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PHYSICAL THERAPY TREATMENT IN A PARA-POWERLIFTING AND PARA-SWIMMING ATHLETE WITH ACHONDROPLASIA. CASE REPORT

Keywords: Achondroplasia; Disabled Person; Rare Diseases; Adapted Sport; Physical Therapy.
Palabras clave: Acondroplasia; Persona con discapacidad; Enfermedades raras; Deporte adaptado; fisioterapia.

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RESUMEN

Introducción. La acondroplasia es una condición congénita causada por una mutación del gen codificador de crecimiento del fibroblasto que afecta la osificación endocondral y genera discapacidad estructural; además, es la causa más común de talla baja desproporcionada. Por su parte, el deporte adaptado es una disciplina deportiva que se ajusta al colectivo de personas con discapacidad y una estrategia diseñada para generar inclusión y mejorar la calidad de vida de sus participantes. Se presenta el caso de una paciente con acondroplasia a quien se le diseñó un plan fisioterapéutico de intervención enfocado a minimizar el riesgo de lesiones y prevenir la pérdida de funcionalidad como consecuencia de la práctica deportiva.

Presentación del caso. Paciente femenina de 27 años diagnosticada con acondroplasia y practicante de levantamiento de potencia adaptado y natación paralímpica, a quien mediante valoración fisioterapéutica y de la aptitud física se le encontraron alteraciones posturales y en el patrón de marcha. Dado que por su condición de base no es recomendable que practique los deportes en los cuales compite, se le diseñó un programa de entrenamiento y recomendaciones dirigido a preservar su funcionalidad y mejorar su desempeño en la práctica deportiva. El plan fue puesto en práctica y tolerado de forma adecuada por la deportista.

Conclusiones. Los planes de entrenamiento que favorezcan el mantenimiento de la condición física óptima de los participantes de deportes adaptados les permiten a estos deportistas realizar su práctica sin afectar su expectativa de vida ni su funcionalidad. El análisis del presente caso muestra cómo el fisioterapeuta desempeña un rol importante en esta población, pues los puede ayudar a disminuir las posibles complicaciones que se deriven de entrenamientos y competencias.

ABSTRACT

Introduction: Achondroplasia is an autosomal dominant congenital condition caused by a mutation of the fibroblast growth encoding gene, which affects endochondral ossification. It is the most common cause of disproportionate short stature, generating physical disability. In the presence of disability, adapted sport emerges as a strategy designed to generate inclusion and, thereby, improve the quality of life of disabled people. The aim was to develop a physical therapy plan that included recommendations to minimize the risk of injury and prevent loss of functionality as a consequence of sports practice.

Case presentation: This is the case of a 27-year-old Colombian athlete diagnosed with achondroplasia who competes in the para-powerlifting and swimming modalities. Physiotherapeutic and physical fitness evaluations were carried out, finding obesity and postural and gait pattern alterations. The available literature does not recommend practicing these sports in this type of patient; however, a training program and recommendations were designed to preserve her functionality and improve her sports performance. The plan was put into practice and tolerated adequately by the athlete.

Conclusions: Training plans that promote the maintenance of optimal physical condition in adapted sports participants allow them to continue practicing their sport without compromising their life expectancy or functionality. The analysis of the present case illustrates how physical therapists play a key role in this population to minimize the possible complications derived from training and competitions.

INTRODUCTION

Achondroplasia is an autosomal dominant congenital disorder caused by the mutation of the fibroblast growth encoder gene (1), which is found in the short arm of chromosome 4 and is a type of chondrodystrophy. It is the most common bone dysplasia and is associated with disproportionate short stature. Its annual incidence worldwide varies between 1/10 000 and 1/30 000 live births (1,2).

Physical examination allows suspecting achondroplasia, which is confirmed by radiological tests. Its phenotype is characterized by proximal shortening of the long bones, trident-shaped hands, normal-length trunk, short vertebral pedicles, squared pelvis, contracted skull base, macrocephaly, hypotonia, and ligamentous hyperlaxity (1,3,4). Intelligence and life expectancy are not affected by this condition, although comorbidities such as obstructive sleep apnea, middle ear dysfunction, and spinal stenosis increase the risk of death (5). In turn, hypotonia, hyperlaxity, overweight tendency, and postural alterations increase the risk of craniocervical junction injury and degenerate articular cartilage, causing gonarthrosis in early adulthood and leading to inability to walk (6).

