ACRODYSOSTOSIS (THE FIRST CASE FROM TURKEY)

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ÖZET: Kliniğimizde acrodysostosis tanısı konulan 17 yaşında bir erkek olgu sunulmuştur. Hastada özel yüz görünümü, periferik kemik bozuklukları ve kon epifizlerin varlığı ile laboratuvar testlerinde biyokimyasal anormalliklerin bulunmayışı tanıyı desteklemektedir.

ABSTRACT: Özden ANAL, Galip KÖSE, Nevbahar TANELİ, Department of Pediatrics, Faculty of Medicine, Dokuz Eylül University, Izmir. Acrodysostosis (The first case from Turkey).

A seventeen-year-old male is presented as a case of acrodysostosis. Diagnosis is established upon findings of characteristic facial features, severe generalized peripheral dysostosis and cone epiphyses along with normal results obtained at routine laboratory examinations.

Key words: Short stature, Peripheral dysostosis, Cone-shaped epiphyses
Anahtar sözcükler: Boy kısalığı, Periferik disostoz, kon epifizler

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Acrodysostosis is a rare syndrome first described by Maroteaux and Malamut(1) after Giedion has called attention to cone-shaped epiphyses in 1967 (2). In 1971 Robinow et al(3) presented twenty cases from different parts of Europe and United States of America. Severe generalized peripheral dysostosis is a cardinal feature of the disease. All the tubular bones of the hands and feet are involved. The epiphyses are affected so growth is compromised. Short stature is found in almost all the cases. Biochemical abnormalities have not been detected. A case is described here displaying the typical features of the disease.

CASE REPORT: A seventeen-year-old white male was born at term but of low birth weight. His health status, motor and verbal development were described to be normal by his mother as compared to his peers. He had managed only to complete his primary education. He was first seen when

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seventeen years old because of complaints regarding his short stature. His height (149 cm) and weight (48 kg) were both below the fifth percentile. The head circumference was 52cm. He had distinct facial features with heavy eyebrows, flat cheeks, a bulbous nose and slightly antevert nostrils (Figs.1a,b). Macroglossia and high arched palate were also noted. There were too many pigmented nevi on the face and upper part of the trunk. His hands were short with stubby fingers and hypoplastic fingernails. The skin and soft tissues appeared to overgrow the underlying bones giving the fingers a bulging appearance. Similar findings were observed in feet and toes (Figs.1c,d). The upper extremities presented cubitus valgus and pronation was limited at the forearms. The external genitalia were observed to be normal for his age.

In the laboratory investigations the results of his routine blood count, blood chemistry and urinalysis were unremarkable. X-ray examination showed short metacarpals and metatarsals and phalangeal cone shaped epiphyses (Fig 2). A relative hyperplasia of the first ray in the feet were noted.

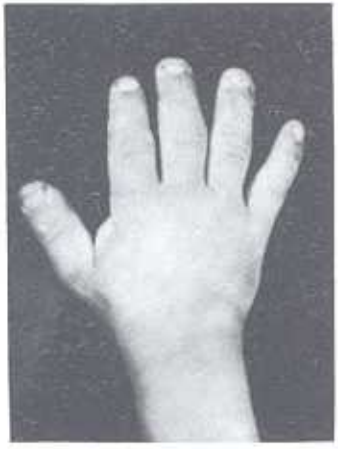
DISCUSSION: In acrodysostosis there is marked peripheral shortening of the bones of hands and feet with cone epiphyses and early epiphyseal fusion(4). Intrauterine growth retardation is present and growth failure is progressive(3). The symptoms of this disease become more apparent as the child grows older. Reiter(5) describes acrodysostosis in a 4 1/2 year old boy with peripheral dysostosis, nasal hypoplasia, mental retardation and impaired hearing. The face of the patient is quite characteristic with a small bulbous nose, maxillary hypoplasia, flat cheeks and mandibular prognathism. Robinow et al(3) report that in their series three adult patients had numerous pigmented nevi, a finding observed also in the present case. Sexual development is normal in some patients while incomplete in others. Normal results are obtained in chromosomal analyses and routine laboratory tests.

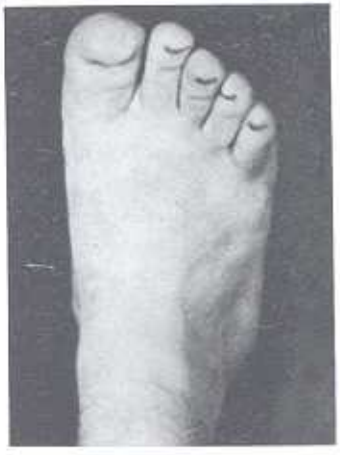
Poznansky et al(4) have examined the pattern of length alterations in the hand bones in cases of acrodysostosis, pseudohypoparathyroidism (PHP) and pseudo-pseudohypoparathyroidism (PPHP). In fact, the differential diagnosis of acrodysostosis includes mainly these two disorders. The pattern profiles for PHP and PPHP show similarities to that seen in acrodysostosis although in these two former conditions shorter metacarpals tend to be restricted to the ulnar side of the hand and phalangeal shortening is relatively mild. As discussed by Reiter(5) a radiological and clinical distinction between acrodysostosis and PHP-PPHP has been made by Giedion and Robinow et al. According to the authors the changes in hands and feet are more marked and the face is more characteristic in acrodysostosis. The soft tissue and intracranial calcifications, biochemical abnormalities in PHP-PPHP are not seen in

this entity.

Our case displays the typical findings in various degrees. The patient's laboratory tests do not reveal any pathological finding. Skeletal malformations are observed on x-ray. This case has so far been the first one reported from Turkey. Almost all of the cases of acrodysostosis reported are sporadic and the presence of a hereditary etiology is still not clear. In the family of this patient no other member with a congenital abnormality is defined.

Fig. 1. a, b. Facial appearance





c and d. Hand and foot short and broad with hypoplastic nails

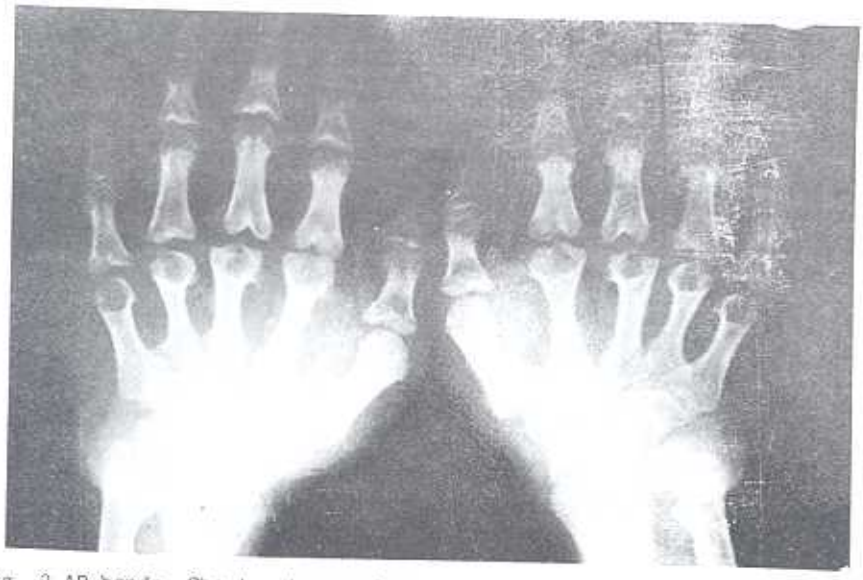


Fig. 2 AP hands. Short metacarpals and phalang... ...shaped epiphyses

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