TETRA PHOCOMELIA - A Case Report

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ABSTRACT

We are presenting a rare case of Tetra-Phocomelia diagnosed by first trimester scan performed at 12 weeks of pregnancy. The postmortem examination confirmed the findings. This is one of the rarest cases which have been reported so far in view of the early gestational age at the time of diagnosis.

DISCUSSION

- Development of limb buds starts from the 26th day of intrauterine life and by the end of the 14th week, they are fully formed. Any disturbance in this pattern can result in limb malformations.
- Tetra-Phocomelia syndrome is a congenital disorder associated with a near or complete absence of all 4 limbs.
- Phocomelia can occur as an isolated skeletal defect or can be associated with other visceral anomalies like Renal, craniofacial, cardiac and uterine anomalies.
- The most common cause of congenital limb amputations is vascular disruption defects, such as amniotic band.
- Other causes include: Teratogenic agents (e.g., thalidomide, vitamin A), Substance abuse like cocaine or alcohol, gestational diabetes, use of X-ray radiation, fetal chromosomal anomalies like trisomy 18 or genetic inheritance (autosomal recessive trait mutation linked to chromosome 8).
- When a genetic syndrome is suspected, evaluation should include a thorough assessment for other physical, chromosomal and genetic abnormalities by a clinical geneticist. Typically, x-rays are required to determine which bones are involved.
- Antenatal ultrasonography remains the hallmark for detecting all fetal anomalies including that of limbs.
- Cytogenetic testing can be performed in cases with suggestive clinical findings.
- Individuals carrying phocomelia syndrome will generally show symptoms of growth retardation prior to and after birth. The syndrome can also cause severe mental deficiencies in infants.
- While there is no cure for phocomelia, treatment options have advanced in the recent years.
- Prosthetics can be used in place of absent limbs, and physical, occupational, and speech therapy among the therapies used to promote the quality of life of the patient.
- Prosthetic devices are most valuable for lower-limb deficiencies and for completely or almost completely absent upper limbs. If any activity in an arm or hand exists, no matter how great the malformation, functioning capacity must be thoroughly assessed before prosthesis or surgical procedure is recommended. Various types of surgery are used to rectify deformations of the face.

INTRODUCTION

- Intratropic reduction defects of the limbs occur when a part of or the entire limb of a fetus fails to form. The upper extremities are more commonly affected.
- The minor limb anomalies are relatively common and can usually be corrected surgically but the major ones are more complex and are mostly caused by genetic or environmental factors.
- Tetra Phocomelia is a rare congenital appendicular musculoskeletal malformation with a subnormal length of the long bones.
- It is seen in about 0.62 per 100,000 live births.
- Most cases are due to primary intratropic growth inhibition or disruptions secondary to intratropic destruction of normal embryonic tissues.

CASE PRESENTATION

We are presenting an interesting case of a 32 years old patient with an unremarkable medical history, who attended to the Fetal Medicine Unit at Southend University Hospital at 12 weeks and 4 days into her first pregnancy, for the dating scan. A single live intratropic pregnancy identified. There was a marked increase of the measurement of the Nuchal Thickness of the fetus (NT 7.7), both fetal feet and hands were found to be closely attached to the body. There was a degree of micrognathia, with narrowing of the Chest. No other abnormalities were identified on the scan. The risk for T21 was 1 in 7.

In view of these findings, a discussion was given about the diagnosis of tetra phocomelia, prognosis and the expected outcome in terms of the limbs anomalies. Options of conservative management ortermination of pregnancy were given. The couple opted for ending up the pregnancy medically and agreed for a post-mortem examination which was performed later and confirmed a diagnosis of four limbs phocomelia, with four ray oligodactyly in the upper limbs.

Genetic study of the baby revealed a normal male karyotype with essentially normal genetic makeup. Of note is that, there was no history of drug intake, radiation exposure, maternal diabetes or family history of congenital anomalies to support the occurrence of tetra-phocomelia in this baby.

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