The Turkish Journal of Pediatrics 31:249 - 252, 1989

ANONYCHIA ASSOCIATED WITH ECTRODACTYLY SYNDROME: A CASE REPORT*

*Türkân Patıroğlu MD***, Enver Hasanoğlu MD****

Key words: anonychia, ectrodactyly

Anonychia associated with ectrodactyly is a rare congenital anomaly which is inherited as an autosomal dominant trait[1]. It was first described by Lees et al[2], in 1957. Ectrodactyly, which is the occurrence of bizarre digital anomalies, has also been noted to be associated with anonychia[2,3]. We report a patient with anonychia associated with ectrodactyly, who also presented with microcephaly.

Case Report

A two-month-old female infant was brought to the Pediatric Department of Erciyes University Hospital with anonychia and a deformed hand. She was the fourth child of healthy unrelated parents. The mother's history revealed no hereditary congenital anomalies. She had not been subjected to roentgenograms nor had she taken any medication during her pregnancy. The patient had two healthy brothers and a sister. The mother was unaware of anonychia in her or her husband's family.

Physical examination revealed an infant weighing 3950 g (10^{th} percentile), height 55 cm (10^{th} percentile), and head circumference 33 cm (below the 3^{rd} percentile). There was an absence of all nails except for the fifth nail of the left foot. Both her thumbs and great toes were shorter than expected. Moreover, she exhibited syndactyly between the fourth and fifth digits (Figs. 1 and 2). All other findings of the physical examination were unremarkable. Roentgenograms of the hand revealed an absence of the distal phalanges of the thumbs, and the distal parts of the other digits were not developed (Fig. 3). Craniosynostosis was not observed on the skull X-ray. Urine and blood amino acids were found to be within normal limits.

* From the Department of Pediatrics, Erciyes University Faculty of Medicine, Kayseri.

