



Case report

Rehabilitation of a pediatric achondroplasia patient – Case report

Alicja Warmowska-Dawid¹ , Danuta Krzywda², Kamila Regin³ , Ireneusz M Kowalski³ 

¹ Department of Nursing, Faculty of Health Sciences, Collegium Medicum, University of Warmia and Mazury, Olsztyn, Poland

² Department of Rehabilitation, Regional Specialized Children's Hospital in Olsztyn, Poland

³ Department of Rehabilitation, School of Medicine, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Poland

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ABSTRACT

Introduction: Achondroplasia is the most common non-lethal osteochondrodysplasia characterized by macrocephaly, frontal bossing and depressed nasal bridge, disproportionate short stature, bowing of the lower legs, rhizomelia and trident hands.

Aim: The aim of this work is to present a clinical case of a achondroplasia patient and effects of early rehabilitation.

Case study: A 3-month-old female infant was admitted to our hospital due to insufficient weight gain. Patient was diagnosed with achondroplasia. Due to the truncal hypotonia and right torticollis, between 4th and 24th month of age, rehabilitation according to Vojta method and Bobath concept was conducted.

Results and discussion: Patient was diagnosed with delayed motor development in comparison to healthy children population. With the use of achondroplasia developmental recording forms psychomotor development was found to be harmonic and normal for this disease entity. As a result of rehabilitation normalization of muscle tone and correction of head position was achieved in the 7th month of age.

Conclusions: (1) Children with achondroplasia require early rehabilitation adapted to the delay in the certain stage of development. (2) In the assessment of psychomotor development and effects of rehabilitation 'achondroplasia developmental recording forms' prove to be useful. (3) Its widespread use by clinicians should be recommended. (4) Development of rehabilitation guidelines for achondroplasia patients requires multicenter cooperation.

Corresponding author: Alicja Warmowska-Dawid, Department of Nursing, Faculty of Health Science, Collegium Medicum, University of Warmia and Mazury, Oczapowskiego 2, 10-719 Olsztyn, Poland. Tel.: +4889 539 32 83.

E-mail address: alicja.warmowska@gmail.com.

1. INTRODUCTION

Achondroplasia is the most common non-lethal osteochondrodysplasia with a prevalence ranging from 0.36 to 0.6 per 10 000 live births.¹ It is caused by a mutation in fibroblast growth factor receptor 3 (FGFR3) and inherited in an autosomal dominant manner. Approximately 75% of cases are the result of de novo mutations. The result of this gain-of-function mutation is impaired endochondral bone growth.² In the majority of cases diagnosis is made after birth based on the specific clinical and radiological picture. Typical cases do not require molecular testing. Characteristic features of achondroplasia include macrocephaly, frontal bossing and depressed nasal bridge, disproportionate short stature, bowing of the lower legs, rhizomelia and trident hands. Motor development of children with achondroplasia is frequently delayed, although they have normal mental capacity.^{3,4} Other serious health consequences in children with achondroplasia are related to hydrocephalus, craniocervical junction compression, recurrent otitis media or thoracolumbar kyphosis.^{5,6} General recommendations for the management and screening for the most common complications have been developed by American Academy of Pediatrics.⁵

2. AIM

The aim of this work was to present clinical case of a female achondroplasia patient and effects of early rehabilitation methods.

3. CASE STUDY

A 3-month-old female infant was admitted to the Regional Specialized Children's Hospital in Olsztyn due to insufficient weight gain (1120 g in 3 months). A child was delivered by cesarean section at 39 weeks gestation due to threatening fetal hypoxia, with Apgar scoring 8/10/10 at 1, 5 and 10 minutes, respectively. Physical examination on admission revealed rhizomelic shortening of the limbs, bowing of the legs, short and broad hands, clinodactyly, relatively enlarged neurocranium (head circumference between 90–95 percentile), prominent forehead, wide nasal bridge, short neck, global muscular hypotonia and delayed psychomotor development. Radiographs of spine, hands and lower limbs, as well as echocardiography and abdominal and cranial ultrasound examinations were performed. Electrocardiography and ultrasound results were normal. Skeletal radiology revealed hyperkyphosis of lumbosacral region, broadened distal femoral and proximal tibial metaphyses, which was suggestive of achondroplasia (Figures 1–2). Based on the whole clinical picture diagnosis of achondroplasia was made (which in 7 months of age was confirmed by molecular testing). Patient was consulted by neurorehabilitation specialist.

After discharge patient was referred to outpatient rehabilitation. On the follow-up visit at 4 months of age



Figure 1. Lateral spine X-ray.

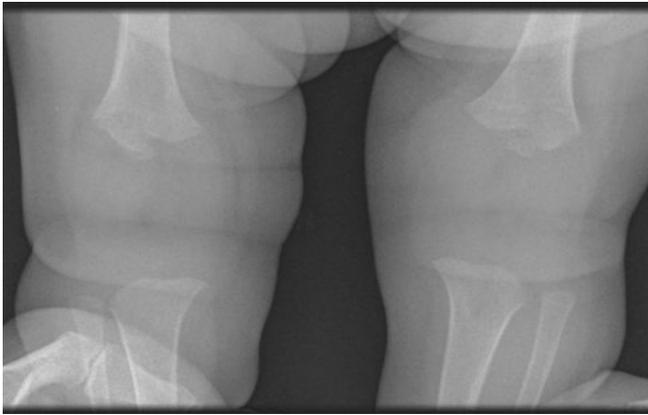


Figure 2. Left knee AP X-ray.



