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RESEARCH ARTICLE

Sirenomelia (Mermaid syndrome): A case report.

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Abstract

Sirenomelia, Mermaid syndrome is a rare and lethal congenital malformation characterized by the fusion of the lower limbs,commonly associated with urogenital and gastrointestinal malformations.The specificetio-pathogenesis of sirenomeliais not well established. We present a case of neonate with clinical features suggestive of sirenomelia.

Background:-

Sirenomelia also known as Mermaid Syndrome or SirenomeliaSequence, is a rare congenital malformationwith an incidence of 1.1–4.2 of 100,000 births.[1]It is characterized by the fusion of the lower limbs with associated urogenital and gastrointestinal malformations thatinclude absent external genitalia, imperforate anus,vertebral abnormalities andrenal agenesis.[2]Sirenomeliainfants may present with Potter's facies, characterized by low-set ears, epicanthic folds, hypertelorismand retrognathia.[3]

Sirenomalia is a fatal anomaly due to bilateral renal agenesis which leads to severe oligohydramnios and lung hypoplasia; although, less severe cases have been reported to survive with appropriate surgical management.[4]The precise etio-pathogenesis of Sirenomelia is not well established.The disorder was formerly thought to be an extreme case of caudal regression syndrome[5], but presently it is reclassified as a separate condition.Early prenatal diagnosis by first trimester scan and counseling of parents should be the aim to minimize the trauma related to the mortality and morbidities associated with Sirenomelia.[6]

Based on the clinical diagnosis we present a case of Sirenomeliain a neonate at birth.

Case report:-

A preterm neonate with 34-36-weeksgestational age, birth weight2450gms, was born out of non-consanguinity to a 22-year-old primi-gravida mother, married since 5yrs.She had primary infertility and received medical treatment for the same, before this pregnancy.Maternal history was not suggestive oftobacco use,diabetes mellitus or any heavy metal exposure.

Antenatal USG performed in third trimesterwas suggestive of severe oligohydroamnios.The baby was delivered by emergencylower segment caesarean section andrequired resuscitation at birth.

On physical examination baby had single umbilical artery with multiple external deformities including a single lower extremity with no feet, imperforate anus and absent external genitalia[Figure 1].Facial featureswith low set

ears,epicanthicfolds,hypertelorism and retrognathiawereindicative of Potter's facies. The neonatesurvivedfor only 30 minutes after birth.

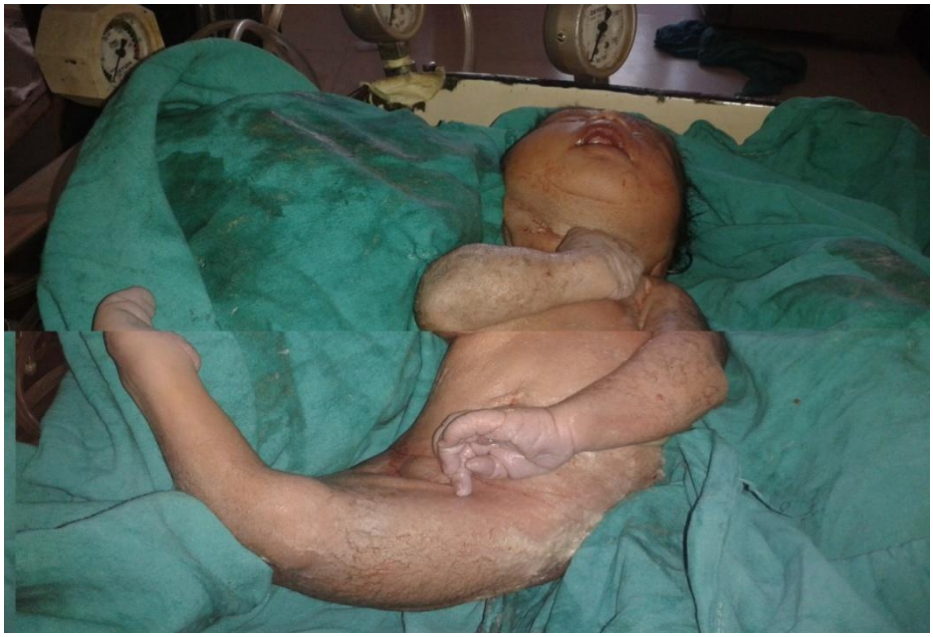

Figure 1:- Photograph showing features of Sirenomelia

Discussion:-

Sirenomelia is a fatal congenital anomaly.Only few cases are reported to survive neonatal period.Treatment of surviving infant involves a multidisciplinary surgical management.[7]

Incidence of Sirenomelia is reported to be 100-150 times higher in monozygotic twins than dizygotic twins or singletons.[8]Sirenomelia hasa strong association with maternal diabetes where relative risk is 1: 200-250 and, up to 22% of fetuses with this anomaly are reported to have diabetic mothers.[9]Few proposed hypotheses for etiopathogenesis of Sirenomeliaare as follows:

1. Defective blastogenesis hypothesis postulates that there is defective development of caudal mesoderm due to teratogenic exposure to tobacco[10],retinoic acid[11]or cocaine[12] and maternal metabolic derangement due to diabetes[9]during the gastrulation stage. Intracytoplasmic sperm injection technique is also proposed to be a causal factor under this hypothesis .[13]
2. The vascular steal hypothesis suggest that fusion of the limbs and agenesis of midline structures results from a deficient blood flow and nutrient supply to the caudal mesoderm.[14]
3. Genetic origin of Sirenomelia is endorsed by experimental animal datain genetically modified mouse strains.[1]

Sirenomelia is classified into seven types according to the presence or absence of bones within the fused lower limbs:[14]

1. Type I fusion involves superficial tissues only with presence of all bones.
2. Type II fusion involves superficial tissues with presence of single fibula.
3. Type III fusion involves superficial tissues with absence of both fibula
4. Type IV involves partial fusion of femur and foot bones.
5. Type V, partially fused femur and tibia with single foot bones.
6. Type VI,single femur and single tibia with no foot bone.
7. Type VII only a single bone is present

Though radiograph was not possible, clinical examination was suggestive of Type V.

Early prenatal diagnosis by first trimester scan and counselling of parents should be the aim to minimize the trauma related to the mortality and morbidities associated with Sirenomelia.

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