mentioned data, it can be concluded that Treacher Collins syndrome is rare and its pathophysiological aspects are not yet fully known. It is known that the main sequelae are facial malformations, airway obstruction, changes in dentition, hearing loss and delayed sensory-motor development.

Physiotherapist can mainly improve neurological deficits such as motor deficits, delays in the development of protective and rectifying reactions, flat feet and valgus, and hypertension. However, in the national and international literature, few studies have been carried out with patients with this syndrome.

Therefore, it is necessary to evaluate some physical and functional aspects and identify shortcomings and physiotherapy intervention, in order to learn about possible treatment and minimize consequences. Thus, the objective of this study was to evaluate the sensory-motor aspects of a child with Treacher Collins syndrome who was undergoing treatment with physiotherapy.

## Materials and methods

### Type of study

This was an observational and descriptive study of a child with Treacher Collins syndrome. Data collection was performed at the Pestalozzi Association of Goiânia. This is a charity that provides care for children from 0 to 12 years of age with multiple and mental disabilities. This research was not funded by any institution.

### Participant

The subject of the study was a female child 9 months of age with a clinical diagnosis of Treacher Collins syndrome. She was also diagnosed by a child neurologist when she was 2 months old.

The patient was attended by physiotherapy interns and she received treatment in the physiotherapy sector at the Pestalozzi Association in Goiânia. In addition to physiotherapy, the child was cared for by a multidisciplinary team, and received other rehabilitation services such as speech therapy and occupational therapy.

### Ethical aspects

The study was conducted after approval by the Research Ethics Committee of the Pontifical Catholic University of Goiás (protocol No. 0769.0.000.168-07), according to Resolution No. 466/12 of the National Health Council. The child's parents agreed to the child's participation as a subject and signed a Free and Informed Consent form; they also authorized publication of the results.

### Material

The following instruments were used for data collection: the child's neurological assessment, the Operational Portage Inventory (IPO) and the Gross Motor Function Measure (GMFM-88). The neurological assessment consisted of data from an interview with the parents and neurological assessment data. In the anamnesis, pre-, peri- and post-pregnancy data and information from the child's medical history were collected, such as hospital admissions, pre-existing diseases, sensory changes and sphincter control.

Neurological evaluation data were collected for muscle tone and trophism, the presence of primitive, deep and superficial reflexes, assessment of range of motion, the presence of shortening and contractures, active movement and postural control.

The IPO was used to evaluate the delay in the child's motor development. This test covers five areas of sensory-motor development for children aged 0 to 6 years of age, including socialization, cognition, language, self-care, motor development and an area for child stimulation, aimed at newborns up to four months of age. The instrument features 580 items divided into five areas according to the age of the child (21, 22).

The GMFM-88 was used to assess the child's motor function. This is a method used to assess motor performance in patients with neurological injuries, such as cerebral palsy and genetic syndromes (23). The GMFM consists of 88 items that assess the gross motor functions of lying down and rolling over, sitting, crawling and kneeling, standing up, walking, running and jumping. First, the total for each item is calculated; then, this total is divided by an exact number, established by the instrument, and multiplied by 100 to give the final score. This instrument was created for the purpose of measuring changes that occur in gross motor function of children with cerebral palsy (23, 24).

## Procedure

The parents were provided with the necessary information about the survey conducted with the child; the physiotherapy assessment was clarified, which consisted of the neurological evaluation, the IPO and the GMFM-88.

The child's sensory motor development was observed throughout the physiotherapy by using the IPO and the GMFM-88 for two evaluations at an interval of 2 months and 9 days. The first evaluation was performed at 9 months and the second at 11 months.

Physiotherapy lasting 35 minutes for each visit was performed twice a week, totaling 21 visits between the first and second evaluation. The proposed treatment was focused on improvement in neurological development and evolution of gross motor function.

The objectives emphasized during treatment were changing certain postures, stimulating neck and torso control, and reactions for protection and balance. In addition, manipulation of sonorous objects in the midline was carried out, with a view to visual and hearing improvement.

## Results

According to gestational data reported by the mother, prenatal care was performed regularly, from the 1<sup>st</sup> to the 9<sup>th</sup> month of pregnancy. The mother had gestational diabetes and reported that she did not use alcohol, cigarettes or drugs.

The child was born by normal delivery in the 39<sup>th</sup> week of pregnancy and weighed 3.21 kg. However, there was a delay in the child's expulsion period during childbirth. Thus, oxygen therapy was necessary, once a day after the delivery.

The child remained in the hospital infirmary for 20 days. On the second day after birth, the doctors identified characteristics of Treacher Collins syndrome; however, the confirmation of the diagnosis occurred in the second month. The child does not take any medication. Physiotherapy was started at only two months, carried out by a multidisciplinary team at the Pestalozzi Association in Goiânia.

With regard to the history of development, the child had head control at 3 months, rolled over alone in the 4<sup>th</sup> month and sat up in the 5<sup>th</sup> month, but without crawling or walking.

