

Type E Brachydactyly with Short Stature in a Mother and Her Two Daughters (A Case Report)

Alişan YILDIRAN, MD.¹, Ayşenur ÖKTEN, MD.², Yusuf GEDİK, MD.²,

¹Department of Pediatrics, Faculty of Medicine, Ondokuz Mayıs University, SAMSUN

¹Department of Pediatrics, Faculty of Medicine, Karadeniz Technical University, TRABZON

- ✓ Type E brachydactyly is a digital malformation that characteristically causes an asymmetrical shortening of one or more metacarpals or metatarsals or both. Although commonly seen as part of a syndrome, it can be inherited as an autosomal dominant characteristic and has been classified into various patterns. Bilginturan et al firstly reported the brachydactyly-short stature-hypertension syndrome in a Turkish family. Schuster et al. mapped the gene causing this condition on 12p. We present a mother and her two daughters with mildly short stature-normal intelligence-normal blood pressure and Type E brachydactyly, and there was no chromosomal aberrations by using cytogenetical techniques.

Key words: Type E brachydactyly, short stature

- ✓ **Bir Anne ve İki Kızında Boy Kısalığı ile Birlikte Tip E Brakidaktili (Olgu Sunumu)**
Brakidaktili bir veya daha fazla metakarp ve/veya metatarsın asimetrik kısalığı ile karakterize bir parmak malformasyonudur. Çoğunlukla bir sendromun parçası olmakla birlikte otozomal dominant geçişlidir ve çeşitli şekillerde sınıflandırılır. Bilginturan ve ark. bir Türk ailesinde brakidaktili-kısa boy-hipertansiyon sendromunu tanımladılar. Daha sonra Schuster ve ark. bu duruma neden olan genin 12p üzerinde olduğunu gösterdiler. Tip E brakidaktili'li hafif kısa boylu-normal zekalı-normal tansiyonlu bir anne ve iki kızı sunulmaktadır. Sitogenetik olarak bir anomali saptanmadı.

Key words: Tip E brakidaktili, kısa boy

INTRODUCTION

Brachydactyly has been classified into various patterns (Table), and some of these patterns can be found as a component of some syndromes⁽¹⁾. In Type E brachydactyly, the primarily shortened bones are metacarpals, especially 4th and 5th, it is inherited autosomal dominantly⁽¹⁾. Type E brachydactyly can be accompanied to hypertension⁽²⁾, to pseudohypoparathyroidism/pseudopseudohypoparathyroidism (PHP/PPHP)⁽³⁾, to atrial septal defect⁽⁴⁾, to exostoses of tibia⁽⁵⁾ and to dysarthria-mental retardation-progressive spastic paraplegia⁽⁶⁾.

We present in this short report a mother and her two daughters with mildly short stature-normal intelligence-normal blood pressure and Type E brachydactyly.

CASE REPORTS

Case 1: A 17-year-old girl was admitted to our clinic with the complaint of short stature. Her physical examination showed a proportionate short stature (length 147 cm, <3 p and weight 50 kg, 10-25 p) with remarkable short hands. She had normal intelligence, and regular menstrual cycles. Her secondary sexual characteristics were in

Table I. Brachydactyly Types and Typical Clinical Features⁽¹⁾.

	Inheritance	Typical hand findings
Type A1	Autosomal dominant	The hands are broad and all of the fingers are short. The most severely shortened bones are the middle phalanges that may be fused with the distal phalanges.
Type A2	Autosomal dominant	The characteristic finding is of a short index finger and 2 nd toe.
Type B	Autosomal dominant	The terminal portion of the index to the 5 th and the toes show absent or rudimentary nails. The big toes and thumbs are separated.
Type C	Autosomal dominant	Fingers 2 and 3 are obviously short and 4 th finger is the longest finger in the hand.
Type E	Autosomal dominant	The primarily shortened bones are metacarpals, especially the 4 th and the 5 th .
Type Pitt	Autosomal dominant	Shortening of the 4 th and 5 th metacarpals and distal phalanges of the same digits are hypoplastic and the nails of these digits are markedly concave.

Tanner stage V. Her arterial blood pressure was 124/76 mmHg.

Laboratory studies revealed unremarkable complete blood count, urinalysis and biochemical analysis for kidney and liver functions. Serum Ca⁺ was 10.2 mg/dl (8.8-10.8), P 3.5 mg/dl (2.7-4.7), alkaline phosphatase 102 u/L (50-130), PTH 87 pg/ml (51-217), FSH 0.82 mIU/mL (1-9), LH (2 mIU/mL (2-12), estrogen 21 µg/24h (5-25), T₃ 323 µg/ml (230-660), T₄ 11 µg/ml (5-12), TSH 3 µg/ml (2-10), Glucose 78 mg/dl (70-105) and rheumatoid factor was negative. Her blood smear was normal. Chromosomal analysis on a peripheral lymphocyte culture revealed a 46, XX, karyotype, confirmed after C (centromere) banding.

The radiograph of hand showed remarkable shortening in the 3rd, 4th, and 5th metacarpals as demonstrated in Figure 1. Her vertebral, pelvic, long bone and feet radiographs were normal.

Case 2: A 15-year-old sister of the index case was 154 cm (10-25 p) in height. Her body weight was 47 kg (10-25 p) and arterial blood pressure 132/56 mmHg. She had normal intelligence and sexual

development. Laboratory findings were unremarkable except for the shortening of the 5th metacarpal bone of the left hand (Fig. 2).

