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Achondroplasia syndrome with clinical manifestations of pneumonia and symptoms of parental social isolation

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Abstract
Introduction: Achondroplasia is an autosomal dominant genetic disorder and is one of the leading causes of dwarfism. In this disorder, the FGFR-3 protein or FGF receptor, which is involved in bone cartilage production, has been mutated. The disease affects one in every 40,000 to 150,000 births. Achondroplasia is a type of congenital short stature that is characterized by short limbs. The main problem in these patients is the conversion of cartilage to bone in the growth plates, especially in the long bones of the limbs. The average height of a man with achondroplasia is about 130 cm and a woman with achondroplasia is about 124 cm. The trunk of these patients usually has no particular problem and the shortness is more in their limbs. Achondroplasia is a genetic disorder and is one of the leading causes of dwarfism. A case of achondroplasia syndrome with clinical manifestations of pneumonia and symptoms of parental social isolation has been reported in this study.
Case report: In this report, a ten-month-old female infant with achondroplasia was observed with clinical manifestations of pneumonia with increased body temperature, coughing, vomiting, anorexia, and wheezing. Blood tests showed; WBC= 12900 µl, ESR= 30, CRP= +1. The patient was treated with ceftriaxone, vial salamol, bromhexine, ondansetron, and acetaminophen ampoules. On the third day of hospitalization, the patient's fever was stopped and blood test was repeated; ESR= 13, WBC= 7900 µl.
Conclusion: According to the present report, it is recommended that in infants with achondroplasia, the increase in body temperature and the WBC, ESR tests should be considered, and the necessary measures should be taken to treat the disease. Also, attention should be paid to the social isolation of parents and necessary measures should be taken in their regard.

Keywords: achondroplasia syndrome, pneumonia, parental social isolation, treatment

Introduction

Achondroplasia is an autosomal dominant genetic disorder and is one of the leading causes of dwarfism. In this disorder, the FGFR-3 protein or FGF receptor, which is involved in bone cartilage production, has been mutated. The disease affects one in every 40,000 to 150,000 births. Achondroplasia is a type of congenital short stature that is characterized by short limbs [1]. The main problem in these patients is the conversion of cartilage to bone in the growth plates, especially in the long bones of the limbs. The average height of a man with achondroplasia is about 130 cm and a woman with achondroplasia is about 124 cm. The trunk of these patients usually has no particular problem and the shortness is more in their limbs [2].

However, it should be noted that at least 80% of the disease is not inherited and it occurs in a person due to a new mutation with no family history. About 20% of these cases are inherited, and the mutation follows a non-inherited pattern on the asexual chromosome. This means that if one parent has achondroplasia, he/she can pass it down to the child. In single-parent cases, the father's age increases the risk of developing a disease of new origin. The incidence of this disease is between 0.5 and 1.5 per 10,000 births [3].

Symptoms of this disorder include short stature with short upper and lower limbs, especially shortness of arms and thighs, limited range of motion in the elbow and difficulty in opening it completely, large head with a prominent forehead, short fingers with middle fingers being apart from the ring finger, normal intelligence, slowing or stopping breathing for a short time, obesity, frequent ear infections, lordosis and kyphosis, spinal canal stenosis, bow legs, lower back pain at puberty, decreased muscle strength, club foot, and hydrocephalus. Before the 20th week of pregnancy, their disorder can only be detected by molecular DNA testing of the fetus and in late pregnancy by analysis of the fetal skeletal picture [4]. Treatments include; control and treatment in infancy and early childhood, use of growth hormone, increasing the length of lower limbs through surgery, and helping to adapt socially due to the psychological aspects of appearance, short stature, and disability. Also, these patients should have the operation for chronic middle ear infarction, hydrocephalus, brainstem compression, obstructive sleep apnea, spinal stenosis, and spinal stabilization [5].

