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Neonatal Achondroplasia: Case Report

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Abstract

Introduction: Achondroplasia, the most common form of skeletal dysplasia with characteristic short limb dwarfism, is a non lethal variant of chondrodysplasia. Although autosomal dominant genes may be the source of inheritance, most occurrences start off as spontaneous mutations.

Case report: Our patient was a term male baby born via elective Caesarean section with birth weight of 2.8kgs. Anomaly scans of 22 weeks of gestation showed no gross anomaly of the fetus.The anomaly scan of 36th week of gestation showed a small size of fetus. On clinical examination, the head circumference was more than normal and the baby had a large head with frontal bossing. There was bilateral symmetrical shortening of upper and lower limbs with short fingers. There was depressed nasal bridge. The abdomen was protuberant and distended.

Literature review: A mutation in the 4p16.3 fibroblast growth factor receptor-3 gene (FGFR3) results in achondroplasia. One parent's achondroplasia increases the infant's probability of inheriting the disorder by 50%, and if both parents have it, the infant's chance increases to 75%. This suggests that the disorder may be inherited as an autosomal dominant characteristic.

Keywords: Achondroplasia; Short stature; Dwarf; Caesarean section; Mutation.

INTRODUCTION

Achondroplasia, the most common form of skeletal dysplasia with characteristic short limb dwarfism, is a nonlethal variant of chondrodysplasia. Although autosomal dominant genes may be the source of inheritance, most occurrences start off as spontaneous mutations. The distinctive features are seen on radiographs of the limbs, pelvis, cranium, and spine. Legs of affected persons are rhizomelically shortened. A normal trunk length, a significant lumbar lordosis, genu varum, a prominent forehead (frontal bossing), midface hypoplasia, rhizomelic shortening of the arms and legs, and a trident hand configuration are among the phenotypic traits.[1] Achondroplasia is a well-known cause of disproportionately small stature, although compared to children and adults, it is more challenging to detect at birth.[2] The majority of people with achondroplasia have normal IQ. Obesity, recurring ear infections, and episodes of slowing or stopping breathing (apnea) are among the health issues linked to achondroplasia. Those who with the disorder typically grow up with bowed legs and a noticeable, lifelong wobble in the lower back (lordosis). Back pain and an irregular

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front-to-back curvature of the spine (kyphosis) are also experienced by some affected individuals. Spinal stenosis is a potentially dangerous achondroplasia consequence.[3] We present a case of achondroplasia that was identified on the first day of life based on radiological and clinical characteristics.

CASE REPORT

Our patient was a term male baby born via elective Caesarean section with birth weight of 2.8kgs. The baby was born out of a non consanguineous marriage to a primigravida mother who is a resident of ahmednagar, Maharashtra.

There was no obstructed labour. The placenta and membranes were completely separated and removed.

Anomaly scans of 22 weeks of gestation showed no gross anomaly of the fetus.

The anomaly scan of 36th week of gestation showed a small size of fetus. It also noted dysplastic bilateral short femur and humerus for age. Both parents are also suffering from achondroplasia and have short stature with no other congenital anomaly.

The baby was delivered via elective caesarean section and cried immediately after birth. There was mild respiratory distress for which the baby was admitted in the NICU for a total of 3 days. After which the baby was transferred to mother-side in the maternity ward. On anthropometric measurements, head circumference is 37 cm, chest circumference is 31cm, length is 44 cm, upper segment is 28 cm, lower segment is 16 cm and the US:LS ratio is 1.75.

On clinical examination, the head circumference was more than normal and the baby had a large head with frontal bossing. There was bilateral symmetrical shortening of upper and lower limbs with short fingers. There was depressed nasal bridge. The abdomen was protuberant and distended.

The baby had normal tone and power.

Blood investigations were sent which came out to be normal.Random blood sugars were normal at the time of NICU admission. X ray of the baby was done which suggested achondroplasia as it showed a broadening of the bilateral femur and humerus's proximal and distal metaphyses, suggesting metaphyseal flaring. Rhizomelic shortening resulted in bilateral shortened femur and humerus. Both hands' metacarpals were short and comparable in length, and the ring and middle fingers were separated (the trident hand).

USG of abdomen and pelvis was normal. Neuro sonography was normal.

The baby was admitted for 2 days till feeding was established and the parents were confident enough and then was discharged.

Fig. 1: Shortening of limbs, large head with frontal bossing, flat nasal bridge and protuberant abdomen.

