FETAL UPPER LIMBS AMELIA: A Case Report and Literature Review

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ABSTRACT: Amelia is complete absence of a limb that presents as an isolated defect or with associated malformations. Congenital limb defects are rare fetal anomalies with a birth prevalence of 0.55 per 1,000. Amelia is an extremely rare birth defect marked by the complete absence of one or more limbs. We present the case of a female fetus born with upper limbs amelia discovered on birth from intermarriage. Our observation supports the hypothesis that this malformation is a genetic defect.

KEYWORDS: Amelia/intermarriage/ genetic defect

1. INTRODUCTION

Amelia is the complete absence of a limb, which may occur in isolation or as a part of multiple congenital malformations. It can present as an isolated defect or with associated malformations, particularly the most frequent defects associated with Amelia were other types of musculoskeletal defects, intestinal defects, some renal and genital defects, oral clefts, defects of cardiac septa, and anencephaly. The incidence of isolated Amelia with or without other limb reductions is 0.4 per 100,000 births. The condition is uncommon and very little is known with certainty about the etiology. Different causal factors like thalidomide, alcohol, amniotic band syndrome, maternal diabetes and autosomal recessive mutations have been proposed.

The diagnosis Amelia syndrome is established clinically and can be made on routine prenatal ultrasonography. We report a case of isolated bilateral upper limb Amelia from an intermarriage (consanguineous couple, parents were cousins 1st degree) discovered at birth.
because the mother didn’t do any antenatal care. The extreme rarity of this congenital anomaly has stimulated us to report this new case presented at our hospital.

2. CASE PRESENTATION

A 19 years old female with no medical or surgical past medical. As familiar history it is a consanguinity marriage, both parents are cousins (1st degree). The patient was gravida 1 para 0, admitted on emergency at 36 weeks of gestational age by last menstrual period; the patient didn’t do any antenatal care. The pregnancy was not medically followed. On admission the patient was at full dilatation, fetal heart sounds were present. The patient was directly conducted in labor ward where she delivered vaginally in cephalic presentation a female baby; APGAR 10 /10, weighing 1.650 kg, with upper limbs Amelia. Otherwise the physical exam was normal. The baby was admitted in neonatology and ultrasound was done for ruling out internal organ malformations and it was normal (Figure1).

Figure 1 Female new born with bilateral absent upper limbs

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3. DISCUSSIONS

Amelia is an extremely rare congenital anomaly with an incidence of 1.5 per 100,000 live births and 7.9 per 10,000 still births. Upper limb Amelia has an incidence of 7 per 1,000,000 live births. [4] Amelia was traditionally thought to be a sporadic anomaly with little risk of recurrence, or evidence of genetic origins.

Usually bilateral brachial Amelia is associated with other malformations such as facial clefts, renal anomalies, ear anomalies and central nervous system malformations. Such infants have a very poor prognosis and usually die within first year of life. Failure of formation of the limb primordial during early embryogenesis may be secondary to vascular, mechanical, or teratogenic exposure Everett F. With complete absence of one or more limbs occurring prior to the eighth week of gestation. Although the teratogenic potential of Thalidomide has been well documented, the spontaneous occurrence of Amelia and limb reduction defects in the general population is rare. Others causal factors are alcohol, amniotic band Syndrome, maternal diabetes. However, different modes of inheritance have been involved in the etiology of Amelia including autosomal recessive, X linked dominant and autosomal mode of inheritance which indicates the genetic heterogeneity of this condition. Amelia with multiple malformations indicates the Roberts Syndrome which is an autosomal recessive syndrome is considered as a single genetic entity and includes various morphologic defects; is not our case. In our case, a part upper limbs Amelia, the baby is normal.

The diagnosis is made by ultrasound at second trimester during morphologic exam. Gross absence of the fetal limbs is seen on ultrasound. Other malformations are also seen, such as some renal and genital defects, spine defect as spina bifida, oral clefts, defects of cardiac septa, and anencephaly. Genetic exam has also to be done; chromosomal analysis was asked to the parents after counseling.

There is possibility of the recurrence of Amelia which has been documented in only a few families. It can be the family in our case because of consanguinity. The parents are informed about recurrence, the useful of chromosomal analysis and antenatal care in other to diagnose such kind of malformations early.

4. CONCLUSION

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Amelia is a very rare form of limb reduction defect. Isolated Amelia without other limb reductions or other malformations is very rare. Among causal factors, autosomal recessive mutations are considerate. The antenatal diagnosis is made by ultrasound during the second trimester. It can be recurrent in some families.

REFERENCES


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