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Short Report

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Digital Amputations Caused By Amniotic Bands a Case Report

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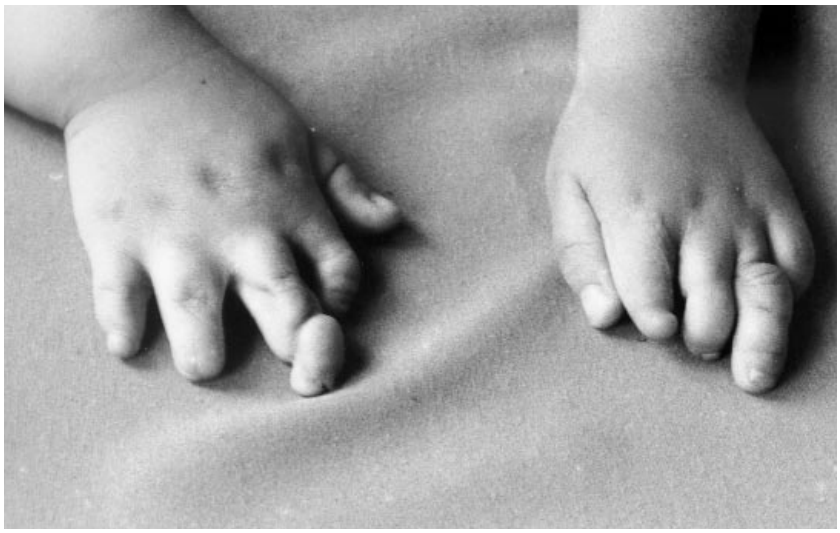


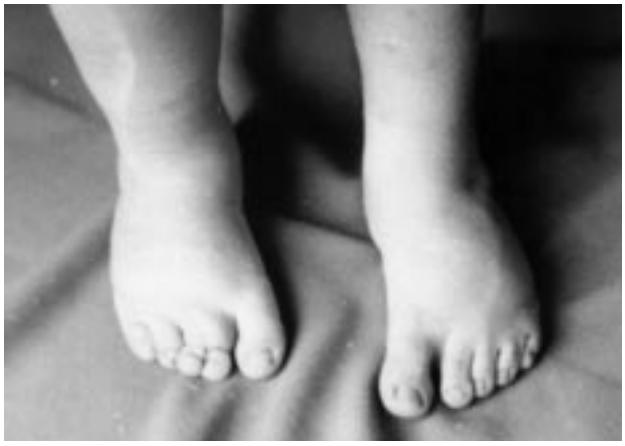
Figure 1. Dorsal view of the hands. Not complete and incomplete amputations.

Introduction

Amniotic band sequences are generally a sporadic condition. The presence of amniotic bands at birth is helpful in making a diagnosis. The incidence of digital amputations occurring sporadically due to amniotic bands is very low (1). Its frequency has been reported as being one in 10 000 - 45 000 births (2, 3). Although digital amputation is the main malformation observed in such cases, cranio-facial malformations are sometimes associated with the deformation (3). The cause of amniotic band sequence is not known. Presently the most widely accepted etiological explanation is that amnion rupture occurs in the first trimester of pregnancy, and fetal parts become entangled in bands which form between a torn amnion and the chorionic mesenchyme. However, not all associated anomalies can be explained by this hypothesis. Of the possible causes, amniocentesis and chorionic villus sampling have been discussed (4). In this report we present an example considered interesting with respect to genetic counselling because of additonal foot deformity.

A 3-and-a-half-year old male was referred to out genetics department because of congenital limb deformities. He was the only child of the family. His mother had not been exposed to any drugs, X-rays or infectious agents during pregnancy. Delivery at 40 weeks gestation was uncomplicated. The birth weight was 3800 g (in the 90th percentile) and length 52 cm (in the 75th percentile). The family history was unremarkable. He had been found otherwise normal at birth.

