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STUDYING THE EFFECT OF A SPECIALIZED KINESITHERAPEUTIC METHOD ON THE MOTOR DEVELOPMENT OF AN INFANT WITH ACHONDROPLASIA

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ABSTRACT

PURPOSE: The aim of the study is to monitor the effect of a specialized kinesitherapy program on motor development in a child with achondroplasia. METHODS: The study cohort was 1 infant with achondroplasia. The infant had physical therapy sessions 5 times a week with a physical therapist and/or with his parents who were trained in kinesitherapy techniques. Specialized kinesitherapy contained specialized methods for working with children and modifications of methods. For the purposes of the study, we applied the following research methods: research of literature sources; examination of gross motor functions using the Gross Motor Function Measure- 88 method; mathematical and statistical methods. RESULTS: We reported a statistically significant improvement in the results of the conducted motor function testing. CONCLUSIONS: Analysis of the results showed that specialized kinesitherapy was effective in improving gross motor skills in an infant with achondroplasia. We believe that specialized kinesitherapy is the main method of therapy for children with hypotonia caused by various pathologies. The early inclusion of kinesitherapy, the regular conduct of kinesitherapeutic procedures and the correctly selected methodology are of key importance for the effect of the therapy.

Key words*:* pediatric physical therapy, child, rare genetic disease

INTRODUCTION

Achondroplasia arises in about 1 in every 25,000–30,000 individuals (1). According to Ireland (2), achondroplasia is the most common form of skeletal dysplasia, resulting in disproportionate short stature, and affects over 250,000 people worldwide. There are different data on the prevalence: according to Pereira (3) it is from 1 in 26,000 to 28,000 live births, according to Gilbert (4) in England every year about 25-30 children are born with achondroplasia. Although achondroplasia is inherited in an autosomal dominant manner, 80% of affected children have a de novo mutation (3).

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The vast majority of individuals with achondroplasia are diagnosed in early infancy, although prenatal recognition has become more frequent and more accurate. No formal clinical diagnostic criteria have been published, but well defined clinical and radiologic characteristics of achondroplasia (5) usually allow for virtual certainty. Clinical features include: Small stature - small size is not a constant feature in infants, who may have lengths within the normal range (6); Short limbs and rhizomelic disproportion - rhizomelic (proximal) shortening is uniformly present, at least in the arms (5, 7), although variable in severity. Often there are redundant skin folds of the upper arms and the thighs; Macrocephaly - head size is usually large at birth and remains so throughout life (6). The anterior fontanel is often large in infancy and may persist to as late as 5 or 6 years of age; Midfacial retrusion - underdevelopment of cartilaginous bones of the face result in flattening of the entire midface and a flat nasal bridge, a short nasal spine and anteversion of

the nose; Small chest - in addition to the chest being often smaller than average (8), the ribs are overly compliant. This results in paradoxical movement with inspiration, which is often misinterpreted as being retractions reflecting respiratory distress; Thoracolumbar kyphosis - virtually all infants develop a dynamic thoracolumbar kyphosis in infancy (9), but this is not present at birth; Lumbar hyperlordosis. Exaggerated lordosis ("swayback") arises when walking begins; Limited elbow extension. Unlike most other joints, the elbows are stiff and may, with age, become progressively stiffer; Short fingers and trident configuration of the hands; Hypermobile hips and knees; Bowing of the mesial segment of the legs - bowing is not congenital. It most often arises in early childhood and may progress at unpredictable rate and extent until growth is completed; Hypotonia - most infants with achondroplasia are hypotonic (10). The combination of joint hypermobility and hypotonia means that many infants will seem particularly "floppy" (1).

Achondroplasia and thanatophoric dysplasia are inherited as autosomal dominant disorders due to heterozygosity for mutations in fibroblast growth factor receptor 3 (FGFR3). FGFR3 disorders are a group of disorders that include thanatophoric dysplasia, achondroplasia, and hypochondroplasia. (11).

Two mutations in this gene account for 99% of cases; both result in an amino acid change (glycine to arginine), which results in continuous activation of the FGFR3 protein. Having a constantly active FGFR3 protein leads to the inhibition of chondrocyte proliferation, which, in turn, inhibits bone growth. The average adult height for patients with achondroplasia is 131 cm for a man and 124 cm for a woman. (3).

There are 2 forms of achondroplasia - heterozygous and homozygous. The homozygous form (child of 2 parents with achondroplasia) was considered lethal.

Achondroplasia, the most common form of chondrodysplasia (inherited skeletal dysplasia), is characterized by a significant delay in the development of communication and motor skills, especially in the first 2 years, according to Ireland (12). Cognitive function is normal in most persons with achondroplasia (13, 14), although it has long been recognized that developmental delays, particularly motor delays, are common (13, 15).

