
Sirenomelia with Spina Bifida

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Sirenomelia is an exceptionally rare congenital malformation characterized by a single fused lower limb. This deformity is variably known as symmelia, sympodia monopodia, sympus and due to its resemblance to the mermaid of Greek and Roman mythology popularly also known as mermaid baby. Sirenomelia has prevalence of 1.5 - 4.2 cases per 100,000 births(1). About 300 cases of this lethal anomaly have been reported in the world literature(2). In India over the last 15 years, 8 reports have appeared describing this condition(3). Most of these reports have only given the description of the dysmorphism but not have made any attempt to study the possible pathogenesis of the syndrome. In the present report we have done a postmortem arteriogram study to find out the possible vascular abnormalities which are supposed to be one of the mechanisms in the causation of this disorder(4). We also describe and discuss the association of neural tube defects along with sirenomelia.

Case Report

A 23-year-old, gravida two, with 8 months amenorrhea was admitted with premature rupture of membranes and onset of labor pain. Her antenatal period was uneventful and there was no history of any drug intake. There was neither a past or a family history of malformations.

A 1.6 kg infant was born by breech delivery. The baby had severe birth asphyxia (Apgar score was 2 at 1 minute and 4 at 5 and 10 minutes). All attempts to resuscitate the baby failed and the baby died after 40 minutes. Physical examination of the infant showed soft tissue fusion in lower extremities except the feet (Fig. 1). The feet were normally formed with toes, normal in number and appearance. Genitalia was represented by a small bud like projection. The anal and urethral openings were absent. Umbilical cord showed a single umbilical artery. The child had normal upper half of the body. The placenta was normal.

Radiological studies demonstrated normal bony structures in the lower extremities and feet. There was bilateral dislocation of hip and crowding of ribs. All the cervical vertebrae and thoracic vertebrae T1 to T4 showed spina bifida defect. The heart was of normal size and lungs were hypoplastic (Fig. 2).
A postmortem arteriogram was done which showed narrowed abdominal aorta which divided at higher level. The superior and inferior mesentric arteries and renal arteries were not visualized (Fig. 2).

Autopsy showed hypoplastic lungs and the rectum ended in a blind cord like structure. Both the kidneys, bladder, pancreas and liver were present and were apparently normal.

Discussion

Sirenomelia is an anomalous development of the caudal region of the body with different degrees of fusion of the lower extremities with or without long bones being present. Along with fused lower limbs sirenomelia is associated with various anomalies like hypoplastic lungs, cardiac defects, absence of genitalia, colon and anus, agenesis of kidneys and urinary bladder, single umbilical artery, tracheo esophageal fistula, radial, thumb and rib defects, vertebral and central nervous system anomalies, etc. (1,5,6). Because of the associated visceral anomalies most cases of sirenomelia are still born or die shortly after birth. Even though consen-
sus is that the anomaly is incompatible with life, two case reports have documented survivors with sirenomelia(7).

Till recently sirenomelia was thought to be a part of caudal regression syndrome resulting from a defect in the formation and development of the caudal region of the embryo. The malformation results from variable disturbances of the structures derived from caudal mesodermal axis of the embryo presumably prior to 4th week and extend to various cranio caudal levels. The exact etiology leading on to the disturbance is unclear(8).

The presence of a single umbilical artery as a constant feature of the syndrome led to an alternative etiological theory. It has been postulated that the umbilical artery arises high up in the aorta and there is hypoplasia of the vasculature distal to it which leads to nutritional deficiency to the caudal half of the body(8). In 1986 Stevenson proposed vascular steal theory and suggested that blood flow is diverted from the caudal portion of the embryo to the placenta through an abdominal artery which arises high in the aorta and assumes the function of umbilical arteries(4). Consequently there may be complete agencies, incomplete or aberrant development of tissues which depend on
nutrition from arteries distal to it. This explains the associated renal, genital gastrointestinal anomalies(4). The single umbilical artery noticed in our patient and the post mortem arteriogram showing narrowed abdominal aorta and iliac arteries favors the vascular insufficiency as the pathogenic mechanism.

Unusual association of anencephaly in 6 cases and limb body wall complex in 3 cases with sirenomelia have been reported in the literature(9,10). Cases of associated meningocoeel, Arnold Chairi malformation, etc. have also been reported(4,9). In the present case there was spina bifida involving proximal vertebrae. Tang et al. in 1991 suggested that embryonic vascular disruption may be responsible for limb body wall complex and sirenomelia(5). Angiographic and vascular anatomic studies in anencephaly and other neural tube defects have not yet received attention(6). This group of anomalies which has etiological heterogeneity may also be attributed to vascular disruption. In that case, vascular defects not only of the lower half of the body but also other arteries may be the pathogenic mechanism of associated defects in sirenomelia.

Seven per cent of cases of sirenomelia are associated with monozygotic twinning(3). Association of various cranial anomalies and observed anomalies in the other twin, points that the process may be related to midline vulnerability in the early process of development. The vulnerability of midline can lead to both duplication and deficiencies(3). The twinning process, in a sense, the ultimate midline anomaly, predisposes the midline to further problems and could account for anomalies observed. Male preponderance is seen in midline defects, so also in sirenomelia(9). The male to female sex ratio is 2.7:1(1). But recently in a report of 11 cases of sirenomelia by Sertori et al. in 1992, the male to female ratio in the total series was 9:0 (with two cases in which sex was undetermined)(8). X linked oligogenes and X linked mutations are suggested to explain the excess of males observed in midline defects(9).

No chromosomal abnormalities are observed in sirenomelia and it usually does not recur in families(5). As this is an uniformly lethal condition the antenatal diagnosis and option of termination of pregnancy gains importance. The antenatal diagnosis of sirenomelia by X-ray and ultrasonography has been reported. By using ultrasonography a diagnosis can be made in second trimester(8). The ultrasonographic features that help in the diagnosis are lack of visualization of both tibia and/or fibula with or without feet or identification of a single femur, convergent femoral bones lying in a side by side configuration that does not change with time and presence of oligohydramnios in the second trimester, premature rupture of membranes, bilateral renal agencies, polycystic kidneys, obstructive uropathy and severe intrauterine growth retardation(8). In situations when ultrasonography is inconclusive or suboptimal, magnetic resonance imaging is useful in confirming the ultrasound impressions and provides more complete characterization of the extent of the fetal anomaly(2).

REFERENCES


Mothers’ Beliefs and Practices Regarding Prevention and Management of Diarrheal Diseases

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Several community-based studies have demonstrated that most of the diarrhea related deaths can be prevented by appropriate and timely use of Oral Rehydration Therapy (ORT)(l-3). However, even after a decade of active promotion, only one third of the diarrheal episodes are treated by ORT(4). Though availability is an important factor but beliefs and attitudes also influence the use of ORT. The objective of this study was to find out the change in mothers’ beliefs and practices after implementation of the diarrheal diseases control programme, and to use the information for improving the promotional strategy.

Material and Methods

Two villages of Raipur Rani block in Haryana, were purposively selected where Diarrheal Disease Control Programme is being implemented for almost a decade. A list of mothers having at least one child of less than 5-years-old was prepared and 48 mothers were selected using systematic random sampling method. Mothers were interviewed in the month of July by using a pretested semi-structured interview schedule.

Results

Most of the mothers described multiple causes for occurrence of diarrhea. Consumption of uncovered food, eating ‘dirty’ or