** Assistant Professor of Pediatrics, Erciyes University Faculty of Medicine.

*** Professor of Pediatrics, Erciyes University Faculty of Medicine.

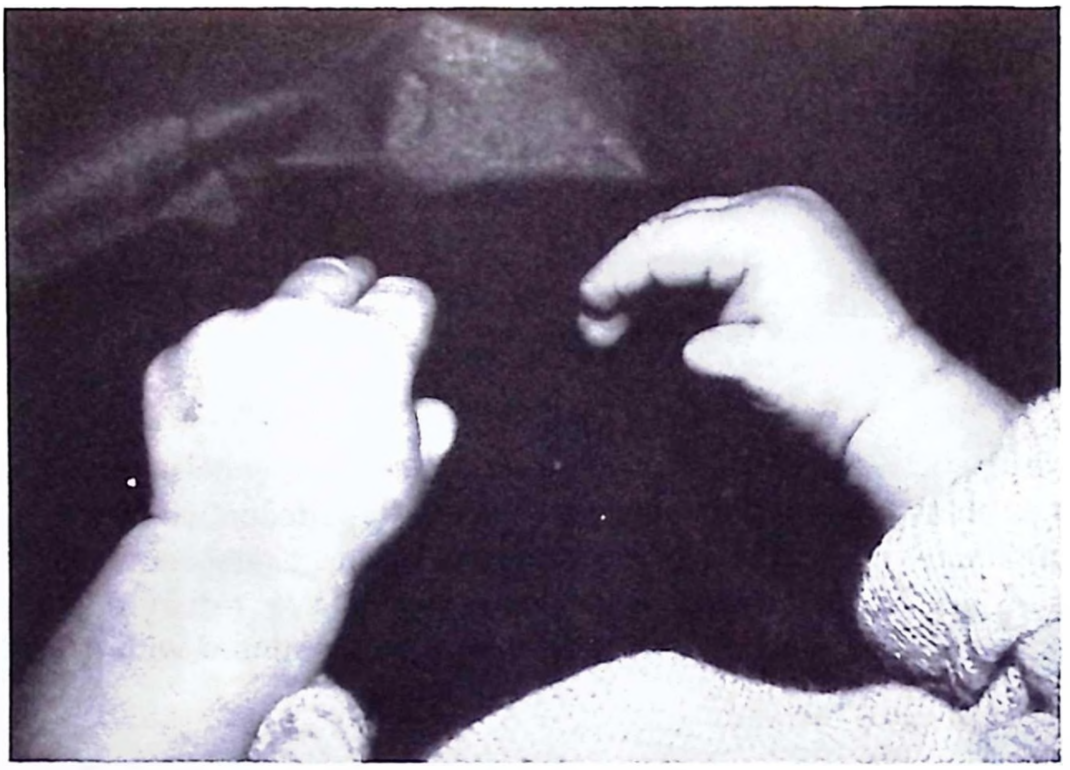

Fig. 1: The patient's hands showing ectrodactyly.

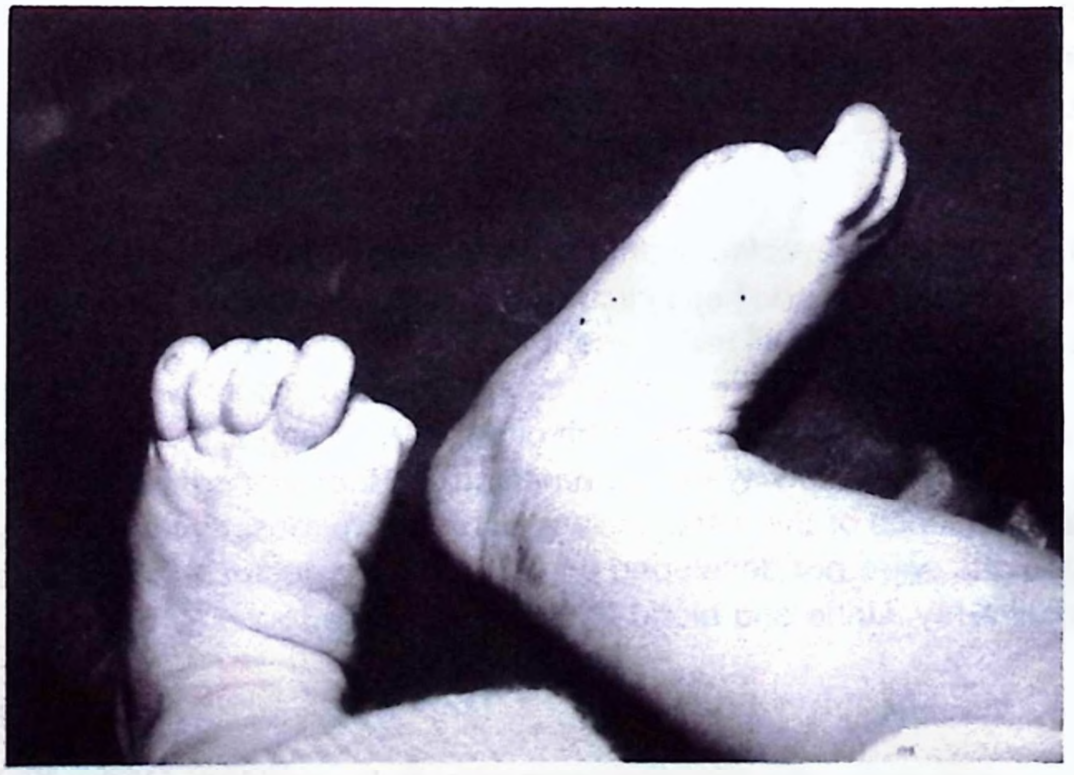

Fig. 2: The patient's feet showing anoncyhia with ectrodactyly.

