

# Phocomelia: An Extremely Rare Congenital Disorder involving the Limbs (Dysmelia)

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## ABSTRACT

Phocomelia—phoco(seal), melia(limbs)—is a rare congenital deformity in which hands or feet are attached close to the trunk, the limbs being grossly underdeveloped or absent. The babies are born with limbs that look like flippers on a seal. The two main etiological factors of phocomelia are genetic inheritance and use of the drug thalidomide. Here, we present a case of phocomelia in second gravida without exposure to thalidomide.

**Keywords:** Phocomelia, Thalidomide, Congenital abnormality.

## INTRODUCTION

Phocomelia is a devastating, rare congenital limb malformation in which the long bones are shorter than normal, with the upper portion of the limb being most severely affected. In extreme cases, the hands or fingers are attached directly to the shoulder and the most proximal elements (those closest to the shoulder) are entirely missing. This disorder, previously known in both autosomal recessive and sporadic forms, showed a marked increase in incidence in the early 1960s due to the tragic toxicological effects of the drug thalidomide, which had been prescribed as a mild sedative.<sup>1</sup>

Symptoms of phocomelia syndrome are underdeveloped limbs and absent pelvic bones.<sup>2,3</sup> Usually, the upper limbs are not fully formed and sections of the hands and arms may be missing.<sup>2,4</sup> Short-arm bones, fused fingers and missing thumbs will often occur. Legs and feet are also affected similarly to that of the arms in hands.<sup>2,4</sup> According to the National Organization for Rare Disorders (NORD), individuals carrying phocomelia syndrome will generally show symptoms of growth retardation previous to and after birth. The syndrome can also cause mental deficiencies in infants. Infants born with phocomelia will normally have a petite head with 'sparse hair' that may appear 'silveryblonde'.<sup>2,4</sup> The pigments of eyes will be bluish. Phocomelia can also cause an undeveloped nose with

slender nostrils, disfigured ears, irregularly petite jaws (also known as micrognathia) and a cleft lip with cleft palate.<sup>2,4</sup>

## CASE REPORT

A 22-year-old, G2P1L2 (twins) lady was admitted to labor room. The chief complaints of decreased fetal movements and leaking per vaginum of 18 hours duration.

She has been married since 7 years, nonconsanguineous marriage. She had first preterm twin delivery at 32 weeks, both females, birth weight 1.25 Kg. Now 5-year-old, alive and well. She was an unbooked case with only one antenatal visit in PHC at 5 months, advised to do USG and other antenatal investigation, but did not get it done, only HIV test was done. No follow-up visits. Patient was taking iron and calcium tablets. No significant past history. No history of thalidomide intake, no exposure to fertilizer like endosulphan, no history of X-ray irradiation. No history of use or exposure of pesticides. No history of congenital anomalies in the family. She is a house wife, husband is a driver, not living close to any farm. No history of diabetes in mother. We could not find any cause (The drug was withdrawn from the market in 1961. In 1998, FDA approved use of thalidomide for treatment of leprosy. We asked history of leprosy in the family to rule out, if the mother has taken the drug by mistake. There is no such history).

On admission, she was afebrile with pulse rate 88/min and blood pressure of 110/70 mm Hg. Uterus was corresponding to 32 weeks size, with breech presentation. She was getting two contractions every 10 minutes. Cervix was mid position, soft, 2 cm dilated, 30% effaced. There was footling presentation with

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cord prolapse and thick meconium. She was placed in a left lateral position and nasal oxygen started. Parenteral fluids were started. In view of footling presentation and cord prolapse with cord pulsation, decision was taken for emergency cesarean section.

As the patient was taken for emergency cesarean section, Ultrasonography was not done.

As it was cord prolapse with cord pulsation, we took patient for emergency LSCS.

*Intraoperative findings:* Female child of 1.2 kg, extracted by footling, baby did not cry after birth. All emergency resuscitation methods were tried. Baby could not be revived and was declared dead after 1 hour. Umbilical cord had two arteries and one vein.

