

A case of phocomelia in young primi gravida

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Abstract

Introduction: Phocomelia is a type of meromelia, in which there is partial agenesis of limb buds. Tetraphocomelia is a severe combination of limb defects in which total or partial agenesis of upper and lower limbs is seen, leading to the proximity of limbs to the trunk of the fetus resembling the flipper of a seal “an aquatic animal”.¹

Keyword: Tetraphocomelia, Thalidomide syndrome, phocomelia, Limbs defects, Teratogenic agents, Hydroamniosis, Genetic inheritance.

Introduction

Phocomelia is a condition that involves malformations of the arms and legs. Although many factors can cause phocomelia, the prominent roots come from the use of the drug thalidomide and from genetic inheritance. Occurrence in an individual results in various abnormalities to the face, limbs, ears, nose, vessels and many other under developments of organs. Although operations may improve some abnormalities, many are not surgically treatable due to the lack of nerves and other related structures. Phocomelia is an extremely rare congenital disorder involving malformation of the limbs (dysmelia); Étienne Geoffroy Saint-Hilaire coined the term in 1836.¹ There is no specific treatment for phocomelia. However, if it is part of a genetic syndrome, surgical intervention may be recommended for associated malformations.^{2,3}

Case Report

25 years old woman came to hospital for USG. She was gravida 1 para 0. She was not sure about her last menstrual period. Her active married life was three years. (non consanguineous marriage). She had taken some irregular treatment to conceive pregnancy, but

records were not available.

Past history: She had no past history of major medical disorders or Drug intake.

Personal History: She has habit of chewing 2 to 3 packets of tobacco per day since last few years. Her husband had habit of smoking 5 to 10 Bidies per day since childhood and chewing 5 to 7 packets of tobacco per day.

Obstetric History: Her uterus was 32 weeks of size with breech presentation amount of liquor was increased (Polyhydroamniosis). Foetal movements were seen and foetal heart sounds were normal.

Ultrasound Examination: USG examination shown Single live Intra uterine viable fetus with breech presentation. Exact maturity could not be derived due to discordance in various organ measurements in fetal biometry. Cisterna magna was dilated, with absence of spine below lumbo-sacral region. Cranio-facial anomalies seen with comparatively large face, small and cut external ear. Fetal ascites was present. Fetal heart rate was normal and fetal echo shows presence of VSD. All peripheral limb bones were very short. Polyhydroamniosis seen AFI-33.09.



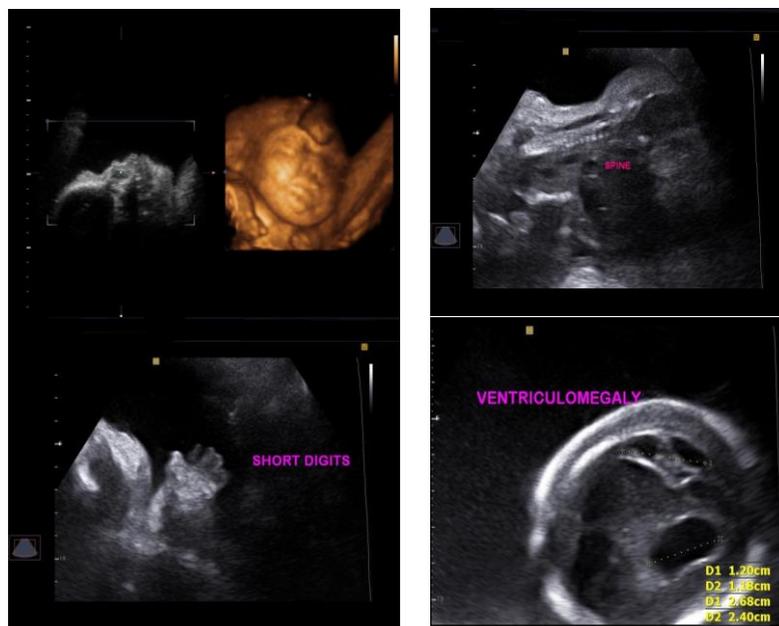


Fig. 1



Fig. 2

Management

Couple and relatives were explained about major fetal anomalies. Induction of labour was planned with informed and written consent. Under USG guidance around 2 liter of liquor drained slowly Trans abdominally. Intravenous antibiotic and other supportive treatment given. Basic investigation done. Labour was induced by dinoprostgel and augmented by oxytocin drip.

