

## Congenital Amputation Involving the Hands and Feet: A Case Report

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### Abstract

**Background:** Congenital amputation of the limbs is not uncommon. However, it is very rare when this involves both the upper and lower limbs.

**Method:** This is a case report of a child who presented with congenital amputation involving both the upper and lower limbs.

**Results:** The patient was a 10 day old baby girl that was delivered by a 21 year old woman. She is the first and only child of the woman, whose pregnancy was uneventful and was carried to term. There is no family history of congenital anomalies. The findings on examination were: amputation of the index, middle and ring fingers at the level of metacarpophalangeal joints on both hands and a partial amputation (at the level of the middle phalanx) of the left little finger. There were forefoot amputations on both lower limbs. Scars were noticed over the amputation stumps with no associated congenital anomaly.

**Conclusion:** Congenital amputation involving all limbs as an isolated entity is a rare condition; the cause of which is probably as a result of congenital amniotic bands.

**Key words:** Congenital amputation, limbs, scar, amniotic bands.

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### Introduction

Congenital amputation of any limb in which there is complete absence of the distal part thereby producing an amputation-like stump has been reported by some authors<sup>1,2</sup>. Usually it is not familial and has no definite inheritance.<sup>3</sup> There is no association with any malformation syndrome.<sup>4</sup> It has been reported to be more common in the upper extremities, but very rare to find simultaneous involvement of both upper and lower limbs<sup>1</sup>. This is a report of a child who presented with simultaneous congenital amputation involving the hands and feet.

### Case Report

This is a 10 day old baby girl that was delivered by a 21 year old woman. She is the first child of the woman, whose pregnancy was carried to term. The pregnancy was uneventful except for a febrile illness she had at the 4<sup>th</sup> month of the pregnancy which lasted for 3 days. The only medication she took during the pregnancy was antenatal drugs which were essentially haematinics. There is no family history of congenital anomalies.

The main findings on examination were on the limbs (fig.1). The right hand (fig.2) had a well developed thumb and the little finger, while the index, middle and ring fingers terminate (amputation) at the metacarpophalangeal joints. On the left hand (fig.2), only the thumb is normal with partial amputation of the little finger at the level of the middle phalanx; while the index, middle and ring fingers were completely absent like that of the right hand. There was loss of the forefoot and toes in both lower limbs, forefoot amputations (fig.1). The stumps had scars marking the points of amputation:



Fig.1: Photograph showing limbs with amputation of the index, middle and ring fingers as well as fore-foot amputation of the feet with scars.



**Fig.2:** Photograph of the upper limbs showing amputation of the index, middle and ring fingers with scars at the point of amputation.

## Discussion

Congenital amputation of the limb is not uncommon but is very rare when it involves all limbs<sup>1, 2</sup>. The entity is characterised by scarring, dimple formation and occasionally an osseous projection at the end of the stump<sup>1</sup>. In our patient, there was obvious scarring at the end of the amputation stumps, a feature that support amputation of this part of the limbs. Such amputations are commoner in the upper limbs compared to the lower limbs while simultaneous occurrence in both the upper and lower limbs is very rare<sup>2</sup>. The incidence is unknown and varies from one community to another. The reason for the spontaneous amputation is unknown. There is no established cause, with no genetic basis, definitely not inherited and has no association with any malformation syndrome<sup>1,4,5</sup>.

Recently, limb amputation as a result of amniotic band syndrome has been reported by Tadmor et al<sup>6</sup>. In their report, through serial Doppler ultrasound, they

demonstrated the relationship between amniotic bands and limb amputation. The foetus they studied had amniotic band originating from the foetal membrane surrounding both limbs with associated swollen feet and amputated fingers. At birth, there was below knee amputation of the right leg and a partial amputation of the left.

Amniotic bands consist of fibrous strands that extend from the outer surface of the chorion into the amniotic cavity<sup>7</sup>. Two theories have been propounded on the aetiopathogenesis of amniotic bands<sup>8, 9</sup>. Both theories have been a subject of controversy, disagree as to whether the amniotic bands themselves cause the foetal anomalies (exogenous) or are a by-product of a primary defect in the embryonic germ disc (endogenous). Despite reports (diagnostic and therapeutic)<sup>6,10</sup>, the concept that amniotic bands cause foetal damage is still being challenged<sup>11</sup>. Even with the controversy, Schwarzler et al<sup>12</sup> have been able to demonstrate (sonographically and by foetoscopy) that amniotic bands cause congenital amputation. These bands were seen extending from the uterine wall and entangling the foetal limbs, with associated amputation of the fingers and toes. Histological examination of the bands confirmed it as amniotic bands with no evidence of fusion with the foetal skin. This feature might explain the successful in utero release of the constriction bands by Quinterero et al<sup>10</sup>.

Congenital amputations involving both upper and lower limbs are very rare. The presence of a scar over the amputation stump in a child born with congenital amputation of any part of the limb will support such diagnosis. Reports have supported the theory that amniotic bands are responsible for such amputation.

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