

A Rare Fetal Congenital Anomaly: Meromelia

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Abstract

Congenital anomalies can be defined as presence of structural or functional anomalies which are present at the time of birth ⁽¹⁾ and are important causes of perinatal mortality in developing countries like India .Preconception and periconception screening can help to decrease the frequency of certain congenital anomalies either by removal of risk factors or by the reinforcement of protective factors .With the advances in imaging and invasive technique, early diagnosis is also possible. A rare case of congenital fetal limb defect-Meromelia is reported which was not diagnosed in repeated antenatal fetal scans. Due to rarity of the condition, gynecologists may not encounter this type of anomaly during their training period, but they should be aware of such condition as they are important causes of perinatal mortality in developing countries like India.

Keywords: Congenital anomaly, Meromelia, Amelia

Introduction

Limb defects are described as meromelia (partial absence) or Amelia (complete absence) of one or more limbs. Phocomelia is a type of meromelia where rudimentary hands or feet's are attached to trunk by small, irregularly shaped bones. Etiological factors include genetic, teratogens (thalidomide use) vascular disruptions and ischemia, chemicals and radiation exposure. Children with limb anomalies have associated malformations like craniofacial, gastrointestinal, heart, kidney and nervous system ⁽²⁾

Case Report

A 25 year old primigravida reported to gynae emergency for the first time in evening with c/o amenorrhoea 9 months with pain abdomen and leaking per vaginum for around 12 hrs. She was a referred case from nearby district hospital, referred for Primi with term pregnancy with Breech presentation in labor; as they had no emergency caesarean facility in their hospital. After detailed history it was found that she was married for 6 years, and was taking some infertility treatment from a quack since 1 year. She had never consulted a doctor and had conceived on the treatment given by the local quack, records of which were not available. Post conception she had never visited a registered doctor and was continuing medicines given by the Quack. By dates (LMP) she was 38 weeks while by dating scan of 11weeks she was 37 weeks. On systemic and obstetric examination patient was well built with good nutritional status and average weight with no evidence of pallor, edema or other systemic illnesses.

On examination, fundal height corresponded to 34 weeks of pregnancy with Breech presentation. She had good uterine contractions with a normal FHR. On P/S examination leaking was observed, liquor was clear, with no bleeding. P/V examination revealed a dilated 4-5cm cervix, almost 50% effaced. Breech was felt at 0 stations, frank breech. Membranes were absent and pelvis appeared adequate. She was advised for a vaginal delivery as all parameters were favorable but patient and attendants refused and demanded a caesarean section as they were previously counselled about fetal risks of breech delivery at the referring hospital and also because of her previous infertility status. The patient was carrying no antenatal investigation, only 3 USG from a remote USG center. First USG was done at around 11wk, 2nd at 22wk and the last was done around 34 wk. All the scans were normal and no gross congenital anomaly was reported. The last scan which was done 3 wks before revealed a SLF of 33wk 6day having Breech presentation with a baby wt. of 2475gm. Amniotic fluid was adequate with placenta anterior in upper uterine segment.

In our hospital all antenatal investigations including virology were done. As she was in active stg. of labor, patient was shifted for emergency LSCS.

The emergency LSCS was uneventful except for the fetal outcome.

Baby details

The congenitally malformed baby had no external genitalia, so gender could not be identified. The baby wt. was 2.1kg with 6, 8 APGAR and immediate cry. The facial symmetry and features were normal consistent with a term baby.

The chest was normally formed but the upper limbs showed defective development. The arms apparently were normal but the forearms were folded at elbows with deformed fingers. The forearms could not be straightened at elbow. Lower limbs were malformed, no external genitalia was visualized only a transverse limb was attached to pelvic girdle. One end of the transverse limb showed a lemon sized soft protrusion with rugosity suggestive of a malformed or underdeveloped scrotum; the tip of which showed urine coming out. No anal opening visualized. A Ryle's tube was passed which showed a patent upper GI tract. A working diagnosis of Phocomelia Syndrome was made and after stabilizing, the baby was referred to a pediatric higher Centre as there was no anal sphincter, for further management (Figure 1, 2, 3).

After the anomalous child birth a detailed history from patients and relatives was sought as regards consanguineous marriage, radiation exposure, febrile illness, rash, drug intake or any possible teratogenic exposure, h/o any anomalous child in family. Other than the treatment given by the quack both preconceptionally and during pregnancy no significant history was elicited. No papers or drug or wrappers were available with patient. The ultrasound were done at a peripheral USG Centre, the USG films were not available only their reports. Post LSCS the patients stay was uneventful and she was discharged (DOPR) on D4 and was advised for stitch removal. The baby as per the attendant was shifted to higher pediatric Centre where they explained a very poor prognosis. The patient did not turn up for stitch removal and telephonically she informed about the death of child on Day 6.

Figure 1



Figure 2



Figure 3**Discussion**

Meromelia is a birth defect characterized by the lacking of a part, but not all, of one or more limbs with the presence of a hand or foot. It results in a shrunken and deformed extremity. Such defects are mainly the results of genetic disorders, but some teratogenic or environmental factors have been identified, such as use of thalidomide from 1957 to 1962 for morning sickness (NVP).

Amelia is the birth defect of lacking one or more limbs. It can also result in a shrunken or deformed limb. Amelia may be present as an isolated defect, but it is often associated with major malformations in other organ systems. These frequently include cleft lip and/or palate, body wall defects, malformed head, and defects of the neural tube, kidneys and diaphragm. Facial clefts, facial anomalies, may be accompanied. The diaphragm may be herniated or absent and one or both kidneys may be small or absent. The complete absence of an arm or leg in Amelia occurs as a result of limb formation process being either prevented or interrupted early in the developing embryo. ^(3, 4)

Roberts - SC Phocomelia syndrome is autosomal recessive genetic disorder caused by homozygous or compound heterozygous mutations in ESCO2 gene on chromosome 8. It is one of the types of Cohesinopathies ⁽⁵⁾. Genetic mapping studies reveal that mutations in cohesion pathways are responsible for multispectrum development abnormalities termed Cohesinopathies. Phocomelia describes defective development of the arms and or legs so that the hands and feet are attached close to the body. Robert's syndrome and SC Phocomelia are considered a single genetic entity, with a wide phenotypic variation comprising craniofacial anomalies, tetra phocomelia and pre and post-natal growth restriction, there is a high neonatal loss rate and if the infant survives there is invariably mental retardation. ^(6, 7)

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