Although physical activity is recommended in patients with achondroplasia, it is advisable to avoid strength sports such as gymnastics, competitive swimming, acrobatics, and contact or jumping sports, as they may be risk factors taking into account the aspects mentioned above (7). However, despite the recommendations, a large number of people with this condition practice such sports competitively in Colombia (8).

Adapted sport is a sports modality in which a series of modifications are made to enable individuals with various types of disability to participate (7). Its purpose is to allow this population to integrate into society and increase their participation. Participants in adapted sports must go through an adaptation process in which training focuses on mobility and acquisition of proper technical sport gestures based on each individual's abilities in order to obtain autonomy and, later, compete (9).

Although preparation and participation in adapted sports competitions can lead to injuries and increased musculoskeletal impairment, these types of sports become relevant in the lives of athletes. They bring benefits, mainly in the emotional and social areas, because they are inclusive and contribute to the overall development of the person with a disability (10). High-performing athletes also receive economic benefits; for example, in Colombia, 125 athletes with physical disabilities compete in para-swimming and 52 in para-powerlifting, and they are paid during the preparation phase for the competitions to which they qualify (8).

There are multiple paralympic sports, but the present case report only analyzes the two sports that the patient practices. Para-powerlifting is an adaptation of powerlifting, and the only discipline in this modality is the barbell bench press, which consists of the development of maximum strength (11). In turn, paralympic swimming includes four styles (freestyle, breaststroke, backstroke, and butterfly), and swimmers may compete individually or in teams combining the four styles in the individual medley or relay races. Swimmers are classified according to the type and severity of their disability and compete in an olympic-size swimming pool (12).

The American Physical Therapy Association (APTA) approach (13), which defines physical therapists as health professionals who study human body movement to preserve the individual's functionality, was used to manage the reported patient. Functionality is understood as the ability of a person to carry out

the activities of daily living according to their context. Consequently, physical therapists have different roles such as rehabilitation and habilitation care; risk prevention and reduction; improvement of physical performance; and primary, secondary, and tertiary care, among others, through the examination, evaluation, diagnosis, prognosis, and intervention phases. The International Classification of Functioning, Disability and Health (ICF) (14) was considered to determine what aspects could be involved in the patient.

With this in mind, the following is the case of a patient with achondroplasia who practices competitive adapted sports in the modalities of para-powerlifting and para-swimming. A physiotherapy plan of secondary intervention was designed for this patient to minimize the risk of injuries, prevent functional capacity loss, and improve fitness to optimize her performance.

CASE PRESENTATION

This is the case of a 27-year-old Hispanic female patient diagnosed with achondroplasia who lives in a low-middle-income household in Bucaramanga and was diagnosed with achondroplasia in the prenatal stage. Physiotherapeutic and physical fitness evaluations were carried out, finding postural and gait pattern alterations.

At the time of the assessment, the patient stated that she was in good health and reported a family history of grandparents with diabetes, high blood pressure, osteoporosis, lupus erythematosus, and two paternal half-siblings with unspecified neurological disorders. Her personal medical history included depression with suicidal ideation arising from society's reactions to her physical appearance, which improved through sports practice and did not require pharmacological treatment.

On physical examination, her blood pressure was 100/60 mm/Hg; heart rate, 80bpm; respiratory rate, 15Brpm; height, 124cm; and weight, 50.3kg, for a body mass index (BMI) of 32.7 (grade I obesity); the percentage of body fat calculated through skin folds was 28%. In addition, the patient reported pain of 4/10 according to the verbal rating scale in the knees when performing activities such as jogging, jumping a rope, and climbing and descending stairs.

The patient stated that she practiced S6 paralympic swimming in free, backstroke and butterfly styles in 2-hour sessions, four times a week, for 2 years; it should be noted that this adapted swimming category includes short-stature athletes (15). She also reported that she had joined the para-powerlifting team 3 months before the assessment and that she trained for 2 hours, 6 times a week, lifting a load of 60kg in the bench press.

