

# BILATERAL UPPER LIMB AMELIA IN A NEONATE IN NORTH-WESTERN NIGERIA: CASE REPORT AND REVIEW OF LITERATURE

**Abstract:** Amelia affects approximately 0.05 to 0.09 out of every 10,000 newborn babies. The complete absence of a limb may occur in isolation or as a part of multiple congenital malformations. The condition is uncommon and very little is known with certainty about the etiology, It could be also be sporadic and can be caused by teratogens such as thalidomide, alcohol, vascular compromise by amniotic bands and maternal diabetes mellitus. Amelia's genesis has also been linked to various types of heredity.

We report a term female neonate delivered by a healthy 30 year old primipara in North-Western Nigeria, with the two upper limbs completely missing at birth, other parts of the body were essentially normal. The maternal history revealed no known identifiable risk factor of Amelia. We are reporting this case because of the rarity of this congenital defect.

**Keywords:** Bilateral, upper limb, Amelia, North-western, Nigeria

## Introduction

The complete absence of a limb may occur in isolation or as a part of multiple congenital malformations [1]. Amelia affects approximately 0.05 to 0.09 out of every 10,000 newborn babies [2], while the prevalence in still birth is 1.41 in 100,000 [3]. The condition occurs due to interruption of development of limb buds between 24 and 36<sup>th</sup> days of fertilization [4]. Amelia is uncommon and very little is known with certainty about the etiology. It could be sporadic [2] and can

also be caused by teratogens such as thalidomide, alcohol, vascular compromise by amniotic bands and maternal diabetes [5]. Amelia's genesis have also been related to various types of heredity, including the autosomal dominant, autosomal recessive, and X-linked dominant modes of inheritance, indicating this condition's genetic heterogeneity [6]. In most cases, even though etiology remains unidentified, a defect in WNT3 gene located at 17q21 which regulates the development of limbs and other organs can lead to this anomaly [7]. Abnormalities associated with Amelia include severe defects of the lungs, vertebrae, heart, internal and external genital system, and anus [8].

We are reporting this case because of the rarity of this congenital defect. There is paucity of data on Amelia probably due to under reporting, under registration of birth defect and because of most pregnancies with Amelia end up as still birth [3].

## **Case report**

### **Patient history**

The baby was a term female neonate, the first child of a 30 year old healthy primipara and 36 years old father. The pregnancy was desired and spontaneously conceived, booked in our tertiary health facility at 3 months of gestation, pregnancy was uneventful and the mother was regular with ANC visits, she had IPT for malaria, tetanus toxoid, folic acid and fersolate during pregnancy. The mother had 3 antenatal scans, one scan each in the first, second and third trimester of pregnancy that revealed no fetal abnormality. The mother however ingested un-prescribed ibuprofen bought in a chemist when she had fever in the first trimester of pregnancy. No maternal history of cigarette smoking, consumption of alcohol, any known teratogenic drug or traditional medicine during pregnancy, neither was

she exposed to radiation during the first trimester of pregnancy. The mother was not diabetic, no history of chronic drug use during pregnancy and had no prenatal invasive procedure. The mother was negative for Hepatitis B and human immunodeficiency virus. There was no history of maternal febrile illness, trauma to the abdomen or vaginal bleeding during pregnancy, no family history of similar illness or consanguinity in the parents. Delivery was via EMLS CS due to failed induction of labour, post-datism and fetal distress. The baby cried immediately after birth, APGAR scores were 7 and 9 at 1 and 5 minute respectively. Meconium was passed within an hour of delivery.

Both parents had tertiary level of education and are secondary school teachers in a private school; the family monthly income is N50, 000.

### **Clinical findings**

Examination findings revealed a neonate that is not in respiratory distress, no cardiac murmur or dysmorphic features. The weight, Length and occipito-frontal circumference were 2.3Kg, 47.5cm and 35cm respectively. The neonate was otherwise healthy-looking except for the two upper limbs that were completely missing with a dimpling in both armpits noticed at birth, with normal lower limbs and female external genitalia, there were no other gross abnormalities on examination.