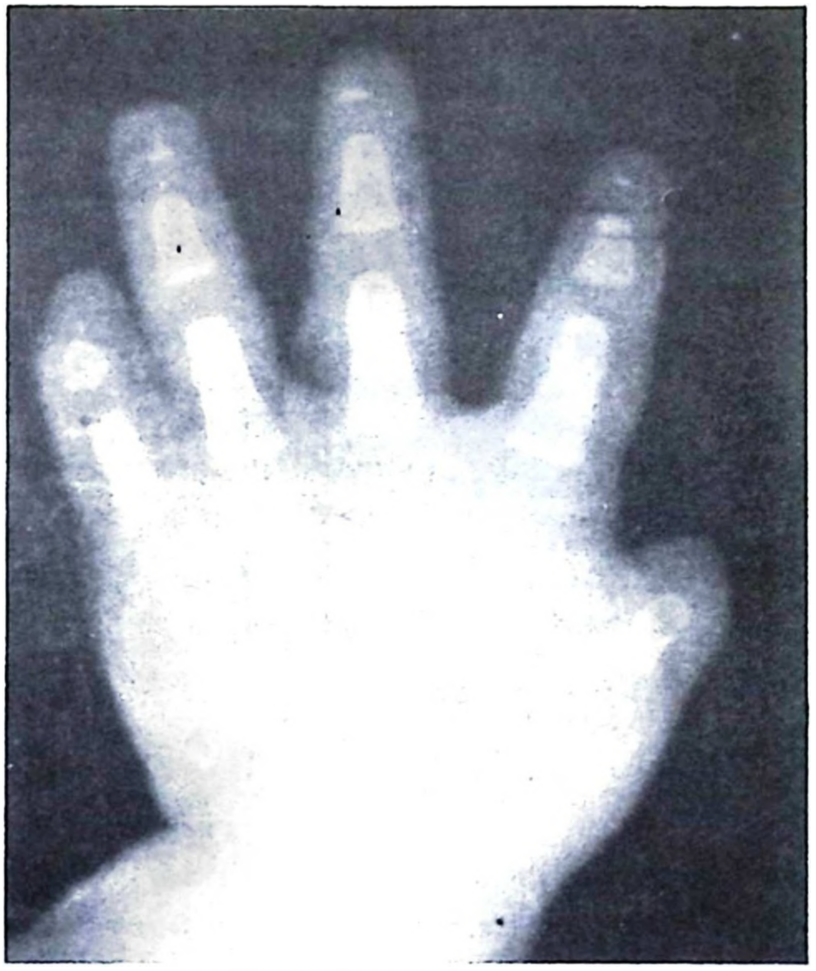

Fig. 3: X-ray of the hand.

Discussion

Cockayne[4] classified anonychia into four types: Type I, the most common, is characterized by a recessive mode of inheritance; Type II is transmitted on an autosomal dominant basis; Type III is characterized by an absence of thumb nails and red, sensitive nail beds, and Type IV comprises the nail-patella syndrome. In 1957, Lees et al[2] reported members of a large family having anonychia occurring with bizarre bony anomalies of the digits while Rahbari et al[5] reported a three-week-old boy with anonychia and associated ectrodactyly.

Our case was diagnosed as having anonychia associated with ectrodactyly since except for the fifth nail all the nails were absent on the left foot. In addition, the thumbs and great toes were shorter than they should have been. There was syndactyly between both of the patient's fourth and fifth digits. All these findings were supported by X-ray studies.

Even though this syndrome is inherited on an autosomal dominant basis[1,6], we were not able to find a case of anonychia with ectrodactyly in the patient's kindred, as had been reported by Rahbani et al[5] in his study.

Our patient was considered to be microcephalic since the head circumference was smaller than expected (below the 3rd percentile). However, the cause of microcephaly could not be determined by physical or laboratory findings.

In our search of the literature, cases of anonychia with congenital deafness or skin lesions have been reported but we could not find a case of anonychia with ectrodactyly and microcephaly. Our case of anonychia with ectrodactyly was coincidentally associated with microcephaly.

Summary

Anonychia with ectrodactyly is a rare inherited autosomal dominant syndrome. A case of a two-month-old female infant presenting with anonychia in association with ectrodactyly and microcephaly is presented.

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