CASE REPORT
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# Physiotherapy intervention in a girl with Pfeiffer syndrome. Case Report

## Intervención fisioterapéutica en una niña con síndrome de Pfeiffer. Reporte de caso

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All relevant data is in the article. For more information, contact the corresponding author.

## Abstract

**Introduction.** Pfeiffer syndrome is an autosomal dominant congenital disorder that affects 1 in 100,000 live births. Variable malformations, developmental delay, and neurological complications occur.

The objective of this case report is to describe the physical therapy intervention on a girl with type 2 Pfeiffer syndrome, which was based on the concept of training focused on specific tasks, the analysis of changes in postural control, and education and empowerment to parents.

**Case presentation.** A girl who began physiotherapy treatment with a frequency of two weekly sessions, from 6 months to two years, when she managed to walk independently for eight consecutive steps. At 12 months, she achieved independent sitting, at 18 months the girl was in the 50th percentile according to the Alberta scale, and at 24 months she was able to take eight continuous steps without assistance and perform bimanual reaching and manipulation with modified objects.

**Conclusion.** Task-based training and a patient and family-centered approach allowed a girl with type 2 Pfeiffer syndrome, with deficiencies in musculoskeletal and movement-related bodily functions and structures, to ambulate independently for eight steps at two years and to be interested in manipulating and exploring her immediate environment. Physiotherapy intervention can benefit children with Pfeiffer syndrome or similar syndromes, who evolve with deficiencies in their development, improving their quality of life.

## Keywords

Child; postural balance; physical therapy specialty; rehabilitation; syndrome; learning; motor activity; acrocephalosyndactylia; family relations; developmental disabilities.

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## Resumen

**Introducción.** El síndrome de Pfeiffer es un trastorno congénito autosómico dominante que afecta a 1 entre 100.000 nacidos vivos. El objetivo de este reporte de caso es describir la intervención fisioterapéutica en una niña con síndrome de Pfeiffer tipo 2, la cual se basó en el concepto del entrenamiento centrado en tareas específicas, el análisis de cambios en el control postural y la educación y empoderamiento a los padres.

**Presentación del caso.** Niña que inició tratamiento de fisioterapia con una frecuencia de dos sesiones semanales, desde los 6 meses hasta los dos años, cuando logró deambular por ocho pasos consecutivos de manera independiente. A los 12 meses logró la sedestación independiente, a los 18 meses la niña se ubicó en el percentil 50 según la escala Alberta y a los 24 meses pudo dar ocho pasos seguidos sin asistencia y realizar alcances y manipulación bimanual con objetos modificados.

**Conclusión.** El entrenamiento basado en tareas y el enfoque centrado en el paciente y su familia permitieron que una niña con síndrome de Pfeiffer tipo 2, con deficiencias en las funciones y estructuras corporales musculoesqueléticas y relacionadas con el movimiento, lograra deambular por 8 pasos de manera independiente a los dos años y se interesara por manipular y explorar su entorno inmediato. La intervención en fisioterapia puede beneficiar a los niños con síndrome de Pfeiffer o síndromes similares, que evolucionan con deficiencias en su neurodesarrollo, mejorando su calidad de vida.

## Palabras clave

Niño; equilibrio postural; fisioterapia; rehabilitación; síndrome; aprendizaje; actividad motora; acrocefalosindactilia; relaciones familiares; discapacidades del desarrollo.

## Introduction

Pfeiffer syndrome, described in 1964 by Rudolph Pfeiffer, is an autosomal dominant congenital disorder with an estimated incidence of 1 in 100,000 live births, without gender preference [1]. It is recognized as the most severe phenotype of syndromic craniosynostosis [2]. According to the severity of the phenotype, it is classified into three clinical subtypes. Type 1 or "classic" with mild manifestations, including brachycephaly, midface hypoplasia, and abnormalities in fingers and toes; it is associated with normal neurological and intellectual development [3]. Type 2 consists of a three-lobed deformity of the skull (cloverleaf skull), extreme proptosis, anomalies in fingers and toes, ankylosis or synostosis of the elbow, developmental delay, and neurological complications; the main neurological abnormalities are mental retardation, learning difficulties, and seizures [4]. Type 3 is similar to type 2 but without a cloverleaf skull [5]. The medical diagnosis is based on the presence of characteristic clinical findings, including craniosynostosis, facial features, and variable malformations of the hands and feet.