Since both modalities practiced by the patient are part of the sports that should be restricted in people with achondroplasia (16), because they may accelerate their degenerative joint process and increase the risk of injury and death due to the movement style that they require, preventive physiotherapy intervention was considered necessary.

Physiotherapy assessment

The patient underwent two physiotherapy assessments, one at the first visit and the other three months later to follow up. The aspects found during the initial assessment are described below.

Given the lack of adapted tests to measure aerobic capacity, the modified Bruce protocol was used, obtaining a VO2 max of 44.18 mL/kg/min (17). The test was suspended before

it could be completed because the patient experienced knee pain after increasing the treadmill incline. Although oxygen consumption was classified as good for age (reference value 39-48.9), the perceived exertion was 4 (moderate exertion) according to the modified Borg scale.

The range of motion was normal, but the patient had hypermobility according to the Beighton scale (18): fifth metacarpophalangeal joint passively flexed until 100°, hyperextension of the elbow and knees, and trunk flexion that allowed the palms of the hands to rest on the floor. Muscle strength was normal on the Robert Lovett scale (19).

Posture was assessed by planimetry with a grid. Asymmetry of the shoulders and genu varum were seen in the anterior view, and genu recurvatum, lumbar hyperlordosis, forward head position, and forward right shoulder were seen in the lateral view; the latter aspect affected the bench press practice because the bar was tilted toward the left side. When evaluating the sole, it was established that the woman had cavus feet (20).

The observational analysis of the gait found that the patient performed the stance and swing phases appropriately, but gait determinants were altered due to increased pelvic rotation and knees in permanent flexion, impacting stance phase knee flexion and causing widening of the base of support (20).

Moreover, it was found that the most affected system in the patient was the musculoskeletal system. She also reported that the most troublesome aspects that affected her personally, according to the CIF (14), were: E325 acquaintances, peers, colleagues, neighbors, and community members; E330 people in positions of authority; E345 strangers; E445 individual attitudes of strangers; and E460 societal attitudes. All these factors made her a victim of discrimination and mockery throughout her life. On the other hand, she said that the following were facilitating aspects: e310 immediate family; e315 extended family; E320 friends; E355 health professionals; E410 individual attitudes of immediate family members; and E420 individual attitudes of friends. This means that the patient feels that her personal (negative attitude toward her condition and emotional instability), family (absence from family events), work (rejection by co-workers and job rejection), and sports (not being able to perform high-impact sports) roles are affected.

According to the APTA, the patient's physiotherapeutic diagnosis was musculoskeletal domain and D pattern: limited joint mobility, motor function, muscle performance, and range of motion associated with bone tissue dysfunction (13).

It should be noted that the medical diagnosis of achondroplasia syndrome caused the patient to have activity limitations such as D4502 walking on different surfaces, D4552 running, and D4553 jumping, and restrictions on mobility such as D470 using transportation, D475 driving, etc. (14).

Physiotherapy treatment plan

The intervention and training plan proposed by the physical therapists consisted of a physical training program that emphasized joint protection based on the patient's conditions and the sports she practiced. Its design included three exercise blocks to strengthen the muscles in a general way (Table 1), allowing the patient to perform other training additional to the regular training. Each activity had a basic description and prescription, which were established according to the physical fitness assessment.

Table 1. Training plan.

