Brief Clinical Report

Acheiropodia: Report on Four New Brazilian Patients

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We report on 4 new patients with acheiropodia ("the handless and footless families of Brazil"). This autosomal recessive condition involves all 4 limbs with a well-defined pattern of defects. Two of the patients described here had a small bone fragment in the upper stumps (Bohomoletz bone), an uncommon finding in acheiropodia.

KEY WORDS: acheiropodia, limb congenital anomaly, autosomal recessive inheritance, consanguinity

INTRODUCTION

The primary manifestations of acheiropodia are the almost uniform absence of the forearm, including elbow joint, defective development of the tibiae beyond their proximal two thirds, absence of the fibulae and feet, and occasionally the presence of an unusual elongated small bone on the tip of the upper limbs. There is a higher than usual frequency of consanguineous marriages among parents with affected children [Freire-Maia and Chakraborty, 1975; Freire-Maia, 1981] and, since the first description of acheiropodia in 1929 by Peacock, it remains known only in Brazilian patients [Grimaldi et al., 1983].

Here we report on 4 new patients with acheiropodia, 2 possessing a Bohomoletz bone, and 2 born to consanguineous parents.

CLINICAL REPORTS

Patient 1

GRS (Fig. 1, an 11-year-old boy, was the sixth child of a 41-year-old G6P6 woman and her 52-year-old unrelated husband, but with their relatives living in the same region through several generations. There were no similar cases in close relatives. Pregnancy was normal. There was absence of trauma or exposure to toxic or infectious agents or to X-rays. The child had a normal term delivery. Birthweight (BW), length (TBL), and occipitofrontal circumference (OFC) were not recorded. Limb anomalies were noted at birth.

Examination at age 11 years showed a tetramelic deficiency with terminal transverse hemimelia at the level of the left elbow and a fingerlike appendage located distally at the tip of the right elbow; in the lower limbs, hemimelia involved the distal portion of both tibiae.

Radiological examination showed abnormally modeled distal humerus with lack of differentiation of the condylar regions, the presence of a short and hypoplastic bone, with a triphalangeal complement on the right (Bohomoletz bone). The tibiae were abnormally modeled with transverse deficiency of their lower third; fibulae and feet were absent.

Patient 2

WSB (Fig. 2), a 1-year-old boy, was the first child of a 17-year-old G1P1 woman and her 18-year-old unrelated husband. There were no similar cases in close relatives. Pregnancy was normal. There was absence of trauma or exposure to toxic or infectious agents or to X-rays. Delivery occurred at 32 weeks gestation through cesarean section. BW was 1,930 g, TBL was 42 cm, OFC was not recorded, and limb anomalies were noted at birth.

Examination at age 1 year showed bilateral terminal transverse hemimelia at the elbow and transverse hemimelia at the middle third of the tibia. Radiological examination showed an abnormally modeled distal humerus with terminal transverse hemimelia at the elbow and bilateral transverse hemimelia at the middle third of the tibiae.

Patient 3

ARS (Figs. 3, 4), a 2-year-old girl, was the first child of a 17-year-old G1P1 woman and her 22-year-old related husband (second cousin). Pregnancy was normal. There was absence of trauma or exposure to toxic or infectious agents or to X-rays. Vaginal delivery was at 32 weeks gestation. BW was 1,600 g; TBL and OFC were not recorded. Limb anomalies were noted at birth.

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Examination at 2 years showed a well-developed metacarpal-like structure at the tip of both elbows with a triphalangeal complement and transverse hemimelia at level of the middle third of both tibia. Radiological examination showed bilaterally mild hypoplasia of the distal portion of the humerus and the presence of the Bohomoletz bone with a distal triphalangeal complement, bilateral transverse hemimelia at the middle third of the tibiae, with absent fibulae.

**Patient 4**

LPD (V-1) (Figs. 3, 5), a 7-month-old girl, was the first child of a 22-year-old G1P1 woman and her 26-year-old related husband (second cousin). Pregnancy was normal. There was absence of trauma or exposure to toxic or infectious agents or to X-rays. The child had a normal term delivery. BW was 2,20 g, TBL was 44 cm, and OFC was not recorded. Limb anomalies were noted at birth.

Examination at age 7 months showed terminal transverse hemimelia at the level of the elbows, and transverse hemimelia at the level of the middle third of both tibiae. Radiological examination showed abnormally modeled condylar region of both humeri with transverse hemimelia at the elbows, transverse hemimelia at level of the middle third of both tibiae, and absent fibula and foot.
DISCUSSION

Acheiropodia is a rare severe autosomal recessive condition [Freire-Maia, 1975; Freire-Maia et al., 1975a,b]. There is a higher than usual frequency of consanguineous marriages among the unions, which produce affected children [Freire-Maia and Chakraborty, 1975]. Since its first description, only Brazilian patients have been reported. The gene is fully penetrant, and expressivity is relatively constant, although some variability can be found not only among different individuals, but interlaterally in the same affected individual [Freire-Maia, 1975]. No other pleiotropic or heterozygous effects has been reported.

One of the most important variables is the occurrence of the Bohomoletz bone, an elongated small bone on the tip of the stumps of the upper limbs, parallel to the axis
of the humerus [Freire-Maia et al., 1978; Grimaldi et al., 1983]. The origin of this bone remains unknown; in some cases, it looks like a proximal portion of the ulna, while in others, it shows little differentiation remaining as an abnormally modeled and hypoplastic bony fragment [Grimaldi et al., 1983]. Another variable is the level of the deficiency of the upper limbs and the humeral diameter from top to bottom, ending in a funnel shaped close to the terminal part, or being smoothly tapered [Freire-Maia et al., 1978].

The present report on these 4 patients describes these classic variations concerning the presence of the Bohomoletz bone (patients 1 and 3), the abnormalities observed in the humerus (patients 1, 2, 3, and 4), the interlateral variability of the anomalies, as well as the presence of parental consanguinity in 2 of the reported patients (patients 3 and 4).

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REFERENCES