Regarding Pfeiffer syndrome, there is evidence on prenatal diagnosis [1,6,7], ophthalmological and visceral complications [5] and surgical reconstruction [2,3]. Despite the fact that neurodevelopmental alterations occur in this syndrome, to date there are no publications or reports related to physiotherapeutic intervention proposals. This case report aims to describe the physiotherapeutic intervention performed on a girl, from six months to two years old, diagnosed with Pfeiffer syndrome type 2 [2].

Following Hadders-Algra's article [8], maturationist theories (1963) considered that motor development was innate, part of man's phylogenetic development, but in recent decades it has been shown that development is influenced by experience and environment. ; For its part, the Systems theory postulates that development is the result of a dynamic process where aspects of the child, his family and environmental conditions are interrelated [8].

The theory of dynamic systems arose in the sixties from the exact sciences, in the nineties it was introduced to psychology and has contributed to the explanation of motor and cognitive development. It states that "when a system of individual parts is put together, its elements behave collectively in an orderly way" [9 p35]. It understands movement as the result of the interaction between physical and neural components. Motor development is considered a non-linear dynamic process, in which the individual's interaction with the environment and the task determine the possibility of developing new and more complex motor skills [10].

Based on the concepts developed in the Dynamic Systems Theory, intervention proposals have been generated, such as the training of specific tasks, which proposes that the learning of motor skills is more effective when carried out in a real context in which problems offered by both the environment and the task must be solved [11].

The achievement of effective treatments in real contexts requires the collaboration of the family and health professionals. For this, an applicable proposal is the model centered on the patient and his/her family, which was created in 1992 by the Institute for Patient and Family Centered Care (IPFCC, for its acronym in English). In this model, family participation is crucial and is developed collaboratively with health professionals. For the definition of the intervention objectives, the priorities, concerns and needs of the family are taken into account. The model is based on four basic concepts: dignity and respect, information exchange, participation and collaboration. Thus, the beliefs and cultural contexts of families are taken into account, with the purpose of supporting coping strategies, motivation and development of care skills [12-13].

The International Classification of Functioning, Disability and Health is a classification system that aims to become the common language for rehabilitation professionals and ideally for the entire multidisciplinary team, as it allows articulating human functioning, understanding the impact of disability and guide the intervention at all times in the life course of individuals and populations.

Historically, physiotherapy interventions in children with motor disabilities have been aimed at remedying alterations in body structure and function (improving range of motion, muscle strength, tone), but currently the importance of focused interventions is highlighted towards the domains of activities and participation [14].

The objective of this case report is to describe the physiotherapeutic intervention in a girl with type 2 Pfeiffer syndrome.

## Presentation of the case

Mixed race girl, with a birth weight of 2862 g, 48 cm tall and a head circumference of 33 cm. She was diagnosed prenatally with Pfeiffer Syndrome type 2. She underwent craniosynostosis correction surgery at 17 days of birth. Subsequently, she presented hydrocephalus, for which they performed a ventriculoperitoneal shunt. From birth she is diagnosed with hip dysplasia, so she requires the permanent use of a Milgram type splint. Desired girl, with parents interested in the minor's rehabilitation process.

The girl, the subject of this case report, began physiotherapy treatment with a frequency of two weekly sessions of 45 minutes, from 6 months to 2 years, when she was able to walk eight consecutive steps independently. Physiotherapy intervention was framed from three perspectives: the components of the International Classification of Functioning, Disability and Health (ICF) [15], the patient- and family-centered approach, and the concepts of specific task training [16].

To assess its functioning, the joint mobility arc test (ROM) was used; Motor development was assessed biannually using the Alberta Infant Motor Scale (AIMS), which is a commonly used standardized instrument for the assessment of motor development from birth to 18 months. The total score ranges from 0 to 58 points and is associated with 6 percentile ranks, where children between the 5th and 25th percentiles have suspicious motor development, and a percentile below the 5th ranks as abnormal motor development [17].

After 18 months, the Gross Motor Function Measure (GMFM-88) Scale was used. The original version contains 88 items distributed in five dimensions: lying down and rolling (17 items), sitting (20 items), crawling and kneeling (14 items), standing (13 items) and walking, running and jumping (24 items). It is considered a reference test that allows the quantification of gross motor function [18]. The use of this scale is determined from the decision tree for the inclusion/exclusion of cases of cerebral palsy, which include syndromes with inclusion criteria for CP, such as: presenting posture and movement disorders , motor function disorders and that is not progressive [19]. To agree on the achievement objectives with the parents, a follow-up was done through the Rehabilitation Problem Solving English problem-solving format (RPS-Form), based on the CIF [20].