Case 3: The mother of the case was 154 cm (5-10 p) in height and showed similar shortening of the 4th and especially 5th metacarpals (Fig. 3). Her body weight was 55 kg (25-50 p) and blood pressure was 125/67 mmHg.

DISCUSSION

Digital malformation that causes an asymmetrical shortening of one or more metacarpals or metatarsals or both is characteristic for Type E brachydactyly. It can be inherited as an autosomal dominant characteristic, however, commonly seen as part of a syndrome. Brachydactyly has been classified into various patterns (Table)^(1,7).

A mother and her two daughters with mildly short stature and Type E brachydactyly were presented here. This family showed autosomal dominant inheritance and their specifically short bones were metacarpals. In the light of these clinical findings, brachydactyly Type E was diagnosed^(1,8).



Figure 1. Propositus's hand and wrist radiograph showed remarkable shortness of 2nd, 3rd and 5th metacarpals.



Figure 2. The radiograph of the hand of the index case's sister showed shortness of the 5th metacarpal.

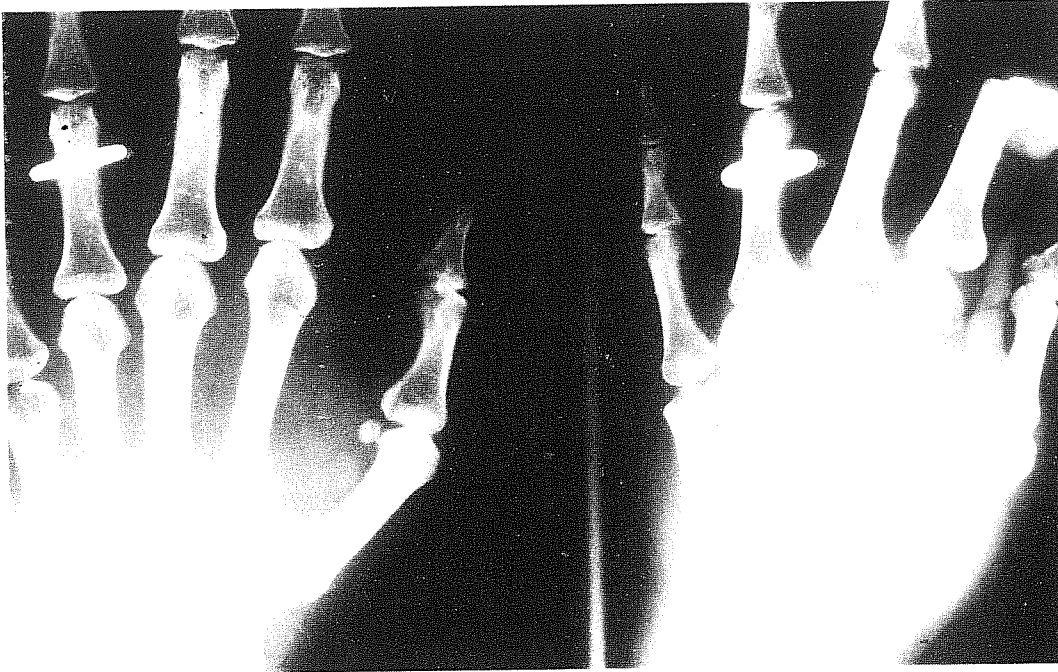


Figure 3. The mother of the case's hand radiograph, shortening of the 4th and especially 5th metacarpals.

Of cases with limb anomaly shows 30%-80% chromosomal aberrations. The expressivity of these anomalies covers a wide range within the morphogenetic pattern because of variable gene expressivity. No entirely specific malformation type is seen⁽¹⁾. We did not find any chromosomal aberrations by cytogenetical techniques.

Bilginturan et al first reported the brachydactyly-short stature-hypertension syndrome in a Turkish family⁽²⁾. Shuster et al. mapped the gene causing this condition on 12p⁽⁹⁾. Czeizel et al.⁽⁴⁾ reported a family were affected with the combination of brachydactyly type E and heart defect, mainly atrial septal defect, type II. Ikegawa et al.⁽⁵⁾ reported disproportionate short stature-type E brachydactyly-exostoses of tibiae in a patient with an XYY karyotype. Lacassie et al.⁽⁶⁾ reported on concordantly affected female identical twins with mental

retardation-dysarthria-progressive spastic paraplegia-brachydactyly type E. Recently, Chitayat et al. named the brachydactyly-short stature-hypertension syndrome as Bilginturan syndrome^(10, 11).

The present family had only mildly short stature accompanying to brachydactyly Type E.

However, an Albright hereditary osteodystrophy (AHO)-like syndrome including brachydactyly type E and mental retardation may be caused by (micro) deletions at chromosome 2q37, this region together with the AHO locus at chromosome 20q13 were considered as candidate loci for brachydactyly type E⁽⁷⁾. Although, we did not find any chromosomal aberrations by cytogenetical techniques, further genetic evaluations by using DNA analysis should be done to determine the syndrome the family has.

In conclusion; complete physical

examination of the patient, evaluation of family members, measuring of blood pressure should be absolutely performed in a patient with brachydactyly. Genetic counselling should also be given to such families.

Geliş tarihi : 25.09.2000

Yayına kabul tarihi : 21.05.2001

Correspondence:

Dr. Alişan YILDIRAN

Ondokuz Mayıs Üniversitesi, Tıp Fakültesi,

Çocuk Sağlığı ve Hastalıkları Anabilim Dalı

55139 Kurupelit, SAMSUN

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