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Case report

Tetra Amelia Syndrome- A Case Report

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ABSTRACT

Abstract: A still born male baby with tetra amelia syndrome is reported , which showed absence of all four limb along with craniofacial abnormalities . Localised malformations of the limbs include absence of bone, extra bones , hypoplastic bones and fusions. Complete absence of a limb is called amelia , almost complete absence with a mere stub remaining phocomelia and partial absence ectromelia . The defects may be transverse or axial . The embryonic correlation of the case is discussed.

1. Introduction

Limb anomalies vary greatly, and presented as complete (Amelia) or , partial (meromelia) absence of one or more of the extremities.Tetra amelia syndrome (TAS) means absence of all four limbs associated with anomalies of CVS, CNS, GIT , urogenital , craniofacial or respiratory system. Meromelia, includes i) almost complete absence with a mere stub remaining is called phocomelia and ii) partial absence of a limb is called ectromelia . Micromelia represents abnormally short extremities[1] . Localized malformations of the limbs include absence of bones , extra bones , hypoplastic bones and fusions. The defects may be transverse or axial[2].

Two main forms of classifications were used widely i) morphologic and ii) casual . The currently used classifications of congenital anomalies of the upper limb is based on that of Swanson. Modified by the congenital malformation commitee of the International Federation of Societies For surgery of Hand (IFSSH) in 1983[2,3]. A case of Tetra Amelia Syndrome in a stillborn male baby is reported here

2.Case report

A stillborn male baby was sent from the department of Obstetrics &Gynecology which showed complete absence of all four limbs (Fig1). It was associated with cleft lip and palate on the right side (Fig2). On further dissection, no deformity was found in the other systems. In this case the mother revealed a history of taking some local herbs for abortion in her first trimester.

Figure 1. Tetra amelia

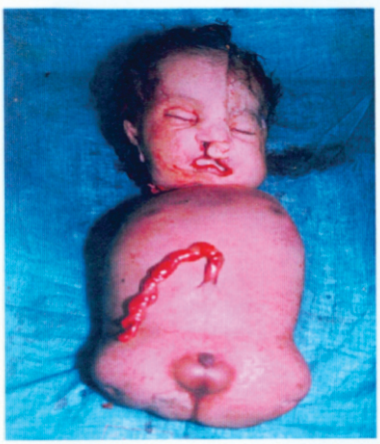
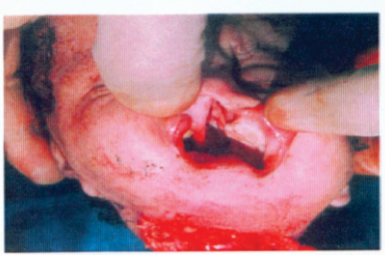


Figure 2. Cleft lip and palate



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3.Discussion

The embryonic forelimb buds appears on the 26th day and hind limb bud appears on 28th day of gestation . By 36th day the upper limb has started to differentiate into its three segments(arm, forearm and hand). In the lower limb the same process occurs shortly afterwards. By the end of the 6th week embryo has acquired a recognizable human forms. The upper limb is fully formed by 12 weeks and lower limbs by 14 weeks. During this period the muscles & nerves also develop and by the 20th week, joint movement is possible [4,5].

Most of the malformations involving limb reductions are due to embryonic insults between 4th and 5th weeks of gestation(1). Maternal smoking has been found to be associated with increased incidence of congenital defects (6). Teratogen induced limb defects have also been documented [1,4]. Some are found to be genetically determined and these usually have an autosomal dominant pattern of inheritance(4). Children born of consangniuos marriage are also reported to have TAS [1,7].

4.Prevalence

This anomaly is extremely rare . Cases are stillborn, or die shortly after birth [8]. Incidence

The limb malformations appears in about 6/10,000 live births , with 3.4/ 10,000 affecting upper limb and 1.1/10000 the lower one [1] . The incidence of congenital deformity of upper limb is also reported to be 1:9400 live birth [7].

These defects are often associated with other birth defects involving the craniofacial , cardiac and GIT, genitourinary and respiratory system [1].

A case of unilateral phocomelia is reported by Kanthimathi. B et al [9].

The present case is a still born male baby which presented as TAS with associated cleft lip and palate on right side. The mother too revealed of taking local herbs as an abortificant.

Currently used classification of congenital anomalies of upper limb is based on that of Swanson .He classified this into six subgroups which describe a clinical picture and do not always relate accurately to the developmental process of the limb. The first group includes the failure of formation of parts of a limb that is caused by developmental arrest affecting the long bones [3]. Our case falls in this subgroup. Others, such as Ogino have proposed improvements to address some of the uncertainties. But these classifications are of little practical value in terms of management of the patients[9].

Amelia can be diagnosed clinically and prenatally by ultrasonography. It has WNT3 gene association [1]

Cases of TAS are usually stillborn or die shortly after birth. Management of the surviving patients will depend on the presence and severity of the associated malformations. This may require the support of several medical disciplines. Cases of phocomelia or ectromelia, either unilateral or bilateral, treatment should be started in childhood as various prosthetic fittings are available(10).

5.Conclusion

Proper health education, antenatal screening of patients and genetic counseling of both parents can further reduce the risk of such congenital anomalies. This case is reported due to its rare occurrence.

Acknowledgements

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