Case Report

Sirenomelia

Dr. Mahesh Shinde[1], Dr. Abhijit Shinde[2], Dr. Sushrut Kumar[2], Prof. Dr. Sunil Natha Mhaske[3], Dr. Suresh Waydande[4]

[1]Junior Resident, [2]Assistant professor, [4]Professor & Head. Department of paediatrics, DVVPF'S Medical College & Hospital, Ahmednagar-414111, Maharashtra, India.

[3]Professor, Department of Pediatrics & Dean, DVVPF's Medical College & Hospital, Ahmednagar-414111, Maharashtra, India

Abstract:

Sirenomelia is a rare and fatal congenital defect characterized by varying degrees of lower limb fusion, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary, and anorectal atresia. We report a case of baby, born with narrow chest, bilateral hypoplastic thumb, fused lower limbs with a single foot and 5 toes, absent external genitalia, imperforate anus and umbilical cord with single umbilical artery. When diagnosed antenatally, termination should be offered.

Key words: Sirenomelia; Caudal Regression Syndrome; Mermaid Syndrome; Potter's Facies

Introduction:

Sirenomelia is a rare and fatal congenital defect characterized by varying degrees of lower limb fusion, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary, and anorectal atresia. The incidence of sirenomelia is 0.8-1 case/100,000 births with male to female ratio being 3:1. There is a strong association with maternal diabetes where relative risk is 1:200-250 and up to 22% of fetuses with this anomaly will have mothers with diabetes.[1]

Anomalies observed in the sirenomelia described as most severe form of caudal regression syndrome, fusion of both lower limbs both lower extremities, presence of single umbilical artery and persistent vitelline artery are major features of serinomalia. Although the primary melecular defect result in serinomelia remains unclear, two main pathogenic hypothesis namely the vascular steal hypothesis and the defective blastogenesis hypothesis are proposed.[2]

The most common feature seen in serinomelia is complete or partial fusion pf both lower limbs into single lower limb, giving it a mermaid resemblance. there are usually many underlying visceral abnormalities that make it incompatible with life with a few rare exceptions of infants surviving with this condition.[3]

Case Report:

A 34-year-old unbooked G3P2L2 at 35 weeks 5 days of gestational age with previous two live vaginal birth was admitted in the labor room with pain in the abdomen. She had no history of prior antenatal care and belonged to a tribal community with lower socioeconomic status. She was otherwise healthy with no known history of genetic or congenital anomaly in her family. On examination, she was observed to be in the second stage of labor with cephalic presentation and regular fetal heart rate. She at a pre-term 2.3 kg baby via normal vaginal delivery with history of birth asphyxia. Baby not cried after birth, no activity and hypotonia present. Immediately intubated with ET NO 3 and bag and tube ventilation given and shifted to Neonatal Intensive Care Unit and put on Controlled Mode of ventilation. Baby has multiple congenital anomalies. The Apgar score was 3 at 1' and 4 at 5 min, 5 at 10 min. On physical examination, the infant showed narrow chest, bilateral hypoplastic thumb, fused lower limbs with a single foot and 5 toes (Figure No. 1), absent external genitalia, imperforate anus and umbilical cord with single umbilical artery There were also prominent epicanthal folds, hypertelorism, downward curved nose, receding chin, low-set soft dysplastic ears and small slit-like mouth suggestive of Potter's facies.

Corresponding Author: *Dr. Mahesh Shinde*

Email ID: *mdshindemd968962@gmail.com*

Address: *Department of paediatrics, DVVPF'S Medical College & Hospital, Ahmednagar-414111, Maharashtra, India*

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X ray Chest suggestive of respiratory distress syndrome (Grade III) with curved spine (Figure No. 2) and X Ray lower limb suggestive of two femur and two tibia with absent fibula. (Figure No. 3) Baby was died on second day of life due to multiple congenital anomalies not compatible with life.