Block	Exercises	Series	Repetitions	Intensity *	Frequency	Tool
Block 1	Warm-up 20 minutes					
	Shoulder external rotation	3-4	10-12	70-75% Rmax	2 times/week	Dumbbells or elastic bands
	Shoulder lateral raise	3-4	10-12	70-75% Rmax	2 times/week	Dumbbells
	Front fly	3-4	10-12	70-75% Rmax	2 times/week	Dumbbells
	Shoulder extension	3-4	10-12	70-75% Rmax	2 times/week	Dumbbells or elastic bands
	Knee extension	3-4	10-12	70-75% Rmax	2 times/week	Machine or elastic bands
	One-leg press	3-4	10-12	70-75% Rmax	2 times/week	Horizontal press
	Abductors	3-4	10-12	70-75% Rmax	3 times/week	Pulley or elastic bands
	Buttocks	3-4	10-12	70-75% Rmax	3 times/week	Pulley
	Core (planks)	4	60 s	Body weight	3 times/week	Mattress
	Trunk extension	4	10	Body weight	2 times/week	Exercise ball
	Dynamic stretching 10 minutes					
Block 2	Warm-up 20 minutes					
	Pulley row	3-4	10-12	70-75% Rmax	1 times/week	Pulley
	Upright row	3-4	10-12	70-75% Rmax	1 times/week	Pulley
	Shoulder internal rotation	3-4	10-12	70-75% Rmax	1 times/week	Dumbbells
	Squats	3-4	10-12	Body weight	2 times/week	Exercise ball
	Knee flexion	3-4	10-12	70-75% Rmax	2 times/week	Machine
	Adductors	3-4	10-12	70-75% Rmax	2 times/week	Pulley or elastic bands
	Quadruped hip extension	3-4	10-12	70-75% Rmax	2 times/week	Ankle weights
	Scissor squats	3-4	10-12	20 kg	2 times/week	Bar
	Dynamic stretching 10 minutes					
Block 3	Warm-up 20 minutes					
	Shoulder flexion	3-4	10-12	70-75% Rmax	1 times/week	Pulley
	Shoulder abduction	3-4	10-12	70-75% Rmax	1 times/week	Body weight
	Shoulder extension	3-4	10-12	70-75% Rmax	1 times/week	Dumbbell or band
	Reaches	3-4	10-12	Body weight	2 times/week	Bosu ball
	Half squat band hip abduction	3-4	10-12	Body weight	2 times/week	Elastic bands
	Gastrocnemius muscle (heel lift)	3-4	10-12	Body weight	2 times/week	Decline bench press
	Triceps	3-4	10-12	70-75% Rmax	2 times/week	Bar
	Biceps	3-4	10-12	70-75% Rmax	2 times/week	Bar
	Supine and prone forearm	3-4	10-12	70-75% Rmax	2 times/week	Bar
	Dynamic stretching 10 minutes					

Rmax: repetition maximum.

* Repetition maximum should be reevaluated every 3 months to carry out the progression of the exercise.

Source: Own elaboration.

Within this training plan, and to reduce the risk of joint injuries due to hyperlaxity, it was suggested to avoid postures involving axial loads on the joints and performing submaximal ranges of motion. The plan also established that before each training, a warm-up should be carried out, including joint mobility and multilateral dynamic exercises, as well as low-impact functional training. It was also recommended to include breathing exercises and adequate hydration throughout the training and make a generalized dynamic stretching to cool down.

Furthermore, the patient was advised to avoid jumping, running, deep squats requiring knee flexion >90°, jogging, using elliptical machines, and ascending and descending stairs, which are all activities that are not recommended due to her underlying condition (21). Also, to reduce future complications, she was advised to avoid high-impact and repetitive activities, as well as continuous traumas that pose a risk of injury to the tibiofemoral joint due to instability. Psychosocial support was also recommended to prevent the relapse of depressive symptoms and emotional distress due to rejection and negative attitudes of the environment.

The training plan and the recommendations were socialized with the patient, who put them into practice and showed great acceptance and satisfaction. The relevance of including this plan in weekly training sessions and following the recommendations mentioned above to optimize the performance of their daily living activities was emphasized.

The intervention plan was designed to ensure an average life expectancy for the patient and maintain functionality. The importance of following the recommendations, especially in sports practice, was also emphasized, as the analysis of the deficiencies and limitations detected in the physical therapy assessment was taken into account. No incidents or adverse events occurred during the implementation of the proposed activities.

A three-month follow-up revealed that the patient was adherent to the training plan, which was confirmed by her para-powerlifting coach, who also reported adequate tolerance to all the exercises recommended. It was established that the athlete complied with the suggested recommendations for joint protection, except for avoiding jumping, an activity that can accelerate the degeneration of the articular cartilage of the knees. She explained this behavior by stating that she perceived that jumping the rope favors her aerobic capacity and helps her lose weight.

