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CASE REPORT

A RARE CASE OF FETUS WITH SIRENOMELIA TO 34 WKS PREGNANT WOMAN WITH PREVIOUS LSCS

Kirti M. Hurakadi[1], Rutuja Kolekar[2]

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ABSTRACT: We report a rare case of a fetus with Sirenomelia to a pregnant woman from a rural, poor background, wherein early diagnosis of this lethal anomaly has been missed in this era of advanced obstetric care. Sirenomelia is a rare developmental anomaly of caudal region with varying degree of fusion of lower limbs, resembling Greek mythological character Mermaid. Antenatal diagnosis of this universally lethal condition is desirable, so that early termination of pregnancy can be offered to reduce the associated morbidity. A good understanding of this rare condition remains central to effective counseling and management of affected couples.

KEYWORDS: Prev LSCS.

INTRODUCTION: Sirenomelia is a rare and fatal congenital anomaly characterized by single fused lower limbs with multiple urogenital and anorectal malformations with an incidence of 0.8-1 case/ 100000 births, with male to female ratio being 3:1.(1) The sequence was originally described by Rocheus in 1542 and Palfyn in 1953 and named after the mythical Greek Sirens. Most of the cases of Sirenomelia results in still birth or die within in a day or two due to congenital complications.

CASE REPORT: An unbooked case of 24 yrs old woman, G2P1L1 with 8½ months of amenorrhea, came to our hospital at 2.30 am with labor pains. She was referred from a Primary Health Centre in view of previous LSCS in labor. History revealed that the patient had not taken regular Antenatal visits. She had only one obstetric scan at 7th month of gestation done at a rural place which showed a 26 weeks single live fetus with breech presentation with no other sonological abnormalities. Patient had labor pains since 4 hrs and leaking P/V since 1hr.

On examination, pt was clinically pale. She had tachycardia and was normotensive. On per abdominal examination, she had a pfannensteil scar, with no scar tenderness. Uterus was 34 wks size with good uterine contractions (3/10/30). The fundal grip suggested of head and lateral grip suggested of back on the left side. Pelvic grip was inconclusive. The fetal heart rate was 150bpm. On per vaginal examination, cervix was fully dilated. The membranes were absent and the liquor was clear. We felt a single limb like bony structure continuous with the fetal spine with no genitalia or anal opening (As expected in a breech presentation). The feet were also not felt. An emergency obstetric scan was done. It revealed a single live intrauterine fetus of 34 -36 weeks of gestation (with respect to BPD & HL) with good cardiac activity and oligohydraminos. The lower limbs and urinary bladder could not be visualized.

After taking consent for VBAC (Vaginal birth after cesarean section), complete breech extraction was done. We extracted a fresh stillborn fetus weighing 2.2kg with potter's faces, normal appearing upper limbs and trunk. The lower limbs were fused in the midline at the thighs and the legs. Feet were absent. Also, baby had imperforate anus, absent external genitalia, absent urinary orifice.

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The postmortem radiograph (X-Ray) showed sacral agenesis, fused hypoplasticpelvis, single appearing femur shaft bone with two distal epiphyseal ossification centers, single small tibia. The Fibula and tarsal bones were absent.

We failed in convincing the relatives to allow the baby to be taken for further evaluations due to their blind beliefs, social stigma and old rituals.

DISCUSSION: Sirenomelia also known as mermaid syndrome is a rare syndrome (0.8 to 1/100000 births).[2] The etiology of Sirenomelia is unclear though it is well known that the embryological injury occurs between 28 and 32 days of life and the site is at the caudal mesoderm. Stevenson et al.[3]

Explains diversion of blood away from the caudal region of the embryo through the abdominal umbilical artery, i.e., "vascular steal", as the primary mechanism leading to Sirenomelia. Other hypothesis being altered oxidative metabolism from maternal diabetes that may cause free oxygen radicals in the developing embryo which may be teratogenic.[4]

Sirenomelia was once considered as a part of caudal regression syndrome, but evidences have shown that Sirenomelia and caudal regression are two different entities.[5] Some authors such as Jones et al[6]have adopted a new classification which distinguishes the following:

