

CASE REPORT

Sirenomelia-The Rare Mermaid Syndrome In Neonate of A Diabetic Mother

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ABSTRACT

Introduction: Sirenomelia or the mermaid syndrome is a rare form of caudal regression syndrome characterised by abnormal development of caudal region of body. Prognosis is very poor because it is accompanied by variable major anomalies, including bilateral renal agenesis, sacral agenesis and imperforate anus.

Case Report: A 34 year old primigravida with history of uncontrolled diabetes mellitus for last 4 years was admitted in emergency of S.N Medical College,agra with postdated pregnancy. Emergency caesarean section was done due to contracted pelvic and with poor bishop score, a severely asphyxiated 2.8 kg baby with fusion of both lower limbs and absence of external genitalia, urethral and anal opening was delivered.

Conclusion: Infants of diabetic mothers have two to three times the average incidence of congenital anomalies including forms of caudal regression. Exact etiology of sirenomelia is unknown but in our case uncontrolled diabetes mellitus, inadequate antenatal care are the proposed causes.

Keywords: Sirenomelia, Vascular steal, Mermaid syndrome.

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INTRODUCTION

Sirenomelia or the mermaid syndrome¹ is a rare form of caudal regression syndrome characterised by abno-

mal development of caudal region of body with varying degree of fusion of lower limbs absent external genitalia, anal imperforation and renal agenesis or dysgenesis. Because of the resultant oligohydramnios, these infants most often have Potter's facies and pulmonary hypoplasia.² Prognosis is very poor because it is accompanied by variable major anomalies, including bilateral renal agenesis, sacral agenesis and imperforate anus. Only four cases of a surviving infant with sirenomelia have been reported.³

CASE REPORT

A 34 year old primigravida with history of uncontrolled diabetes mellitus for last 4 years was admitted in emergency of S.N Medical College, Agra with postdated pregnancy. There was no antenatal check up. On examination: B.P 110/70 mm of Hg, pulse - 84/min, respiratory rate -22/min. On per abdomen examination - fundal height-34 weeks, lie-longitudinal, presentation cephalic, fetal heart rate-130/min. Uterine contraction 1in 10min lasting for 10-15sec. Per vaginal examination: os closed, cervix uneffaced, show absent, pelvis was contracted. On investigations- Hb-11gm%, TLC- 4500 cells/cumm, DLC- P78L18M4, Random blood sugar 218 mg/dl, HbA1c 8.2(suggestive of poorly controlled diabetes). Urine routine- albumin traces, sugar 2+, puscell-2-4, epithelial cell 1-2 SGOT/PT-23/32, Serum Creatinine-1.1 (0.6-1.5). HIV -Non Reactive, HBsAg-Non Reactive.

Emergency caesarean section was done due to contracted pelvic and with poor bishop score, a severely asphyxiated 2.8 kg baby was delivered. Physical examination of baby (Figure-1a,b) showed fusion of both lower limbs and absence of external genitalia, urethral and anal opening. The upper half of the body was normal. Postnatal ultrasound showed absence of bladder, ureters and both kidneys. Family refused autopsy study. Cytogenetic analysis revealed normal female karyotype (XX).

DISCUSSION

Infants of diabetic mothers have two to three times the average incidence of congenital anomalies. These



Figure-1a,b: SIRENOMILEA, showing fused legs, giving them the appearance of a mermaid's tail.

include neural tube defects, cardiac defects (transposition of the great vessels, coarctation of the aorta, VSD, ASD, cardiomyopathy), situs inversus, renal anomalies (hydronephrosis, renal agenesis, multicystic dysplastic kidney, duplication of the renal tracts), intestinal atresia, and forms of caudal regression including sacral agenesis.^{4,5}

Lynch and Wright reported a case⁶ where the mother had diabetes and the infant had sirenomelia with renal agenesis and an absent right radius. There was also a hypertrophic cardiomyopathy and a bicuspid pulmonary valve. It bears resemblance to the mermaid Greek mythology. Affected individuals exhibit a variable range of defects, including hypoplasia and or fusion of lower limbs, vertebral abnormalities, renal agenesis, imperforate anus, and anomalies of the genital organs.⁷ The exact etiology of sirenomelia is not known. It is reported to occur with teratogenic agent like cadmium and lead. Maternal diabetes mellitus, genetic predisposition and vascular hypoperfusion have been proposed causes.

Stevenson et al.⁸ reported in 1986 that the aetiology of sirenomelia was explained by vascular steal theory. They documented the common feature of a single large abnormal artery arising from high in the abdominal cavity, which they thought diverted nutrients from the caudal end of the embryo in a vascular steal phenomenon. The associated anomalies are variable and involved multiple organ systems. The most common anomalies include a single umbilical artery, malformation of urinary tract, lower gastrointestinal tract and external genitalia⁹.

Sirenomelia sequence is classified into three groups according to the number of feet present. The most common of the three conditions is SYMELIA APUS, in which both legs are merged completely into a single lower extremity. In this condition both feet are absent or rudimentary. SYMELIA UNIPUS shows a presentation of one foot, two femora, tibiae and fibulae. In SYMELIA DIPUS, two distinct feet are

present but are malrotated and resemble fins. Our case was compatible with symelia apus.

CONCLUSION

In our case, mother was known diabetic with inadequate antenatal care and uncontrolled blood sugar levels belonging to low socioeconomic status.

Sells et al.,¹⁰ found that intellectual function in infants where mothers had controlled blood sugar level was not different from controls, whereas there was a correlation between delayed development or major congenital malformations in the infant and poor maternal diabetic control. Mermaid syndrome is a fairly rare condition coupled with slim chances of survival of baby making the study of the condition very difficult.

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