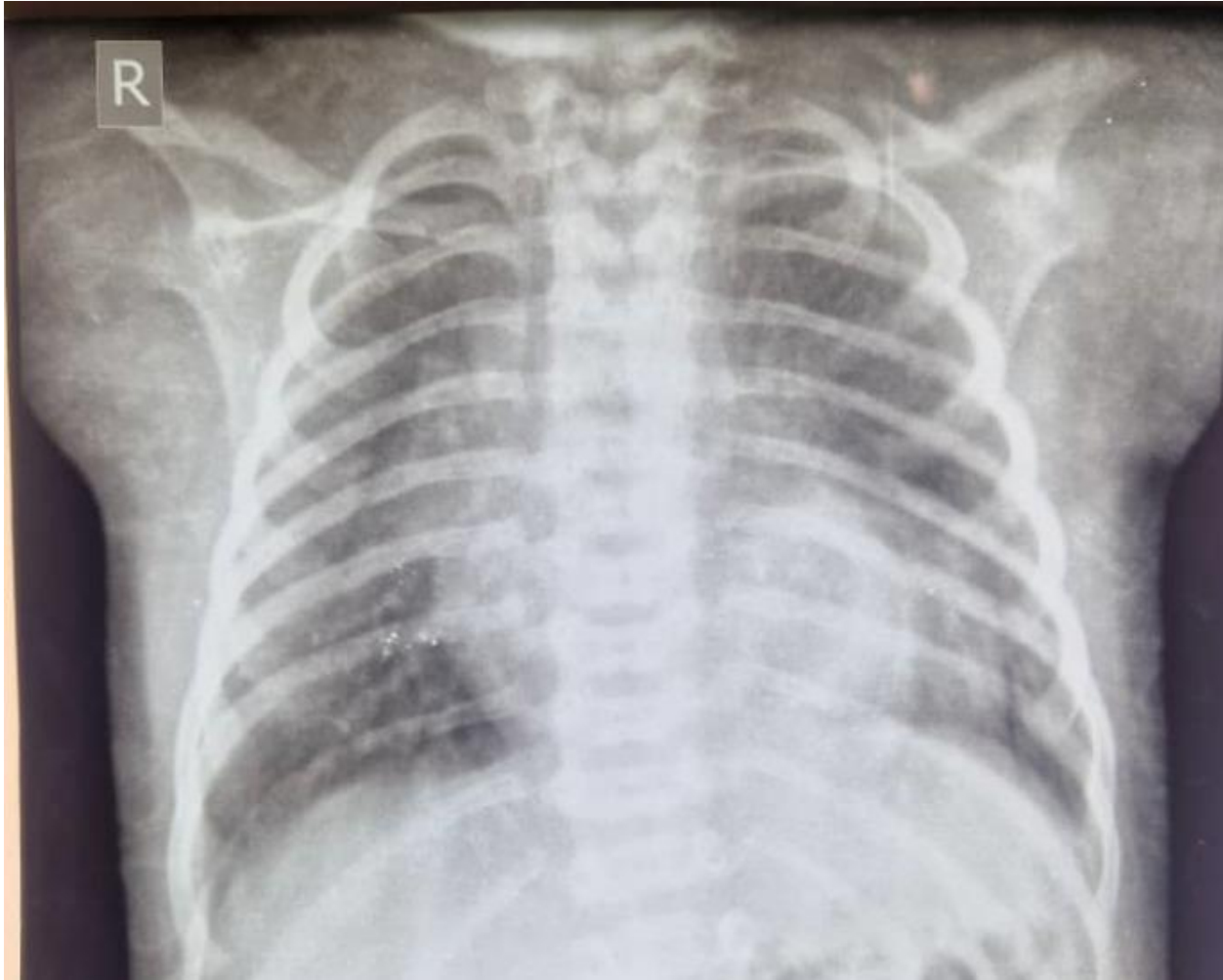
**Figure 1: Clinical image of the neonate showing complete absence of both upper limbs and normal lower limbs**



**Figure 2: Clinical image showing the complete absence of both upper limbs and a dimple in the right armpit**



**Figure 3: Clinical image of the neonate showing complete absence of both upper limbs**



**Figure 4: X-ray of the chest and both shoulders showing bilateral upper limb Amelia**

The abdominal ultrasound and Echocardiography revealed normal findings.