- **Caudal regression syndrome (CRS) (also called Caudal Dysplasia Sequence):** The principal elements of this are defects in the development of the sacrum, up to agenesis, and alterations of the spinal cord with, as a corollary, more or less severe urinary incontinence and misplacement of the lower limbs. Renal agenesis and anal imperforation are inconstant characteristics. CRS is compatible with life, but at the price of heavy motor and sphincterian sequelae.
- **The Sirenomelia Sequence.:** This is associated with renal agenesis, an absence of both external and internal genital organs (With differentiated gonads: the sex ratio is 3 boys: 1 girl), anal imperforation with absence of a rectum, digenesis of the sacrum to agenesis, sometimes associated with lumbar vertebral digenesis and a variable degree of atrophy and an inconstant fusion of the lower limbs. The inconstant fusion of the lower limbs and their misplacement have two possible explanations: an absence of the cleavage of the common bud of the lower limbs by allantois, or an absence of the caudal extremity, which prevents the normal rotation of the lower limbs, thus leading to their fusion on the mid line.

Twin pregnancies and Sirenomelia sequence: 9% to 15% of the cases of Sirenomelia are associated with twin monozygotic pregnancies. The relative risk is multiplied 100-fold in the case of twin pregnancies.[7]

PROGNOSIS: Sirenomelia is fatal in most cases because of the characteristic pulmonary hypoplasia and renal agenesis. About 50% of the children are born alive after eight or nine months of pregnancy. Death occurs within the five days following birth.[8] Post-natal management requires the presence of kidneys**,** even if they are dysgenetic. Murphy et al[9]reported one case where a child born with Sirenomelia survived**;** the infant was neurologically normal and had fused lower extremities**,** an imperforate anus, colon atresia, bilateral fused pelvic kidneys with renal dysplasia and sacral dysplasia**,** and genital abnormalities. Laparotomy and colostomy were performed**,** and an eventual separation of the lower extremities was planned. Clarke et al[10] report on a three-month-old infant whose Sirenomelia was diagnosed prenatally. The infant was neurologically normal and has fusion of the lower limbs with associated renal dysplasia**,** an imperforate anus**,** pelvic and sacral dysplasia**,** and genital abnormalities. The infant's anomalies were compatible with life and surgical separation of the

lower limbs was done. Managing Sirenomelia is difficult and quite costly, requiring several interventions, the results of which are unpredictable.

Antenatal Diagnosis: Since the prognosis is bad, the management of Sirenomelia is difficult, with unpredictable results. It seems more than justified to put the emphasis on antenatal diagnosis in order to choose the cases with better prognosis. Antenatal diagnosis is based on a morphologic ultrasound study (Oligohydraminos, bilateral renal agenesis, a single lower limb, a unique umbilical artery, absence of a bladder, undetermined external genitalia, anorectal atresia, lumbosacral agenesis). In Sirenomelic fetuses, bilateral renal agenesis causes severe oligohydraminos, hindering a reliable sonographic evaluation of the lower extremity in the second and third trimester. In some cases, bilateral renal agenesis is the only sonographic finding. Amnioinfusion and high-frequency transvaginal ultrasonographic probes have proven to be very useful in such a situation.[11]MRI (Magnetic Resonance Imaging) permits the evaluation of visceral lesions, avoiding the call for amnioinfusion (Which carries a risk of fetal injury, infection, membrane rupture and placental injury).

CONCLUSION: This rare syndrome, the Sirenomelia sequence, will continue to divide the scientific community as to its etiopathology and its classification. However, to be pragmatic, it seems that the emphasis should be placed both on prenatal diagnosis and genetics to ensure an optimal management that would consequently be less demanding both from a psychological point of view and a health cost point of view, especially in resource poor settings like India.

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AUTHORS:

1. Kirti M. Hurakadi
2. Rutuja Kolekar

PARTICULARS OF CONTRIBUTORS:

1. Assistant Professor, Department of Obstetrics & Gynaecology, HSK's Hospital & Research Centre & SN Medical College, Bagalkot.
2. Senior Resident, Department of Obstetrics & Gynaecology, HSK's Hospital & Research Centre & SN Medical College, Bagalkot.

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NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Kirti M. Hurakadli,
C/o. B. M. Hurakadli,
Nandi, Near Water tank,
Bhagawan Mahavir Road,
Bagalkot-587101,
Karnataka.
E-mail: mh_kirti@yahoo.co.in
rgundalli@yahoo.com

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