The first attempt to provide standards for comparison of development in a child with achondroplasia to similarly affected peers was that of Todorov et al (15). The resultant tool, patterned after the Denver Developmental Screening Test. (1)

The screening study was conducted with 197 children with achondroplasia. Screening results show a delay in gross motor skills in children with achondroplasia compared to their peers. No speech delay was reported (15).

Although there is some information regarding the development of children with achondroplasia, no study has simultaneously assessed the pattern of skill development in multiple key developmental domains. A study of 20 families of children with achondroplasia born between 2000 and 2009 in Australia and New Zealand used retrospective questionnaires to determine child developmental data related to significant achievements and milestones. The aim of this study was to determine the age range and developmental sequence of the acquisition of gross motor, fine motor, feeding, and communication milestones for a prospective, population-based cohort of Australasian children with achondroplasia, and to compare this with previously reported prospective data. A secondary aim was to develop a clinical developmental recording form to assist clinicians/therapists in monitoring and recording development in young children with achondroplasia across multiple skill areas. Families were asked to document age of acquisition for 41 gross motor activities; fine motor skills and key moments of communication and feeding. Results are compared with previously available information on motor skill development from a cohort of American children with achondroplasia. Results: Although the results support previously reported delays in gross motor, communication and other skill development, fine motor development does not appear to be as delayed as previously thought (12).

In 2019, Pauli published an extensive review of the current and updated information on achodnoplasia. In it he points out that despite

what has been studied in the last 50 years and there is a lot of information, there is still a lot of unexplored information on the subject.

METHODS

We monitored the effect of regular specialized kinesitherapy in an infant with achondroplasia. The study lasted until the patient reached 30 months of age (corrected age). To assess the effect of the therapy, we applied a study of gross motor skills using the Gross Motor Function Measure - 88 method (16). We performed the initial gross motor examination at the patient's first appointment when the patient was aged 3 months corrected (4 calendar months). The patient is a female infant born by Caesarean section at 35 weeks of pregnancy from the 2nd abnormal pregnancy. After birth, stigmata for achondroplasia were established. Sequencing of the FGFR3 gene was performed, which established a mutation c.1138G>A(p.G380R) in the 8th exon, in a heterozygous state, which confirmed the diagnosis - Achondroplasia. In addition, macrocephaly with hydrocephalus was detected (x-ray, CT, MRI and monthly TFE were performed), therefore, on 10/20/2020, a neurosurgical intervention was performed with a ventriculoperitoneal shunt (VPS). Mitral insufficiency grade I-II was established by echocardiography.

On initial motor status examination, severe general hypotonia was observed, making antigravity movements of the head, torso, and limbs impossible. The upper limbs were abducted and the lower limbs were abducted and externally rotated. Control of the head from the prone position was also difficult due to the larger size of the head. An asymmetric position of the neck with rotation to the left and tilt of the head to the right is noted. This predisposition to cervical asymmetry made it impossible to fully rotate the neck to the right from the prone position and from the supine position. An umbilical hernia is observed at rest and a diastasis when engaging the abdominal muscles. Presence of excessive lumbar kyphosis, typical of achondroplasia.

Specialized kinesitherapy contained specialized methods and modifications of methods. Kinesitherapy was conducted at home 5 times a week for 40-60 minutes. The parents were trained in the implementation of the kinesitherapy program, in correct positioning and in the correct execution of activities of daily living. We performed the Gross Motor Function Measure-88 (16) at the patient's initial examination when she was 3 months corrected (4 calendar months) and then repeated the examination 4 times at 12th, 18th, 24th and 30th months (corrected age). For the purposes of kinesitherapy were provided:

- Pickler's triangle 40x65 cm in size. We modified the triangle with thinner poles with a diameter of 1 cm to provide a better grip;
- Fitness ball (Swiss ball) with a diameter of 75 cm;
- upholstered track with a size of 90x200 cm;
- balancing disk with a diameter of 33 cm;
- Bosu ball size 63x18 cm;
- foam roller with size 15x60 cm;
- TheraTogs suit;
- 80x200 cm hammock;
- stimulating spiky massage ball with a diameter of 7 cm;
- wooden roller with a size of 5x15 cm;
- textile elastic band with a resistance of 14 kg;
- psychomotor module "Stairs" size 90x50x60 cm;

The methodology included:

- exercises with a large fitness ball (Swiss ball);
- elements of Doman's method, with a modified track placed at an angle, stimulating basic locomotion – crawling, rolling from a prone position to a supine position and vice versa, creeping (17);
- exercises on a hammock;
- Bosu ball exercises;
- modification of kinesiological flossing according to the RockFloss method (18);
- stretching for torticollis;
- massage with a spiky massage ball for upper and lower limbs, and for paravertebral muscles;
- compression massage with a wooden roller for upper and lower limbs and for paravertebral muscles;
- stimulating free movements of the upper and lower limbs and the torso;
- positional therapy;
- application of Theratogs costume (19);
- verticalization exercises.