Some of the changes reported in comparison with the initial assessment include weight loss, reaching 48kg; remission of knee pain during jogging, jumping, and climbing and descending stairs; and improvement in sports performance by increasing bench press load to 65kg.

DISCUSSION

Achondroplasia is considered a rare disease (22), so there is not enough literature to guide intervention plans or sports practice recommendations. However, as with athletes who do not have any disability, people with achondroplasia must follow a structured fitness program before participating in high-level competitions (23). It is also recommended to consider the specific characteristics of the athlete when designing a training plan for patients with this condition, for example, the shortening of their limbs, since this aspect can lead to earlier fatigue and the need for longer and more frequent rest periods. Furthermore, to practice adapted powerlifting, it may be useful to use a thinner training bar because these athletes fail to make an effective grip by not being able to fully close the hand due to their short fingers, which favors wrist injuries (15).

The proposed recommendations in the present case were based on the proper positioning of

the body and the importance of avoiding maximal ranges of motion that could cause injury due to the patient's hyperlaxity. However, during press bench press exercises, the athlete is exposed to spine overextension, which could be harmful due to her history of spinal stenosis. In this regard, previous studies report that an exercise plan that emphasizes the strengthening of the trunk's anterior muscles allows minimizing the extent of extension and favor a more aligned posture of the spine (24). This aspect should be considered at the time of exercise prescription.

The limitations of the case include that there are no validated tests to measure aerobic capacity, bearing in mind that the test used did not measure the actual aerobic capacity of the patient because its score of perceived exertion on the modified Borg scale was 4. It is not useful either to establish BMI in people of short stature, so the results obtained in these aspects of the assessment may not be reliable. In this regard, Sims *et al.* (25) proved that people with achondroplasia have a higher energy expenditure when walking and running because cadence increases, so VO_2Max should be calculated through indirect calorimetry, which could provide a more reliable measurement.

Also, despite the existence of scales adapted to measure BMI, Wagner & Sandt (26) proposed that the predisposition of people with achondroplasia to gain weight is associated with the fact that they have less body area to distribute the extra mass because their bones are small, but the other structures have a normal size. This aspect favors the increased risk of injury and should therefore be considered when devising weight-loss strategies.

The main strength of this clinical case is that the training and recommendations plan was developed in an interdisciplinary manner, with the participation of a physician, a physiotherapist, three eighth-semester physical therapy students and a specialist in physical culture and sports. This allowed considering different relevant points of view. In addition, the patient's adherence to the plan allowed evaluating its effectiveness (27).

Since there are no well-defined or standardized physical training protocols for people with achondroplasia (28), coaches of athletes with this condition should consider collaborating with physical therapists to make an appropriate assessment of all deficiencies and risk factors that may occur. This will also allow designing a training plan to enable athletes to perform optimally and minimize the risk of injury.

CONCLUSIONS

A physical therapy management plan was designed for a patient with achondroplasia after analyzing her impairments and limitations. This plan was intended to provide the necessary care so that, despite the impact that her sports practice could have on her joints, the patient could continue to perform her practices without affecting her life expectancy or functionality.

Adapted sport is a tool that helps people with disabilities explore their maximum potential, favoring their inclusion and participation in society. However, due to health implications, the practice of high-performance sports must be monitored according to the athlete's physical condition.

The analysis of the present case shows how physical therapists play an important role in physiotherapy assessment and diagnosis and in the design of secondary intervention plans to prevent and minimize the possible complications resulting from sports practice, which can have a detrimental effect on athletes' quality of life.

PATIENT'S PERSPECTIVE

When asking the patient for her opinion on the proposed training plan and recommendations,

she said she felt comfortable with it because it included various activities that provided her with tools to strengthen her training. She also stated that she tolerated well the exercises and complied with most of the recommendations, resulting in significant improvements in her overall health. She added that she could even perform activities that are not recommended for her condition, such as jumping rope, which she does not wish to withdraw from her training plan because it is her preferred type of cardiovascular exercise.

ETHICAL CONSIDERATIONS

Resolution 8430 of 1993 of the Ministry of Health of Colombia (29) was taken into account to prepare this case report, and the informed consent of the patient was obtained.

CONFLICTS OF INTEREST:

None stated by the authors.

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