Through neurological assessment, characteristics such as agitation and global normotonic and normotrophic muscles without any change in tactile, thermal and pain sensitivity were observed. The child had a positive Babinski sign on the left foot, sat up on a ring, showed postural control in some postures and presented dextroconvex scoliosis in the thoracic region, with no presence of shortening or deformity.

As for the neuro-physical-functional examination, the child had decreased deep tendon reflexes bilaterally (triceps, biceps, patellar and Achilles). One could observe the presence of glabellar eye, corneal-palpebral and cochlear-palpebral reflexes, postural reflex of escape from asphyxia, and gait and positive support.

Also observed were cervical rectification and dissociated body reaction and a decrease in optical and labyrinth rectification reaction. Forward, sideways, backward and break-falling protective reactions were found. In relation to balance reactions, on all fours, kneeling and standing up positions, without support, were decreased.

In assessing socioeconomic conditions, the parents are the ones responsible for the child, reporting that she is their only daughter. They live in an assigned residence housing seven people, and only one is responsible for obtaining the family income (the child's father). The residence has water, electricity and sewage benefits.

In the evaluation conducted with the GMFM-88, it was observed that the child presented developmental delay in the standing posture, as well as transferring to high postures. In the evaluation conducted with the IPO, the child presented delays in the areas of motor development, socialization and cognition (Tables 1 and 2).

Table 1 illustrates that after physiotherapy the child showed the strongest growth in items A (lying down and rolling over), B (sitting up) and C (crawling and kneeling) in the final evaluation with the GMFM-88. However, the biggest difference found between the initial and final evaluation with the GMFM-88 was in items D (standing up) and C (crawling and kneeling).

Table 2 shows that after applying the IPO, the child had improved sensory-motor evolution following the treatment performed. Yet progress was noticeable from the first to the second evaluation, which can be observed in all aspects.

**Table 1** - Results of the GMFM-88 evaluation of a child with Treacher Collins syndrome after physiotherapy – Goiânia, 2010

| GMFM-88                              | IA (%) | FA (%) | FS (%) |
|--------------------------------------|--------|--------|--------|
| Item A: lying down and rolling over  | 88.2   | 100    | 11.8   |
| Item B: sitting up                   | 76.6   | 95     | 18.4   |
| Item C: crawling and kneeling        | 40.4   | 64.2   | 23.8   |
| Item D: standing up                  | 0      | 61.5   | 61.5   |
| Item E: walking, running and jumping | 0      | 12.5   | 12.5   |

Note: IA = initial assessment; FA = final assessment; FS = final score.

**Table 2** - Results of the Operating Portage Inventory evaluation of a child with Treacher Collins syndrome – Goiânia, 2010

| Operating Portage Inventory | IA (%) | FA (%) | FS (%) |
|-----------------------------|--------|--------|--------|
| Child stimulation area      | 100    | 100    | 100    |
| Motor development area      | 38     | 100    | 62     |
| Socialization area          | 26     | 100    | 74     |
| Self-care area              | 100    | 100    | 100    |
| Cognition area              | 11     | 100    | 89     |
| Language area               | 100    | 100    | 100    |

Note: IA = initial assessment; FA = final assessment; FS = final score.

## Discussion

According to the mother, upon performing prenatal treatment, she had not taken any drugs. With regard to diseases associated with pregnancy, she presented gestational diabetes. According to data from the literature, about 25% of newborns from diabetic mothers presented neonatal complications. As for congenital malformations, these outweigh by almost three times those observed in the general population and are a major cause of perinatal mortality (25).

Although the child's mother presented gestational diabetes, we cannot exactly discern whether that was the primary cause for the development of Treacher Collins syndrome. Scientific and technological advances have led to the improvement of glycemic control during the gestational period, reducing perinatal

morbidity and mortality associated with diabetes in pregnancy (26).

The mother reported a delay in the expulsion period during labor. We suggest that pregnant women be evaluated periodically to avoid the results described above.

Not only the glucose test, but also other tests should be performed on pregnant women for early diagnosis of prenatal and postnatal mutations. These allow the identification of the disease in only modestly affected individuals, as well as assistance in genetic counseling for the family in future pregnancies and anticipation, if possible of treatment. DNA analysis of the individual with the syndrome is also recommended (2).

Regarding family history, it was reported that the father has a family member who was born with hydrocephalus. It is estimated that over 50% of cases

of children affected by other syndromes arise as new mutations, i.e., as the first case in the family. However, before diagnosing a case as coming from a new mutation, it is necessary to conduct a thorough examination of family members, with a view to detecting possible signs of the syndrome (27).

