A TYPICAL CASE OF SIRENOMELIA - THE MERMAID SYNDROME

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ABSTRACT

BACKGROUND

Sirenomelia is a very rare and a lethal polymalformative syndrome characterised by fusion of lower limbs.

Case characteristics: Live sirenomelic baby born to a 22-year-old primigravida mother at 35 weeks of gestation with a normal antenatal scan.

Outcome- Infantogram showed a single femur and an incomplete tibia. USG abdomen revealed B/L renal agenesis, the intestines ended with a blind ileal pouch. Colour Doppler showed the presence of a single large umbilical artery. The baby died at 10 hours of life.

Message- It is a very rare and a lethal congenital anomaly. Efforts should be made to diagnose it prenatally and medical termination should be offered.

KEYWORDS

Sirenomelia, Caudal Regression Syndrome, Anhydramnios, Polymalformative Syndrome.


BACKGROUND

Sirenomelia is a rare polymalformative syndrome,[1] characterised by fusion of lower limbs. It is incompatible with life because of severe congenital anomalies associated with it.

We are reporting a case of a live born sirenomelic baby, to a 22-year-old primigravida mother, extracted through caesarean section at 35 weeks of gestation.

We are reporting this case because of its rarity, term live birth, radiological misdiagnosis and its presentation with all the typical features.

CASE REPORT

A 22-year-old primigravida from a tribal community with a married life of 1 year, second degree consanguinity, was admitted in labour. She was healthy during her pregnancy. No history of fever with rashes, diabetes, radiation exposure and substance abuse. USG done at 32 weeks was normal. No history of congenital anomalies in her family. She underwent a C-section, in view of breech presentation and delivered a 1.2 kg baby. The baby had a weak cry at birth. No active resuscitation was made. The baby survived for 10 hours.

On examination, the baby had prominent epicanthal folds, hypertelorism, low set ears, cleft lip, cleft palate, retrognathia, a single lower limb with absent digits, absent external genitalia, imperforate anus and an umbilical cord with a single umbilical artery.

Infantogram showed a single femur and an incomplete tibia with gross scoliosis. USG abdomen revealed B/L renal agenesis, absent ureters and urinary bladder. The intestines ended with a blind ileal pouch with no colon, rectum and anus. Colour Doppler showed the presence of a single large umbilical artery.

Figure 1
DISCUSSION

Sirenomelia as mentioned before is one of the rarest congenital anomalies. The incidence is 0.8 to 4.2 cases/10,000 births.[1] Most of them die and present as still births. 9 to 15% of sirenomelia are associated with twin monzygotic pregnancies.[1] There is a strong association with maternal diabetes as suggested by Welch et al[2] and about 22% of foetuses with this anomaly will have mothers with diabetes.[3] Maternal use of tobacco, [3] cocaine,[4] heavy metal poisoning like cadmium,[5] radiation exposure are also identified as some of the risk factors.

Earlier it was considered as a severe form of caudal regression syndrome; however, now it is identified as a separate entity.[1] Pressure theory, theory of primary failure, theory of nutritional failure, theory of caudal regression, theory of neural tube over distension, etc.[6] have explained the pathogenesis of sirenomelia. Of the many proposed theories, the vascular steal hypothesis by Stevenson et al[7] and defective blastogenesis hypothesis[3] are the most popular ones. According to the former one, there is decreased blood supply to the caudal mesoderm which results in agenesis of midline structures and subsequent approximation of both lower limb fields. Defective blastogenesis hypothesis states that a teratogenic event occurs during gastrulation stage at around 4th gestational week, resulting in abnormal development of caudal structures.

It is incompatible with life because of its association with several visceral abnormalities especially due to renal agenesis and pulmonary hypoplasia.[1] Exceptional cases without renal agenesis have survived. Clarke et al[8] reported a sirenomelic baby diagnosed antenatally with fused limbs and renal dysplasia, on whom surgery was planned at 3 months of age. But such interventions are costly and are unpredictable.

The facial abnormality usually seen in sirenomelic infants is Potter’s facies secondary to anhydramnios. In our case along with typical description of Potter’s facies,[3] there was B/L cleft lip and cleft palate unlike the case reported by S K Hatwar[6] where there was no facial dysmorphism. The baby had respiratory distress and eventually died of respiratory failure. This could be secondary to pulmonary hypoplasia. In that case it would constitute Potter’s syndrome (Oligo/anhydramnios associated with pulmonary hypoplasia).
As according to the classical description, our case had a single limb with no digits. Infantogram revealed a single femur and an incomplete tibia. Hence, it belongs to Stocker and Heifetz classification type VI.\(^9\)

The infant had B/L renal agenesis with absent bladder and ureters. The gastrointestinal tract ended as a blind ileal pouch with imperforate anus and there was a single large umbilical artery. These features were similar to many previously reported cases.

The baby died 10 hours after birth. Autopsy was not done as per the parent’s request.

In this case, USG done at around 32 weeks failed to pick up the gross anomaly. Similar failures have been reported earlier.\(^1\) Antenatal detection of sirenomelia is not straightforward. Secondary to B/L renal agenesis, there is severe oligohydramnios or anhydramnios obscuring the foetal parts during sonographic examination. High frequency transvaginal probes following amnioinfusion are very useful in these situations. Hence, cases associated with oligohydramnios early in second part of pregnancy must be carefully screened for renal agenesis or dysgenesis. It is also very important to screen diabetic pregnant women, pregnant women with substance abuse for sirenomelia as early detection will allow early termination of pregnancy.

CONCLUSION
It is a very rare and a lethal congenital anomaly. Efforts should be made to diagnose it prenatally and medical termination should be offered.

Educating mothers on substance abuse, optimum control of diabetes could possibly reduce its incidence.

REFERENCES