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Sirenomelia: A Case Study.

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ABSTRACT

Sirenomelia is a rare anomaly. It involves caudal region of the body. Presents with fusion of lower limbs in which Genito urinary, Gastrointestinal, Cardiovascular and Neural tube anomalies are seen. We present a case with severe oligohydramnios, with features of Sirenomelia. We present another case of sirenomelia to make an earlier diagnosis and terminate the pregnancy early and safely.

Keywords: Sirenomelia Anomaly, stillborn, malformation, mermaid syndrome.

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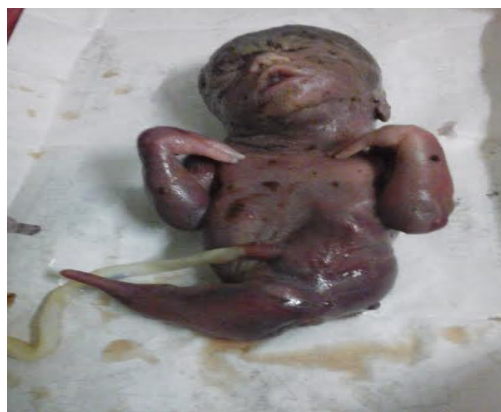
INTRODUCTION

Sirenomelia or Mermaid syndrome was described by Rocheas in 1542 and Palfya in 1553. Homer explained that sirens are tempting singing creatures that seduced sailors to their deaths. Many European countries referred sirens to female hybrids. Incidence is 1 in 1, 00, 000 live births. This malformation is the complete or partial fusion of the lower limbs. It includes renal agenesis, ambiguous external genitalia, imperforate anus and blind Intestinal loop. Double inferior Vena cava and angiomatous lumbosacral myelo cystocele are also associated. Most of the Sirenomelia are stillbirth. Only a few were alive after delivery. It is associated with maternal diabetes. We present a fetus with features of sirenomelia.

Case study

A 25 year old non- diabetic primigravida with an supervised pregnancy of 21 +3 weeks was admitted for MTP. Routine blood investigation normal. TRIPLE screen had low risk , TORCH investigation revealed Rubella Ig G and CMV Ig G reactive. Ultrasonography at 13 week revealed no anomaly. Repeat scan revealed a dead fetus of 21 +3 weeks, severe oligohydramnios? , spina bifida ? / tibia with absent fibula , ? Single umbilical artery. Came to us for second opinion. Repeat Ultrasonography revealed 21 weeks, severe oligohydramnios, B/L renal agenesis/ nonfunctioning kidney, single umbilical artery. Severe caudal malformation, including fused lower extremities. Till then pregnancy was uneventful except for vomiting in 1st trimester and no significant medical, personnel, obstetric history and no congenital anomalies in family. There was no history of any drug intake. It was non consanguineous marriage.

She underwent MTP. Placenta was removed by evacuation and curettage. Expelled a dead, malformed fetus of unidentified sex weighing 400 g. The stillborn had fused lower limbs and absence of anus. Postmortem radiography showed sacral agenesis, a hypoplastic pelvis, and fused lower extremities with tibia but absent fibulae. Autopsy examination confirmed renal agenesis and revealed hypo plastic lungs, absence of anus. A single umbilical artery was present.



Contraceptive advice was given for 4 months. Genetic counseling was advised. Informed about sporadic incidence and negligible recurrence of human sirenomelia. She was asked to go to specialized prenatal center before next pregnancy.

CONCLUSION

Sirenomelia is a multisystemic human malformation of unknown etiology. Clinical observations have provided important insights into the problem and given rise to causal hypotheses that can now be tested and analyzed at the molecular level owing to the availability of animal models.

DISCUSSION

Mermaid syndrome or sirenomelia is a rare disorder. Infant is born with fused legs, giving the appearance of a mermaid's tail. Incidence is 1 in every, 100,000 births. The term "sirenomelia" comes from "siren" or "mermaid."

Malformations include a single lower limb, the presence of two feet or one foot or both feet could be missing. Has one [1] umbilical artery instead of two. Most of them are stillborn and the rest are fatal within a few days of birth. Complications are associated with the kidneys, bladder, genitals, heart deformities, and lungs. Cause of death is due to main organs doing not form normally.

The three survivors are Milagros Cerran, Shiloh Pepin and Tiffany Yorks.

Mermaid Syndrome Genetic

Mermaid syndrome develops early in fetal development when blood is diverted from cartilage, bones, muscles, nerves and blood vessels (the caudal structures) to the placenta. It's this lack of blood flow that causes the limbs to fuse, as the lower extremities are starved of oxygen and nutrients. Why this happens is still a mystery.

Several ideas have been put forward and these include poor health of an expectant mother, exposure to large amounts of radiation that creates genetic mutations, and/or a major illness suffered by the expectant mother.

There may be a series of genetic mutations, but if there are, they have not been identified. However, it has been established that sirenomelia can have a genetic basis in mice. Sirens have been observed when some strains have been crossed and as a consequence of mutations that boost the levels of retinoic acid. Researchers have also reported that sirenomelia has been caused by a loss of Bone morphogenic protein 7 (Bmp 7) [2] in combination with a complete loss or half dose of another Bmp protein - called twisted gastrulation (Tsg). Bone morphogenic proteins are multi-functional growth factors that play crucial roles in bone and cartilage development.

All incidences of Mermaid syndrome have occurred as isolated cases in families. It could be that each case represents a new spontaneous mutation.

There is also a possibility that sirenomelia does not have a genetic component; some doctors postulate that an environmental trigger could cause the malfunctions with the blood circulatory system that result in organs not developing correctly.

Another idea is that it could be caused by direct damage to the caudal region of the embryo by excessive distention of the neural tube.

Treatment

The condition is usually fatal, but for those that survive surgery is the only option to separate limbs. Other treatments depend on the extent of the birth defects.



REFERENCES

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- [2] Lise Zakin et al. Development 2005;132:2489-2499.