Patient introduction

A ten-month-old female infant was admitted to the pediatric ward of Khatam Al-Anbia Hospital in Gonbad-e Kavous with a complaint of fever and shiver, wheezing, coughing, and vomiting. Three days before her hospitalization, she had

suffered from train coughs, which were accompanied by green sputum secretions that lasted for about a minute. The coughs were controlled by nebulizer therapy. The day before the hospitalization, she had a shiver, followed by a 39-degree fever. The mother used acetaminophen syrup for her infant fever and washed her feet by cold water, but they did not work. Also, she has been vomiting since the day before her hospitalized, and she did not even want to breastfeed, and her low appetite caused weakness and fatigue. The infant's mother did not report a history of hospitalization, drug, or food allergies in the infant. The infant has been born by virginal delivery in full term. The mother did not mention a family history of genetic disease. The family's two previous children are healthy. Parents had no family relationship. From the age of seven months, maternal ultrasound has been used to diagnose fetal achondroplasia syndrome. The baby's head circumference at birth was more than normal. Preliminary examinations showed a body temperature of 39° C, normal head circumference, and anomalies due to achondroplasia syndrome in the upper and lower limbs, with the hands and feet smaller than normal. The infant's legs were bow shape (Figure 1). Parents, especially mothers, were reluctant to be in public because of their infant's physical appearance and were socially isolated. Preliminary tests including the following blood tests were taken: White blood cell (WBC)= µl12900, (normal range= 4400-11000), hemoglobin (HB)= 11.3 gm / dl (normal range = 12-16 gm / dl), ESR= 30 (normal range= less than 20), CRP= +1, urea (BUN)= 13.6 mg / dl (normal range= 8-25 mg / dl), creatinine (Cr)= 0.7 mg / dl (normal range= 0.3-0.7 mg / dl). A chest x-ray was performed for the patient. Patients with the diagnosis of sepsis and pneumonia were treated with 200 mg of ceftriaxone ampoule as BD, 100 mg of bromhexine ampoule as TDS, 2.5 mg of vial salamol inhaler, 75 mg of acetaminophen ampoule as PRN, and 1mg of ondansetron ampoule as PRN. The FFP received 10 cc / pkgr. An Air humidifier was administered at night during hospitalization. Chest physiotherapy was performed daily for three days. Three days later, the WBC, HB, ESR, CRP tests were repeated as follows: (WBC)=7900 µl, ESR=13, and CRP= -. For the first two days of hospitalization, the patient had a fever of over 38 degrees, which was controlled with acetaminophen injections. From the third day of hospitalization, the patient's fever was stopped. Her wheezing was also relieved. The child's vomiting and anorexia were also relieved.

Fig 1

Fig 2

Fig 3

Discussion

Achondroplasia is a type of disorder of bone growth that causes shortness of arms and legs and abnormal short stature. The autosomal dominant mutations in the fibroblast growth factor 3 receptor cause achondroplasia, which is the most common form of dwarfism in humans. It is a genetic disorder that causes shortening of the limbs. The head is often large and the forehead is prominent, making virginal delivery difficult. In this report, a rare case of achondroplasia with clinical manifestations of pneumonia and social isolation of parents has been introduced, which was associated with increased WBC and ESR. In this case study, a newborn baby with achondroplasia was born through virginal delivery from a 21-year-old mother in 2020. In this case, achondroplasia was diagnosed based on prenatal ultrasound findings, clinical features, and neonatal radiological findings. The infant was admitted to the neonatal intensive care unit for observation and was not discharged the day after the examination because no complications were observed [2].

In two children with achondroplasia that abnormal bone growth has been developed in the area of the distal radial ulnar joint (DRUJ), the problem could not be detected by a single osteochondroma in hertology, but radiographic images, location, and symmetry suggested the rare disease of dysplasia epiphyseal hemimelia (DEH). In one child, the

symptoms were relieved with surgery. These are the first cases of DEH in achondroplasia. Due to the rarity of DEH, further research is needed to better understand the possible association with achondroplasia. In this study, for the management of treatment, joint decision based on the symptoms was recommended [7].

A case study in 2017 reported that a 61-year-old man with a history of achondroplasia complained of back pain, radiculopathy, and neurogenic congestion. In radiographic and MRI images of the patient, kyphosis and spinal stenosis were confirmed. This is the first adult to develop achondroplasia, in which spongelectomy and lateral arthrodesis were used to treat severe kyphosis and lumbar spinal stenosis [8].

These infants are at risk for infections, including middle ear infections and pneumonia. Therefore, along with other diagnoses, an increase in body temperature should be considered. The parents of these children are exposed to social isolation. Given the above statements, early diagnosis and treatment of these patients to reduce the risk of infections, accompanying symptoms, and social isolation of parents is very important.

Patient consent

Consent to publish the case report was obtained.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship. Planning, conduct, reporting, conception and design, acquisition of data: SK, LM. Analysis and interpretation of data: AH, KB. Patient management: KB, AH.

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