The neonate had uneventful nursery care for evaluation and was discharged on day five after delivery, while the baby was on admission the parents were unwilling to see the baby and were also very worried about the deformity of their child and afraid of the child being stigmatized.

The parents had several counseling sessions and were also advised to continue with follow up visit with the paediatricians, orthopedic surgeon and social welfare. The neonate was referred to an orthopedic surgeon in view of the need for prostheses as the child develops.

## **Discussion**

There are few case reports on Amelia in Nigeria; Benjamin et al [1] in Zaria, Nigeria reported absence of the upper left arm and forearm, and fusion of both big and middle toes and absence of the entire small toe on the left foot. In a 12-week-old male child delivered by a 26 years old mother with associated unilateral left cleft lip and palate. Also, Dalvi et al [2] in Nagpur, India, reported complete absent of the right lower limb and the left lower limb was hypoplastic and bilateral renal agenesis, absence of both anus and genitalia in an arbutus, delivered by a 30-year-old mother in good health, with no consanguinity or significant family history.

In IR Iran Eghbalian et al [5] reported a male neonate with the right arm completely missing with a cherry sized skin bulging on the lateral end of the right clavicle and imperforate anus, the first child of a 22 year old father and 26 year old mother. Pakkasjarvi et al [9] in Finland reported a total of 30 cases of amelia and phocomelia, of which 23 had amelia, and 7 were cases of phocomelia. Of the 23 patients with amelia, 10 were boys and 10 girls, and the gender was unknown in three cases. Thalidomide was not an etiological factor in any of the cases. In Pakkasjarvi et al [9] report, amelia affected the lower limbs in 70% of the case, 69% of the patient with amelia associated with urinary tract abnormality, 63% with axial skeletal abnormality, 56% abdominal wall defect, 56% sexual

differentiation abnormality, 50% with gastro-intestinal abnormalities. Unlike in their reports [1, 2, 5, 9], our patient had no other associated abnormality.

It is worthy of note that in most of the reports [1,3,5,9] and ours the mothers were observed to be young. The reason why the condition is commoner in children of young mothers is not known. Risk factors for amelia and studies remain mostly uncharacterized [9]. Also in some of the reports [1, 5] the neonates were males, this is in not in tandem with our report that is a female, while another study [9] reported equal gender predilection.

In our report the neonate had congenital deficiency of both upper limbs, this corroborates the finding of a multicenter study that observed that the upper limbs were more involved than lower limbs [3], but in contrast to the finding of Pakkasjarvi et al [9] that reported that the lower limbs were commonly affected than the upper limbs.

In this index case the mother had three antenatal scans, one scan each in the first, second and third trimester of pregnancy that revealed no fetal abnormality. Likewise, Richmand et al [10] reported that 3 out of 4 cases of amelia were not diagnosed inutero.

The parents of this neonate were unwilling to see their baby while on admission in the neonatal unit for the first few days due fear and guilt, sense of loss and denial. When a child is born with congenital limb deficiency it affects not only the child, but also the parents, family and friends, parents experience grief with the birth of a child with any deformity [11]. Hence parents need support and guidance from the health care providers.

One huge challenge in this patient with bilateral upper limb Amelia is the accessibility and cost of a prosthesis that is as functional and cosmetically pleasing

as possible in a resource constraint setting like ours and also having indigent parents. The functional use of prosthesis is a skill which many amputees do not have the patience, intelligence, or motivation to learn [1]. However, this child is yet to be fitted prosthesis for Amelia. However, we believe that when fitted at the appropriate age it will help the child's body image and may be essential for normal neuromuscular function and development.

## **Conclusions**

Our patient had sporadic form of Amelia. Further studies are necessary to find out the exact etiology and risk factors of Amelia. Antenatal screening, karyotyping analysis, and genetic counseling are recommended to reduce the risk of such congenital anomalies.

## **Declaration of patient consent**

Consent was obtained from the parents to publish the images and other clinical information of the patient.

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