*Baby anomalies:* Underdeveloped limbs, fused fingers and toes, dextrocardia, lung hypoplasia on left side, cleft palate, disfigured ears, sparse hair, polydactyly (Figs 1 to 3).

Chromosomal study of the baby advised but patient refused. We asked for autopsy of the baby, patient's husband and relatives refused.

## DISCUSSION

The first case of phocomelia was described in Germany in 1956. Father of the child was working as a pharmacist, mother had received thalidomide for nausea of pregnancy. After delivery baby was found to have had no arms and only vestigial flipper-like hands.<sup>5</sup>

Thalidomide was released into market in 1957 in West Germany. Initially, it was used as a sedative or hypnotic, thalidomide also claimed to cure anxiety, insomnia, gastritis and tension.<sup>2,6</sup> Later it was used to combat against nausea and alleviate morning sickness in pregnant women. Thalidomide became an over the counter drug in Germany around 1960, and could be purchased without a prescription. Shortly after the drug's selling, in Germany, between 5,000 and 7,000 infants



Fig. 2: Short lower limb with polydactyly and fused toes

were born with the qualities of phocomelia.<sup>2,7</sup> Out of these children merely 40% of them survived. Research also proves that although phocomelia was nonexistent through the 40s and 50s, by time the drug was released in Germany in the 60s, cases of severe phocomelia amplified; the direct cause was linked to thalidomide.<sup>2,7</sup> The statistic was given that '50% of the mothers with deformed children had taken thalidomide during the first trimester of pregnancy'.<sup>2,7,8</sup> Throughout Europe, Australia, and the United States, 10,000 cases were reported of infants with phocomelia; only 50% of the 10,000 survived.<sup>2,7-9</sup> Thalidomide became effectively linked to death or severe disabilities among babies. Those subjected to thalidomide while in the womb experienced limb deficiencies in a way that the long limbs either were not developed or presented themselves as stumps. Other effects included: Deformed eyes, hearts, alimentary, and urinary tracts, along with blindness and deafness.<sup>2,7,8,10</sup>



Fig. 1: Underdeveloped upper limbs with fused fingers



Fig. 3: X-ray showing short limb bones

## GENETIC INHERITANCE

According to 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.<sup>2,4</sup>

There are several attachment points in a chromosome, the centromere is the major connection point and where the immensity of the work generates. An individual containing phocomelia will have chromosome copies that do not connect at the centromeres, making them unable to line up accordingly. As a result, the cell becomes incapable of division or slow in the process; because of this the newly made cells contain an excessive or reduced amount of chromosomes.<sup>2,11</sup> In phocomelia the cells cease to develop, or die, preventing proper development of the limbs, eyes, brain, palate, or other structures.<sup>2,11</sup>

## CONCLUSION

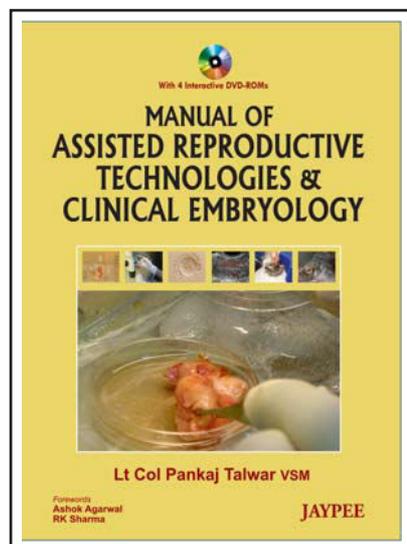
Most of the anomalies can be diagnosed antenatally if we do the anomaly scan at around 18 to 20 weeks of gestation. Hence, we should ask all the pregnant mothers to have at least one USG at 18 to 20 weeks to rule out any anomalies in the baby.

We have asked the patient and relatives to take folic acid preconceptionally, regular antenatal visit is must. We have advised triple test or amniocentesis in next pregnancy, routine diabetes screening to rule out other congenital anomalies. Since phocomelia is diagnosed with USG, we can do anomaly scan at early second trimester.

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# Book Review



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## Manual of Assisted Reproductive Technologies and Clinical Embryology

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