Discussion

First case of Phocomelia was described in Germany in 1956. Father was pharmacist and mother took medicine thalidomide for nausea during her pregnancy. After deliver baby had no arms and vestigial flipper like hand.⁴

Thalidomide was first marketed in 1956 in West Germany. Primarily prescribed as a sedative or hypnotic, thalidomide also claimed to cure "anxiety, insomnia, gastritis, and tension".⁵ Afterwards,

it was used against nausea and to alleviate morning sickness in pregnant women. Shortly after the drug was sold in West Germany, between 5,000 and 7,000 infants were born with phocomelia (malformation of the limbs). Only 40% of these children survived.⁶

The symptoms of phocomelia syndrome are undeveloped limbs and absent pelvic bones. However, various abnormalities can occur to the limbs and bones.⁷ Usually the upper limbs are not fully formed and sections of the "hands and arms may be missing." Short arm bones, fused fingers, and missing thumbs will often occur. Legs and feet are also affected similarly to the arms and hands. Individuals with phocomelia will often lack thigh bones, and the hands or feet may be abnormally small or appear as stumps due to their close "attachment to the body."⁸ Individuals carrying phocomelia syndrome will generally show symptoms of growth retardation previous to and after birth. It can also cause severe mental deficiencies in infants. Severe symptoms of phocomelia are encephalocele and hydrocephalus; causing vomiting and migraine. An abnormally shaped uterus (bicornuate), clotting disorder, malformations in the kidney and heart, Shortened neck, and urethral abnormalities.

When an individual is born with phocomelia due to drugs or pharmaceuticals, it is known as thalidomide syndrome. The symptoms of thalidomide syndrome are defined by absent or shortened limbs; causing flipper hands and feet. According to Anthony J Perri III, and Sylvia Hsu they can additionally receive: Palsy disorder of the face, Ingrown genitalia, disorders to the limbs, Ear and eye abnormalities; resulting in limited/complete loss of hearing or sight, Gastrointestinal and genitourinary tract

disorders, distorted digestive tract, heart, kidney, Undeveloped/missing lungs. The infants that were exposed to thalidomide during development phases had a 40% chance of survival.⁹ The McMredie-McBride hypothesis explains that the limbs of the infants become malformed as a result of the thalidomide harming the neural tissue—simply because the neural tissue has such a large impact on formation and development of the limbs.¹⁰

Genetic inheritance

According to National Organization for Rare Disorders (NORD): when phocomelia is transmitted [in its familial genetic form] it is seen as an autosomal recessive trait and the mutation is linked to chromosome 8.⁸ A study of Roberts Syndrome, a genetic disorder showing similar symptoms to phocomelia, has shed light on the possible causes.¹¹ An individual afflicted with Roberts Syndrome will have chromosome copies that do not connect at the centromeres, making them unable to line up accordingly. As a result, the newly made cells contain an excess or reduced number of chromosomes. In both Roberts Syndrome and phocomelia the cells cease to develop, or die, preventing proper development of the limbs, eyes, brain, palate, or other structures.

Treatment

Project Orphan Anesthesia is a project whose aim is to create peer-reviewed, readily accessible guidelines for patients with rare diseases and for the anesthesiologists caring for them. The project is a collaborative effort of the German Society of Anesthesiology and Intensive Care, Orphanet, the European Society of Pediatric Anesthesia, anesthesiologists and rare disease experts with the aim to contribute to patient safety.¹²

Prosthesis is a synthetic alternative for missing limbs, teeth, and various other body parts. Advances in prosthetic limbs have increased greatly during the twentieth century. The use of new materials such as modern plastics, complex procedures and better pigments have created lighter in weight and more realistic looking artificial limbs. With the advancement of myoelectric prosthetic limbs, patients are able to move their limbs without the use of cords or other devices. The myoelectric limbs can detect electric signals from the nervous system and muscles. They were first used on adults, but now they are being fitted to children.¹³ Patients that receive a loss of limbs due to phocomelia are typically treated with prosthetics. Infants at the age of 6 months are recommended to have a prosthetic mitten fitted; enabling them to get used to the prosthesis. A hook will be added when the child reaches the age of 2 years. Eventually the patient may receive a myoelectric prosthetic limb. Patients are treated in this way due to the lack of understanding at a young age and the absence of necessary tissues and

bones to hold the prosthetic limb.

Conclusion

Most of the anomalies can be diagnosed antenatally if we do the anomalies scan around 18 to 20 weeks of gestation. So we should advice patient to come for regular antenatal visits and anomalies scan around 18 to 20 weeks of gestation. Once there is one anomaly try to search for another one. We should give folic acid 5 mg daily pre conceptionally to prevent neural tube defect.¹⁴

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