Physical examination revealed a boy of normal



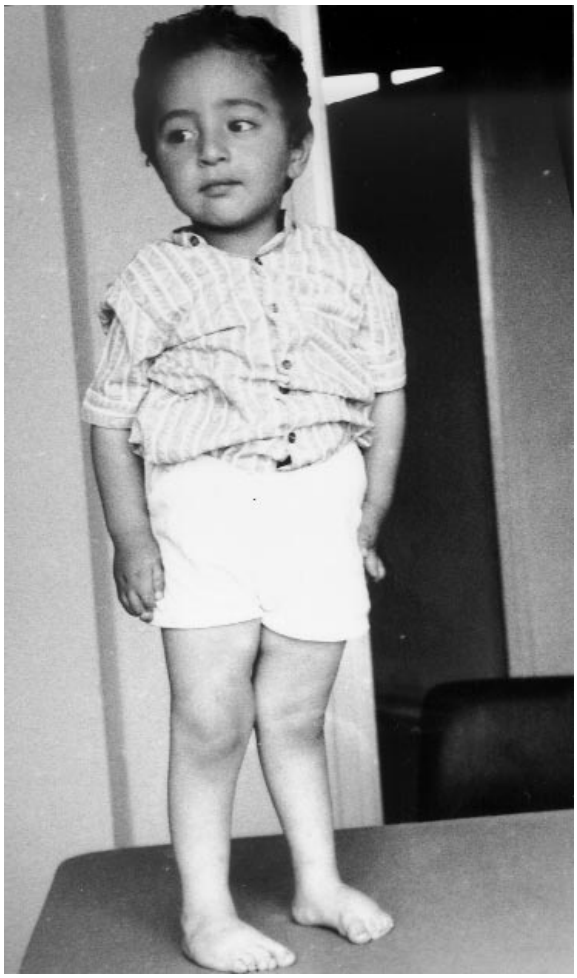


Figure 2. Syndactyly between 2nd and 3rd toes of right foot.

appearence with normal somoto-mental development, his height being 106 cm (in the 90th percentile) and weight 17.5 kg (in the 75th percentile). The head and neck were also normal except for a mobile preauricular mass on the left.

On the right hand the thumb was normal, while the second, fourth and fifth fingers were amputated through the distal interphalangeal joints; in the third finger the amputation was incomplete (Figure 1). On the left had, the first finger was also normal and the second, third and fifth fingers were amputated through the distal interphalangeal joints while there was a furrow on the proximal interphalangeal joint of the fourth finger (Figure 1). The amputation was apparently incomplete. The patient also exhibited pes varus deformity in both feet and equine deformity, regressed by treatment. On the right foot there was syndactyly and construction between the second and third toes (Figure 2).

Routine laboratory analyses including karyotping were within the normal limits. Pedigree analysis was not found to be contributory.

Discussion

There were both complete and incomplete amputations of the digits on both hands due to amniotic bands, associated with pes equinovarus deformity and unilateral syndactyly. Various theories have been proposed to explain the etiology of malformations observed in such instances, which were sporadic in almost all cases (5). These deformities are related to amniotic membranes. It has been suggested that traumas to the amniotic sack are the major causes of bleeding into the amniotic cavity in the third trimester (1, 6). Several experimental studies supporting traumatic etiology after the performing of amniocentesis have been reported (5).

Although the most frequent organs involved in this process are the fingers, these occur with some other anomalies, such as drop-foot and cleft palate and lip (7, 8). In our case, pes equinovarus deformity of both feet (only varus after correction) occurred with incomplete syndactyly of one foot associated with finger deformities. This has not been reported before. Apparently, this would lead to some confusion so far as genetic counselling is concerned (9). We estimated that the risk of recurrence of these anomalies is approximately 2 percent (8).

Congenital amputations should be distinguished from congenital hypoplasia and/or aplasia. The latter are due to transverse defects and hypoplasias are usually caused by monogenic mutations. In general, monogenically determined syndromes are severely damaging to the fetus, with a high risk of recurrence (often 25%)/ (8). Ultrasound plays a leading role in prenatal diagnosis of both transverse defects and amniotic bands and their complications. Amputations due to amniotic bands are

generally asymmetrical (8). Clinical findings and even anamnesis should enable differential diagnosis in most cases. Differentiation is important in genetic counselling and evaluating the risk of recurrence.

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