In the initial evaluation, generalized body asymmetry was observed with greater tension in the left side of the body, difficulty in straightening the neck, inability to sustain the sitting position, lack of mobility in the spine and pelvis with thoracolumbar kyphosis and retroverted pelvis, rejection of the bipedal position, with the presence of a negative support reaction and sensory aversion to contact with objects. The girl used some communicative strategies such as social smile and response with sounds to the voice of familiar people. Regarding the ROM, involvement was found at the level of the elbows, hands and knees (Table 1) and according to the AIMS she was in abnormal motor development with a percentile less than 5 (Table 2).

**Table 1. Compromised ranges of joint mobility (Articular Mobility Arcs: ROM)**

| | **July 18/ 2018 (6 months old)** | | **December 18/ 2018 (1 year old)** | |
|---|---|---|---|---|
| | LEFT | RIGHT | LEFT | RIGHT |
| Elbow Flexion | 110° | 115° | 110° | 115° |
| Elbow Extension | 30° | 30° | 30° | 32° |
| Metacarpophalangeal Flexion | 60° | 60° | 63° | 62° |
| Proximal Interphalangeal Flexion | 60° | 60° | 65° | 65° |
| Distal Interphalangeal Flexion | 40° | 40° | 40° | 40° |
| Knee Flexion | 110° | 110° | 113° | 113° |
| Knee Extension | 20° | 20° | 20° | 20° |

**Table 2. Assessment of motor development according to the Alberta scale**

| Measurement moment | 6 months | 12 months | 18 months |
|---|---|---|---|
| Punctuation | 7 | 50 | 58 |
| Percentile | Abnormal development (below the 5th percentile) | Percentile 25 | Percentile 50 |

The intervention was carried out in a pediatric neurorehabilitation center, in the regular presence of the mother as the main caregiver and at home, during all the moments in which the parents challenged, motivated and accompanied the girl. The work was carried out in an interdisciplinary way, with intervention in the areas of physiotherapy, speech therapy and occupational therapy.

During the course of the intervention, the objectives were defined with the parents, and were based on two basic aspects: promoting independence in the performance of activities of daily living and favoring recreational activities corresponding to the chronological age of the girl. The physiotherapist devoted significant time to health education for the parents, where she illustrated the process of motor development, taking into account that she was the first daughter of the couple, the motor learning strategies that were emphasized were prioritization of trial-error, the importance of the context and the self-initiation of the movement.

For the physiotherapy intervention, five key aspects were worked on.

1. The concepts proposed by the specific task training model were worked on, as progress was made in postural control, taking into account the biomechanical characteristics of the girl and the achievement of the specific task, carried out with the least energy expenditure and in a functional manner. An example of this is the acceptance of sitting displacement, even when she managed to crawl at four points, performed on occasions with dissociated patterns.
2. During the intervention at the rehabilitation center, work was done to achieve early postural control, and for this, aspects such as alignment, balance reactions in different postures and transitions, and the progressive decrease of the support base were emphasized to promote movement control within the limits of stability. The analysis of the small motor changes were the parameter to progress in the motor demands.
3. Adjustments to toys to facilitate the development of motor skills and manipulation with an emphasis on playful and daily activities, favoring exploration and self-initiation of movement. The modifications to the toys and the different tools to carry out the activities were carried out at home, according to the observation of the girl's needs.
4. Adoption of the model centered on the user and his/her family, in which parents assumed their role and fostered, in the home environment, the progressive increase in motor challenges present in carrying out daily activities. They participated in providing the girl with possibilities to experiment and find successful motor solutions.
5. Continuous use of the Milgram type splint, ordered to prevent the increase of hip dysplasia.

At 12 months, the initial asymmetrical posture managed to align; the girl achieved independent sitting and bilateral use of the hands, using toys or modified objects. The initial means of displacement she performed was sitting and, eventually, independently modified crawling. The ROMs did not change significantly (Table 1) and according to the AIMS she was located in the 25th percentile (Table 2).

