Acromicric Dysplasia: Misdiagnosed as Achondroplasia: Case Report

Akondroplazi Olarak Yanlış Tanı Alan Akromikrik Displazi Olgusu

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Geliş Tarihi/*Received:* 17.01.2009
Kabul Tarihi/*Accepted:* 17.09.2009

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ABSTRACT Acromicric dysplasia is a rare inherited disorder characterized by abnormally short hands and feet, growth retardation and mild facial abnormalities. The usual age at diagnosis is around 3 years. The condition appeared to be sporadic but the observation of vertical transmission in reported three families was consistent with an autosomal dominant mode of inheritance. We described a 3-year-old boy with small nose, anteverted nares, small abnormal hand-foot, short stature and normal intelligence. Roentgenograms of the hands were characteristic: the metacarpals and the phalanges were short and stubby without shortness of the long bones. The differential diagnosis with achondroplasia and the other skeletal dysplasias with shortening of the hands and feet were discussed in this paper.

Key Words: Shorthand; achondroplasia

ÖZET Akromikrik displazi, boy kısalığı ile birlikte el ve ayak parmaklarında belirgin kısalık ve tipik yüz görünümü ile karakterize nadir bir iskelet displazisidir. Sıklıkla tanı 3 yaş civarında konulmaktadır. Olgular genellikle sporadik olup 3 ailede otozomal dominant vertikal geçiş de gözlemlenmiştir. Biz de 3 yaşında küçük burun, antevert burun kanatları, küçük el ve ayakları olan, kısa boylu ve normal zekâlı olguyu sunduk. Röntgenogramlarda karakteristik olarak karpal ve tarsal kısalık uzun kemikler etkilenmeden görülmektedir. Kısa el ve ayaklarla giden akondroplazi ve diğer iskelet displazileri ile arasındaki fark bu yazıda tartışılmıştır.

Anahtar Kelimeler: Stenografi; akondroplazi

Turkiye Klinikleri J Pediatr 2010;19(3):265-7

Acromicric dysplasia is characterized by markedly short hands and feet, dwarfism to variable degree and characteristic face with narrow palpebral fissures, short stubby nose and anteverted nostrils.[1] The usual age at diagnosis is around 3 years. The condition appeared to be sporadic but the observation of vertical transmission in reported three families was consistent with an autosomal dominant mode of inheritance.[2] Here, we described a boy with dysmorphic face, small abnormal hand-foot, short stature with normal intelligence.

CASE REPORT

A 3-year-old boy was referred to our department for evalution of short stature and abnormally short hands and feet (Figure 1a). He was the third child

of consanguineous parents. He had no problems during the neonatal period and no history of seizures. Short stature was noticed at 3 years of age. Mental development was normal. There was no difficulty in vision or hearing. There was no similar feature in the family history and the parents were of normal height and proportion. The patient weighted 14 kg (50th percentile according to the Turkish population) and his height was 78.6 cm (<3rd percentile). His head circumference was 47.5 centimeters (50th percentile). On examination, the skull was dolicocephalic with frontal bossing and occipital prominence. He had broad, depressed nasal bridge and anteverted nostrils. Fingers were short with mild contractures and he was unable to make a complete fist. Pronation and supination were normal. The thumbs were short, flat and no nail problem. Movements at other joints were normal. The radiographs showed were short and broad metacarpals. The phalanges were short with shortening most marked in distal phalanges (Figure 1b). Laboratory examinations, including complete blood count, serum electrolytes, liver function tests, alkaline phosphatase levels, somatomedines and thyroid function tests were all within normal limits. Both a dopamine stimulation test and an insulin tolerance test revealed a normal growth hormone response. Ophthalmological examination was normal. He was diagnosed as acromicric dysplasia due to clinical findings.

DISCUSSION

In 1986, Maroteaux et al described a novel bone dysplasia in six unrelated children presenting with short stature, short hands and feet, normal intelligence, mild facial dysmorphic features, and X ray characteristic abnormalities of the hands. They called this condition "acromicric dysplasia".[1] Only a few cases with features of acromicric dysplasia have been reported since then, but all children presented