Figure 3. Physiotherapy with the use of Bobath concept.

decreased muscle tone in the head-trunk axis in reactions according to Vojta, abnormal traction response, improper prop in prone position, hands clenched into fists, hips in flexed position and asymmetric positioning of the head – right torticollis were found. Since 4 month of age the patient was rehabilitated with Vojta method and Bobath concept. Rehabilitation aimed at normalization of muscle tone, correction of torticollis and stimulation of psychomotor development. Up to 24 months of age 7 therapeutic sessions

(10 meetings of 45 minutes) were conducted by physiotherapist during outpatient, stationary and home visits (Figure 3). The patient was regularly checked up every 6–8 weeks by rehabilitation specialist. For the assessment of motor development of the patient in relation to the expected age of achievement of certain gross motor skills and communication, called developmental milestones in patients with achondroplasia, ‘achondroplasia developmental recording forms’ were used.⁸

4. RESULTS

Based on clinical observations delayed motor development was found as compared to healthy children population with the use of Denver II test.⁷ Development of communication capacity during the observation period was however within normal range. In the period of intensive rehabilitation from 4 to 24 months of age harmonic development of the patient was observed, which was within normal range for this disease entity.^{8,9} Particular data is presented in Table 1. In relation to the expected age of achievement of certain developmental milestone in patients with achondroplasia, it was found that the patient was in the 50th percentile for creeping. Age of achievement of such milestones as head stability in prone position, rolling over, crawling, sitting from standing position, standing without assistance and independent walking were above median and below 90th percentile. Analysis of communication milestones revealed that the patient began to smile earlier than half of the patients studied by Ireland et al.⁸ The child was in 50th percentile for waving ‘bye-bye.’ Nodding head for ‘yes,’ playing peek-a-boo, imitation of words, using first words with meaning and following simple commands was between 50th and 90th percentile. However, saying ‘mama’ and identifying body parts – in 90th percentile. As a result of rehabilitation normalization of muscle tone and correction of head position was achieved in 7 month of age. However, in 24 months of age hyperkyphosis of lumbosacral region of the spine was still observed (Figure 4). The description involves major milestones included in the ‘developmental recording forms’ developed by Ireland et



Figure 4. Hyperkyphosis of lumbosacral region.

al.⁸ on gross motor skills and communication, in which the delay in children with achondroplasia is observed. The ones not included in the documentation based on information provided by the parent were omitted.

5. DISCUSSION

Medical problems of achondroplasia patients result from abnormal endochondral bone growth. They involve numerous systems and organs and are observed already since birth. The average length at birth is 47.7 cm for boys and 47.2 cm for girls.⁴ In the later development children with achondroplasia have short stature. Anthropometric parameters are evaluated according to special charts for growth, weight, head circumference and chest circumference. In newborns transient truncal hypotonia is frequently observed, which contributes to the delay in the achievement of certain milestones during the first two years of life.^{4,10,11} These delays involve the development of gross motor skills, communication (single words, word combination) and locomotor skills.^{8,9} Delayed speech development is explained by frequent infections of the middle ear, which can result in hearing loss. Articular laxity together with muscle hypotonia and macrocephaly contribute to the development of thoracolumbar hyperkyphosis in the majority of infants with achondroplasia. Recommendations restrict unsupported sitting until 12–15 months and using carriers or car seats in which the spine of an infant is C-shaped. Thanks to that, number of cases of hyperkyphosis requiring orthopedic corset treatment has significantly reduced.¹² In older children with achondroplasia limitation of elbow and hip extension as well as knee and hand joint hypermobility are also present.² Exercise tolerance and muscle strength is also reduced, which may be associated with lower muscle mass, impaired coordination and abnormal body proportion.¹³ According to the recommendations of American Academy of Pediatrics a child with achondroplasia should be under the care of multidisciplinary team.⁵ Patients who present with motor deficits require complex analysis and implementation of the available rehabilitation methods.^{14,15}

Bobath concept and Vojta method used in patient's therapy are the leading neurorehabilitation methods which are widely used in rehabilitation of children with movement disorders.^{16,17} Its overall objective besides normalization of muscle tone and inhibition of abnormal movement patterns is stimulation of psychomotor development.^{18–20} Bobath concept is considered a problem-solving approach, rather than a rehabilitation technique. Therapist aims at encouraging and increasing the child's ability to move and function in as normal a way as possible.^{20,21} Bobath concept has been found effective in rehabilitation of pediatric population with cerebral palsy in terms of motor development.^{22,23} Vojta method involves triggering reflex motor reaction by a sensory stimulation of exteroceptors and proprioceptors of the patient and seems to be an efficient rehabilitation approach for children with delayed motor development.²⁴

However, in the literature there is no information on the effectiveness of these methods in rehabilitation of children with achondroplasia.

6. CONCLUSIONS

Children diagnosed with achondroplasia require early rehabilitation adapted to the delay in the certain stage of development and their individual capabilities.

In the assessment of psychomotor development and effects of rehabilitation 'achondroplasia developmental recording forms' prove to be useful. Its widespread use by clinicians for the assessment of development of patients with achondroplasia should be recommended.

Development of rehabilitation guidelines for achondroplasia patients requires multicenter cooperation in order to obtain larger study group.

Conflict of interest

The authors report no conflict of interest.

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