The child in this study had physical characteristics of Treacher Collins syndrome, similar to the items already described, in which the syndrome is characterized by antimongoloid inclination of the palpebral fissures, lower eyelid coloboma, micrognathia and hypoplasia of the zygomatic arches and microtia. In addition, one can also observe an inclination of the palpebral fissures, palpebral ptosis, coloboma of the lower eyelids, thinning eyelashes, hypoplasia of the malar and zygomatic eminences, midface hypoplasia with cleft of the secondary palate or high-arched palate, absence of velum and extension of a "temporal hair tongue" along the cheeks (2).

Upon the performance of the functional-physical examination, the child presented postural control, cephalic support, sitting up with and without support and standing with support, but had no support in the posture on all fours, kneeling and half-kneeling. This report differs with data reported in the literature, which shows significant shortcomings in both cognitive and motor skills in children with low weight, compared to those with normal weight. Other variables may play a key role in psychomotor development, such as social and economic characteristics (28, 29, 30).

The child did not perform transference to higher positions independently, sat up on a ring, and presented no shortening or deformities, and the sensory systems were normal. Intense tearing of the eyes was observed, especially in the left eye, a fact evidently due to bad facial formations. The data similar to those found by Bezerra et al. (2).

Upon postural examination, the child presented dextroconvex scoliosis in the thoracic region of the spine due to psychomotor retardation. The child also did not present marching, but adopted high postures with the help of the caregiver. These data confirmed the reports of evaluations of children with Treacher Collins syndrome, which presented delayed physical development resulting from the difference in acquisition of the levels of integration of the central nervous system, slowness in rectification and protective reactions, functional scoliosis and delays in walking during neuro-functional development (18).

According to the neuro-physical-functional evaluation, the child presented decreased deep tendon reflexes bilaterally: triceps, biceps, patellar and Achilles. We noted the presence of primitive glabellar, corneal-palpebral, and cochlear-palpebral reflexes, postural reflexes for escape from suffocation, gait and positive support, with the other reflexes tested being absent. There was the presence of cervical rectification and dissociated body reaction, decreased optical and labyrinthine rectification reaction, the presence of forward, sideways, and backward protective reactions and fall-breaking reactions. In relation to the child's balance reactions, the all fours, kneeling and standing up positions, without support, were decreased.

According to Tamaki (18), children with this syndrome present difficulty evolving balance reactions due to the presence of asymmetry and the delay in the maturation of the rectification and protection reactions, which limit them in a unique way. These results were partially corroborated in this study. Thus, children with the characteristics of these syndromes need constant stimulation and should receive repetitive training so that they can have more effective development. Abnormal development, especially of cervical and labyrinthine rectification reactions, damages the acquisition of body straightening and subsequent balance (18).

Regarding treatment, changes in posture showed higher development of the child in skills related to the horizontal position of item A (lying down and rolling over) in the final evaluation. When comparing the initial and final evaluation with the GMFM-88, the biggest difference was observed in the skills of item D (standing up). This result is probably due to the actual characteristics of the motor development of children with or without Treacher Collins, because at nine months of age a typical baby may not be able to assume a standing posture alone, adopting it at between 10 to 12 months (22, 27).

Motor development is related to function, maturation and the learning process. Increased skills in the child's motor development area were observed in this study, when cervical and torso control, and protective and balance reactions, were worked on. Supposedly, these findings are due to the sensory-motor period of the first year of life, where most of the child's repertoire consists of skills involving the use of the hands, exploring their environment with the use of visual, auditory and tactile-kinesthetic sensorial systems (27, 29).

In the reevaluation performed with the GMFM-88, it was possible to observe that the child showed evolution in the total score of the instrument after physiotherapy. Whereas, in the result obtained with the IPO, the child presented development in all of the items evaluated, after physiotherapy intervention. No studies were found that evaluated children with Treacher Collins syndrome by the methods used, or for the results for physiotherapy. Despite this fact, it was noted in the study that these instruments can also be administered to children with genetic syndromes.

### Final considerations

The child studied presented all the physical characteristics of Treacher Collins syndrome, such as facial malformations, dextroconvex scoliosis and delayed motor development. Upon examination, there was good postural control and head support, remaining sitting with and without support and standing up with support, without performing marching. The child did not perform transference to higher positions independently, and the sensory systems were normal. Several reflexes were not intact and some were missing. Equilibrium reactions were mostly diminished.

After physical therapy intervention, it could be seen by the GMFM-88 that the child was showing better performance in lying down, rolling over, sitting, crawling, kneeling and standing up. Whereas, with the (IPO) instrument, the child showed evolution in all of the items evaluated, i.e., improvement in motor development, socialization, self-care, cognition and language.

Nevertheless, the child needs stimulation at higher postures, in both physiotherapy and as it should be done in the home, since the main problems are with sensory-motor deficits. The parents should also be encouraged to participate more effectively in multidisciplinary treatment, since absenteeism was common, and was one of the main limitations of this study. The family should also receive genetic counseling. Since this was a case study of a rare syndrome, it is necessary to conduct further research to assess the effectiveness of physiotherapy for sensory motor function in children with Treacher Collins syndrome.

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