Figure No.1: Clinical photograph of Sirenomelia

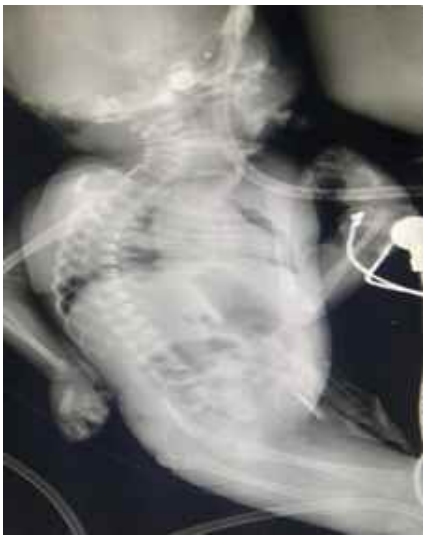

Figure No.: 2 X-ray chest

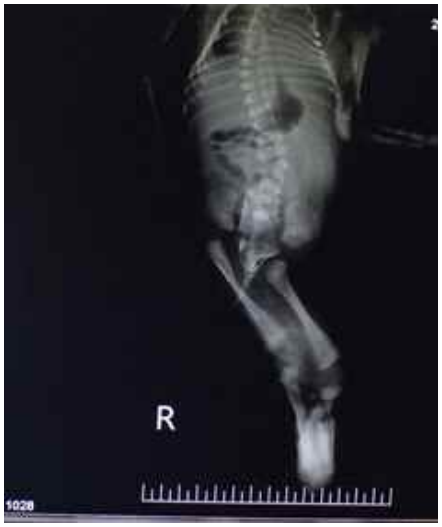

Figure No.: 3 X-ray Lower Limb

Discussion

Anomalies observed in sirenomelia are described as the most severe form of caudal regression syndrome. Fusion of the lower extremities, presence of single umbilical and persistent vitelline artery are major features of sirenomelia.[4,5] Although the primary molecular defect resulting in sirenomelia remains unclear, two main pathogenic hypotheses namely the vascular steal hypothesis and the defective blastogenesis hypothesis are proposed. According to vascular steal hypothesis, fusion of the limbs results from a deficient blood flow and nutrient supply to the caudal mesoderm, which in turn results in genesis of midline structures and subsequent abnormal approximation of both lower limb fields. However, in defective blastogenesis hypothesis, the primary defect in development of caudal mesoderm is attributed to a teratogenic event during the gastrulation stage.[6] Such defect interferes with the formation of notochord, resulting in abnormal development of caudal structures, Maternal diabetes, tobacco use, retinoic acid and heavy metal exposure are possible environmental factors. In our first case, there was history of tobacco use before and during pregnancy, while in the second case the mother had overt diabetes.[7] Sirenomelia is usually fatal within a day or two of birth because of complications associated with abnormal kidney and urinary bladder development and function.

In literature approximately 300 cases are reported worldwide of which 14 are from India. In most of the cases the diagnosis was performed after birth. In antenatal period, sirenomelia can be diagnosed as early as 13 weeks by using high resolution or color Doppler sonography. The condition is usually incompatible with life due visceral abnormalities especially that of renal system. Exceptional cases without renal agenesis have survived, the best example being Tiffany Yorks, a 13-year-old girl who was born with fused legs. Over the years, she has undergone numerous operations to separate her lower extremities.[8] The facial abnormality usually found in sirenomeliac infants known as Potter's facies, which includes large, low-set ears, prominent epicanthic fold, hypertelorism, flat nose and receding chin. In both of our cases, features of Potter's facies were present. When features of Potter's facies are combined with oligamnios and pulmonary hypoplasia it is known as Potter's syndrome,which was present in our second case. In our first case, the right thumb was hypoplastic, which was also previously reported. Stocker and Heifetz classified Sirenomeliac infants from Type 1 to Type VII according to the presence or absence of bones within the lower limb. Although we did not have radiographs to classify our case with certainty, nevertheless based on external examination, we suggest our first and second case belonged to Type IV (partially fused femurs and fused fibula) and Type I (all thigh and leg bones are present), respectively.[9]

Conclusion:

Sirenomelia is a rare and lethal congenital anomaly. When diagnosed antenatally, termination should be offered. However, prevention is possible and should be the goal. Regular antenatal checkup with optimum maternal blood glucose level in preconceptional period and in first trimester should be maintained to prevent this anomaly.

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