Extended Abstract Journal of Congenital Disorder 2020 Vol.2 Issue.1

A Case Study on Tetraphocomelia

Sessions W

Department of Pediatrics, Texas Tech ,Health Sciences Center, School of Medicine,Amarillo, Texas, USA

Abstract:

A newborn female presented with gross tetraphocomelia lacking features of an associated syndrome or relevant family or birth history that would cause the patient to be born with this anomaly. Tetraphocomelia is characterized by severe symmetrical limb reduction in utero. Several syndromes are associated with this finding: Robert's syndrome, Thrombocytopenia with Absent Radius syndrome, Grebe Syndrome, Waardenber syndrome, and Holt-Oram syndrome. Furthermore, certain in utero exposures, such as thalidomide, alcohol and cocaine, are also associated with similar musculoskeletal deformations. Due to the lack of associated symptoms or significant history, our case did not fit into any specific syndrome and appears to be the result of a sporadic, non-hereditary limb deficiency involving all four limb buds. This unique presentation of a rare congenital anomaly was possibly caused by amniotic bands or a vascular accident in utero, resulting in an isolated occurrence during the critical period of limb development, between fourth and the eighth week of gestation.

Phocomelia is a rare congenital malformation in which infants present with flipper like upper and/or lower limbs. Although various etiological factors are contributed, the main etiological factors are thalidomide drug use and genetic inheritance. Genetic inheritance is due to the malformation linked to chromosome 8. Here a case of Tetraphocomelia in a 20 weeks still born male fetus is presented and a review of relevant aspects of Phocomelia is also discussed.

We report a case of term live baby with tetra-phocomelia born to a 35-year-old G3P2L2A0 with history of consanguineous marriage. She was an unbooked case from a tribal community with no previous antenatal visits. At 39 wk of gestation, she was admitted to our hospital with complaint of pain abdomen and on examination was found to be in second stage of labour.

She delivered vaginally a term live 2.5 kg female baby with multiple anomalies. 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. The neonate also had multiple facial abnormalities like hypertelorism, microretrognathia and partial cleft palate. Further investigations revealed no abnormalities of internal organs. At present the baby is being followed up at our paediatric department. The case is reported owing to its rarity and term live birth.

Authors present a very rare case of tetra-phocomelia evaluated by antenatal ultrasonography. It is a condition seen in 0.62 per 100,000 live births. This is a congenital chromosomal abnormality involving the musculoskeletal system. Primi gravida with spontaneous conception after a long period of infertility underwent early anomaly scan. Patient was not aware of the last menstrual period hence; NT scan was missed. Routine early anomaly scan done between 16-18 weeks of pregnancy diagnosed a fetus with Tetra-Phocomelia. Due to the lack of associated symptoms or significant history, our case did not fit into any specific syndrome and appears to be the result of a sporadic, non-hereditary limb deficiency involving all four limb buds. Second opinion obtained from a fetal medicine consultant who confirmed the diagnosis.

Hence, decided for mid trimester termination and fetus was expelled after 8 hours. Fetus was not sent for pathological analysis. Tetra-phocomelia is a rare congenital anomaly and it may be associated with other deformity also. 1st case of phocomelia was described after the intake of thalidomide. In this condition hands and feet are seen as small flippers of a seal. The differential diagnosis includes sporadic phocomelia, Holt-Oram syndrome, thrombocytopenia-absent radius syndrome (TAR syndrome), Robert's syndrome, and thalidomide-induced phocomelia. Here authors are presenting a rare case of Phocomelia where there is no history of drug intake or family history. This has to differentiate from thrombocytopenia-absent radius syndrome (TAR syndrome), sporadic phocomelia, Holt-Oram syndrome, Robert's syndrome, and thalidomide-induced phocomelia.

Corresponding Author:

Sessions W

Department of Pediatrics, Texas Tech Health Sciences Center, School of Medicine
Amarillo, Texas, USA
Email: William.S.Sessions@ttuhsc.edu

References

- Shonubi AM (2006) Asymmetrical Tetraphocomelia with Radiohumeral Synostosis. Annals of Saudi Medicine 26: 318-320.
- https://emedicine.medscape.com/article/159911-overview.
- Bermejo SE, Cuevas L, Amar E, Bianca S, Bianchi F, et al. (2011) Phocomelia: A Worldwide Descriptive Epidemiologic Study in a Large Series of Cases From the International Clearinghouse for Birth Defects Surveillance and Research, and Overview of the Literature. American Journal of Medical Genetics 4: 305-320.