Fig. 2: Happy parents with the baby

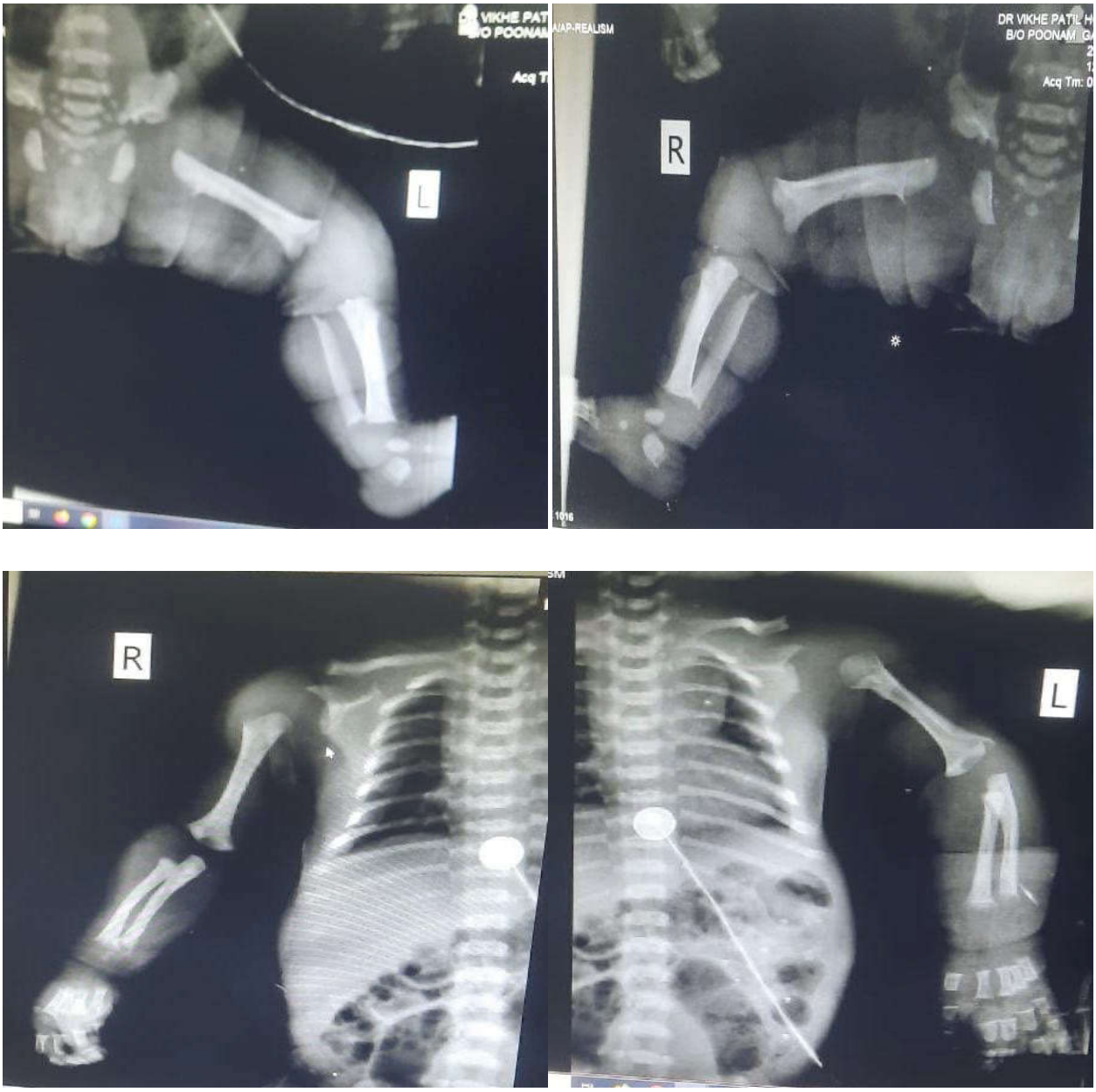

Fig. 3: Multiple x-rays suggestive of bilateral shortening of femur and humerus

DISCUSSION

A mutation in the 4p16.3 fibroblast growth factor receptor-3 gene (FGFR3) results in achondroplasia. One parent's achondroplasia increases the infant's probability of inheriting the disorder by 50%, and if both parents have it, the infant's chance increases to 75%. This suggests that the disorder may be inherited as an autosomal dominant characteristic. Nonetheless, the majority of instances manifest as spontaneous mutations, meaning a kid with achondroplasia can have parents without the condition. In our instance, both of our parents were having achondroplasia and having short stature. In the developing world, the diagnosis is primarily dependent on clinical and radiological findings. (3,4) The global incidence of achondroplasia is 1/77,000–1/15,000.[5]

Because of the shorter long bones, ultrasounds typically reveal the suspicion.[5] In our instance, dysplastic short femur and humerus was suggested by third-trimester ultrasonography. Six the majority of affected individuals' distinctive clinical and radiological symptoms can also be used to make the diagnosis.[7] Despite the fact that diagnosing a case

at birth is more challenging than diagnosing one in a child or adult, our case was diagnosed at birth because of a suggestive ultrasound, radiological evidence, and unique clinical features. Clinically, the patient exhibits a protuberant abdomen, a big head with frontal bossing, a disproportionate shortening of the long bones, and a flattening of the nasal bridge. Achondroplasia can be diagnosed by careful observation because its traits are quite distinctive. But due to financial crisis the patients genetic testing for FGFR3 gene could not be done.

In a family with sporadic instances, the estimated probability of recurrence is 1 in 443.[8] It is stated that one of the parents' mosaicism is to blame for this. Achondroplasia carries a 50% chance of recurrence in kids of either sex if one of the parents has the condition. 25% of offspring will be normal, 50% will be heterozygous, and 25% will have a homozygous mutation if both parents are afflicted. Achondroplasia homozygous is invariably fatal.[9]

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