Until reaching the age of 15 months, the patient conducted kinesitherapy classes 5 times a week lasting 40-60 minutes, working with a kinesitherapist once a week, the rest of the procedures were performed by the parents, who were trained. From 15 to 30 months of age, the patient performed kinesitherapy 5 times a week, 3 times a week with a kinesitherapist and 2

times a week with the parents. The first 12 months the procedures included positional therapy, massage of upper and lower extremities, and paravertebral with a massage ball with spines; massage with a wooden roller for upper and lower limbs, and paravertebral; modification of kinesiological flossing according to the RockFloss method of upper and lower limbs; stretching for torticollis; rollerblading exercises; exercises on a fitness ball; stimulation of locomotion on a modified Doman track placed at an angle of 30°; stimulation of free movements of the upper and lower limbs and the torso; positional therapy for neck, torso and lower extremities. After the 12th month, we added to the therapy: a Theratogs costume; hammock exercises; Pickler's triangle exercises; we changed the tilt angle of the modified Doman track to 45°; After the 24th month we added: crawling, creeping and standing on psychomotor module "Stairs"; Bosu ball exercises; standing up with support on various modules, equipments and furniture.

RESULTS

The statistical analysis of the data showed a significant improvement in the results of the applied Gross Motor Function Measure - 88 - at the initial testing, performed at 3 months of corrected age of the patient, the result was 1.96%; at the 2nd testing, performed at 12 months corrected age, the result was 10.58%; at the 3rd testing, performed at 18 months of corrected age, the result was 17.46%; at the 4th testing, performed at 24 months corrected age, the result was 23.8%; at the 5th testing performed at 30 months corrected age, the result was 40.28% **(Figure 1).**

Gross Motor Function Measure - 88 consists of 5 sections: "Lying and rolling", "Sitting", "Crawling and kneeling", "Standing", "Walking, running and jumping". The improvement in patient outcomes was mainly due to improved skills in the Lying and Rolling section.

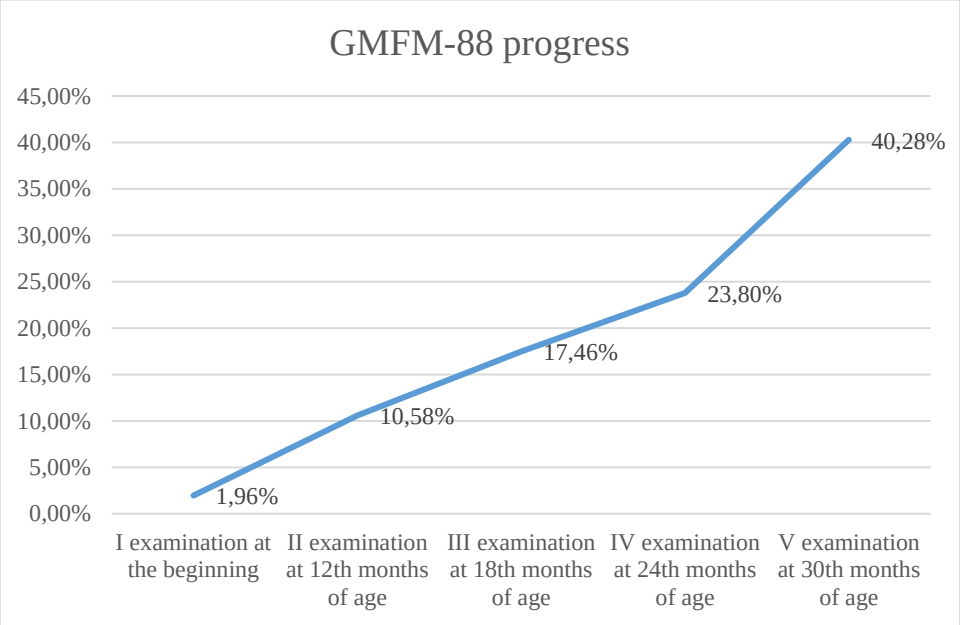

Figure 1. Gross Motor Function Measure – 88 Results

CONCLUSION

Specialized pediatric kinesitherapeutic method effectively treats motor deviation in one infant with achondroplasia. The effect of the therapy on the infant should continue to be monitored. However, the study was done with only one child with achondroplasia, so a study involving more participants should be done to conclude with certainty that the therapeutic approach is effective.

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