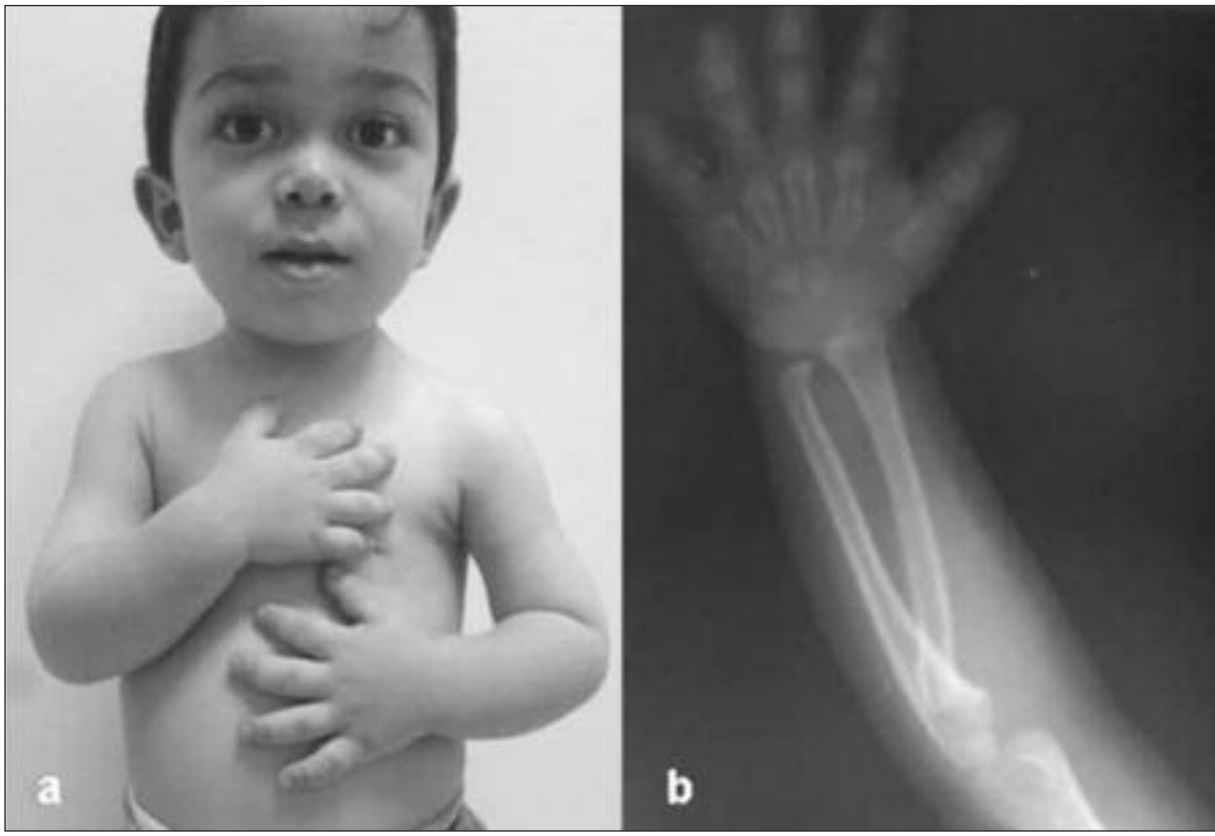

FIGURE 1a: Characteristic face with narrow palpebral fissures, short stubby nose and anteverted nostrils and short hands-feet without rhizomelic shortness of arms and legs. **1b:** Short and stubby metacarpals and phalanges.

overlapping features with geleophysic dysplasia or Moore-Federman syndrome.[3,4] Shortening of the tubular bones of the hands and feet is common to several syndromes, including acromicric dysplasia, Trichorhinophalangeal syndromes (Type I and II), Geleophysic dysplasia, acrodysostosis, Saldino Mainzes syndrome, Albright hereditary osteodystrophy and acromesomelic dysplasia. Other infrequent features included well developed muscles, a hoarse voice, generalised joint limitation in some patients, frequent ear, tracheal, and respiratory complication, and spine abnormalities. Our patient had a dysmorphic face with abnormally short hands and feet without rhizomelic shortness. No signs of visceral storage were found, which rules out geleophysic dwarfism. Achondroplasia's common features include disproportionate short stature with short limbs, particularly rhizomelic shortening, true megalencephaly with hydrocephalus in a minority, midface hypoplasia, a trident hand configuration, and joint hyperextensibility. However; our patient did not have any typical features of achondroplasia such a as a small thorax, trident configuration of the fingers, and rhizomelic shortness of arms and legs, vertebral defects including U-shaped vertebrae and narrowing of the interpediculate distance.

The child described above had characteristic clinical and radiological features of acromicric dysplasia. The main features of acromicric dysplasia are short stature, short hands and feet, and a characteristic facial appearance. Therefore, acromicric dysplasia can be differentiated from the other skeletal dysplasias due to clinical and radiologic features only.

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