

Tetra-amelia syndrome: a case report

Síndrome de tetraamelia: informe de un caso

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ABSTRACT

A still born male baby with tetra-amelia syndrome is reported, with craniofacial abnormalities and pulmonary agenesis. It presented complete absence of all four limbs in association with cleft lip and palate on the right side. The mother medical history revealed no remarkable details. The fetus died shortly after its birth. The embryonic correlation of the case is discussed. Proper health education, antenatal screening and genetic counseling can reduce the risk of such congenital anomalies.

Key words: tetra-amelia syndrome; still born; genetic disorders; malformations.

RESUMEN

Se informa el caso clínico de un mortinato del sexo masculino con el síndrome de tetraamelia, que mostró ausencia de las cuatro extremidades, unido a anomalías craneofaciales y agenesia pulmonar. Presentó ausencia total de las 4 extremidades unido a labio leporino, paladar hendido del lado derecho. Los antecedentes maternos no revelaron detalles considerable. El feto falleció poco después de su nacimiento. Se discute la correlación embriónica del caso. La educación para la salud adecuada, la pesquisa prenatal y el consejo genético pueden reducir el riesgo de estas anomalías congénitas.

Palabras clave: síndrome de tetraamelia; mortinato; trastornos genéticos; malformaciones.

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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.⁽¹⁾

Tetra-amelia syndrome is a very rare disorder characterized by the absence of all four limbs. ("Tetra" is the Greek word for "four" and "amelia" refers to the failure to develop before birth of an arm or leg). This syndrome can also cause severe malformations of other parts of the body, including the face and head, heart, nervous system, skeleton, and genitalia. The lungs are underdeveloped in many cases, which makes breathing difficult or impossible.⁽²⁾

As children with tetra-amelia syndrome have such serious medical problems, most are stillborn or die shortly after birth. The condition has been associated with a mutation in the WNT3 gene in one family, and it appears to be inherited in an autosomal recessive manner.

Treatment for those that survive depends upon the presence and severity of the associated symptoms and may require the coordinated efforts of a team of specialists.⁽³⁾

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 committee of the International Federation of Societies for Surgery of Hand (IFSSH) in 1983.^(2,3)

The overall prevalence of congenital limb amputation is 7.9/10,000 live births. Most are due to primary intrauterine growth inhibition, or disruptions secondary to intrauterine destruction of normal embryonic tissues. The upper extremities are more commonly affected.⁽³⁾

The case of Tetra Amelia Syndrome in a stillborn male is reported here.

Case presentation

A stillborn male baby was sent from the Obstetrics & Gynecology Ward showing complete absence of all four limbs. It was associated with cleft lip and palate on the right side.

On further dissection, others abnormalities were found in other systems such as generative organ apparently male showing a small and pediculated mass with tubular appearance as a penis, bilateral pulmonary agenesis, cardiovascular malformation and craniofacial anomaly with completely open fontanel.

The mother medical history did not reveal remarkable details.

Discussion

The embryonic forelimb buds appears on the 26th day and hind limb bud appears on day 28 of gestation. By 36th day, the upper limb has started to differentiate into its three segments (arm, forearm and hand). In the lower limbs the same process occurs shortly afterwards. By the end of 6 weeks the embryo has acquired a recognizable human form. The upper limb is fully formed by 12 weeks and lower limbs by 14 weeks.

During this period the muscles and nerves also develop and by the 20th week, joint movement is possible.

Most of the malformations involving limb reductions are due to embryonic insults between 4 and 5 weeks of gestation.⁽¹⁾ Maternal smoking has been found to be associated with increased incidence of congenital defects.

Congenital limb deficiencies have many causes and often occur as a component of various congenital syndromes. The most common cause of congenital limb amputations are soft-tissue and/or vascular disruption defects, such as amniotic band-related limb deficiency, in which loose strands of amnion entangle or fuse with fetal tissue.⁽³⁾

Teratogen induced limb defects have also been documented. Some are found to be genetically determined and these usually have an autosomal dominant pattern of inheritance. Children born of consanguineous marriage are also reported to have TAS. This anomaly is extremely rare.^(1,4)

Cases are stillborn, or die shortly after birth. Management of the surviving patients will depend on the presence and severity of the associated malformations.⁽⁴⁾

The limb malformations appears in about 6/10,000 live births, with 3.4/ 10,000 affecting upper limbs and 1.1/10000 the lower limbs.^(5,6)

These defects are often associated with other birth defects involving the craniofacial, cardiac and GIT, genitourinary and respiratory system.⁽¹⁾ The present case is a still born male baby which presented as TAS with associated cleft lip and palate on right side, craniofacial, cardiac, genitourinary and respiratory system.

The first group includes the failure in the formation of a limb that is caused by developmental arrest which affects the long bones. Our case is included in this subgroup. Others, have proposed improvements to address some of the uncertainties. But these classifications are of little practical value in terms of management of the patients.^(7,8)

Amelia can be diagnosed clinically and prenatally by ultrasonography. It has a WNT3 gene association.

This may require the support of several medical disciplines. In cases of phocomelia or ectromelia, either unilateral or bilateral, treatment should be started in childhood as various prosthetic fittings are available.⁽⁹⁾

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.

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