At 18 months, the girl was included in the normal curve on the AIMS scale, reaching the 50th percentile (Table 2). Functionally, she was able to climb onto a chair, supporting herself with her upper extremities, showed bipedal postural control during reaching, and walked with the assistance of another person or pushing a heavy object. At 24 months, she was able to take eight continuous steps without assistance and perform reaching and bimanual manipulation with modified objects. At this age, as a parameter for evaluating motor functionality, the GMFM was used, finding that in Dimension A she achieved a percentage of functionality of 94.1%; for Dimension B, 96.6%; in Dimension C, 88.0%; for dimension D, 94.8%; and for dimension E, 48%, with a global functionality of 84.3%.

During the implementation of the intervention program, there were no adverse events.

## Ethical considerations

The girl's parents signed the informed consent. The endorsement of the Institutional Committee for the Review of Human Ethics of the Faculty of Health of the Universidad del Valle was obtained, with code RC 003 - 021, and the authorization was obtained from the Rehabilitation Center where she received the physiotherapy treatment for the publication of this case report.

# Discussion

In the present case, the physiotherapeutic intervention performed on a girl from 6 months to two years of age, and the motor achievements achieved, are described. The focus of the intervention was centered on the treatment based on the training of specific tasks, the model centered on the user and the family of her, which are congruent with the theory of dynamic systems. In addition, the components of the International Classification of Functioning, Disability and Health were taken into consideration for the intervention. Park Me et al. [13] describe that the approach centered on the patient and his/her family allows to go beyond a focus focused on the achievement of motor skills to give great importance to empowerment, the exchange of information, shared decision-making, the participation of the family in care, education and training.

The girl's parents attended a high percentage of the sessions ordered; They showed the ability to promote movement possibilities, modify the tasks and the tools to support her in finding a solution to the various motor problems that were presented to her on a daily basis. The intense practice of the various motor tasks allowed emphasis to be given to exploration and to find solutions to motor problems that arose through trial and error; Toovey et al. [16] mention that when working with children with neurological injury, the most used strategy is repetitive practice, with positive effects when performed with high intensity. On the other hand, Lucas et al. [21], in their meta-analysis on interventions to improve gross motor performance in children with developmental disorders, report that task-oriented interventions may explain their greater effect because they make it possible to carry out fun and functional activities, capable of being integrated into daily life as leisure or recreation tasks.

The intervention was based on enabling exploration, self-initiation of movement and postural control, for which surrounding objects were modified. This aspect was essential for the achievements achieved, since, as mentioned by Lobo et al. [22], the ability to explore the environment through interaction with objects, sitting up, and locomotion greatly facilitate development.

The use of modified toys was a successful strategy to achieve postural control while sitting, quadruped, and biped, because, although many movements such as reaching sometimes caused loss of balance, the interest in reaching the various toys was the reason to persist in postural control. Hadders-Algra [8] describes that when balance is disturbed in sitting infants, they learn to select the most adaptive postural adjustment from their repertoire.

The intervention plan was based on the scientific evidence published in relation to physiotherapy intervention in children with neurodevelopmental disorders, such as the inclusion of the family in the rehabilitation processes and the approach oriented towards carrying out activities and participation which favored a successful rehabilitation process, in which, despite the deficiencies in musculoskeletal and movement-related body functions and structures, intervention was made to promote mobility and the performance of tasks related to their age in their home environment .

As limitations, it is identified that interdisciplinary work is not reported and the lack of information about the cognitive level of the girl, which can largely justify the motor acquisitions achieved. An important limitation is that motor achievements are described according to the results of the application of standardized scales and that these are reported every six months, as is done in the institution. A qualitative description that would illustrate the dynamics that occurred in daily interaction with the family would have enriched the presentation of this case report.

## Conclusion

The ICF-based intervention, task-based training, and patient- and family-centered approach enabled a girl with Pfeiffer syndrome type 2 with deficiencies in movement-related and musculoskeletal body functions and structures to achieve ambulation by eight steps independently by age two and take an interest in manipulating and exploring their immediate environment. Although the intervention depends on the particular needs of each child, it can show a way for families, clinicians and physical therapists to inform treatment for children with this or similar diagnoses.

Although these findings cannot be extrapolated, the positive results showed that physiotherapy intervention could benefit children with Pfeiffer syndrome or similar syndromes, who evolve with neurodevelopmental deficiencies, improving their quality of life.

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