ORIGINAL RESEARCH PAPER
Volume-9 | Issue-5 | May-2020 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

SIRENOMELIA : A RARE CASE REPORT

Paediatrics

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ABSTRACT

Mermaid syndrome is also known as 'Sirenomelia', a fetal syndrome diagnosed in neonates with characteristicfeature of complete or partial fusion of lower limbs. We report a case of preterm baby delivered to a 28 years old mother. Examination of the baby revealed fused both lower limbs and feet with 10 toes, absent external genitalia and features of Potter's facies like slanting forehead, depressed nasal bridge and low set ears. Baby died one day post birth due to severe respiratory distress. Autopsy findings revealed absent kidneys, urinary orifice and imperforate anus.

KEYWORDS

INTRODUCTION :

Autopsy plays an important role in identifying cause of death in neonates. It also gives inputs to clinicians and parents to plan further pregnancies considering neonatal deaths due to congenital anomalies. Mermaid syndrome is also known as 'Sirenomelia', a fetal syndrome diagnosed in neonates with characteristic feature of complete or partial fusion of lower limbs.

Mermaid syndrome is an extremely rare anomaly, an incidence of 1 in 1,00,000 births, in which a baby born with legs joined together featuring a mermaid like appearance(head and trunk like human and tail like fish). The disorder is referred as symmelia or sympodia or monopodia or sympus but most commonly as the 'mermaid syndrome'since the fusion of lower limbs gives a characteristic mermaid like appearance[1]. So far 300 patients with this rare anomaly have been reported in the world. The incidence of male : female is 3:1[2]. Gastrointestinal, urogenital anomalies and single umbilical artery are clinical outcome of this syndrome.

CASE REPORT :

A preterm neonate with ambiguous genitalia and fused lower limb was born to a 28 year old mother. The neonates birth weight was 1.5 kg, baby was shifted to NICU in view of multiple congenital anomalies but died one day post birth due to severe respiratory distress. The mother did not have any health issue or history of anydrug intake and radiation exposure. Her antenatalUSG scan showed gross oligohydramnios and partially visualized bilateral kidneys. Her anomaly USG scan showed normal liver, bile duct, intestine but bilateral kidneys were not visualized. The new born baby had gross anomalies like Potters' facies, which include slanting forehead, depressed nasal bridge, low set ears. The infant also had featureslike left hand polydactyly, fused both lower limbs and feet with 10 toes, absence of external genitalia as shown in photograph 1.

Photograph 1 - Mermaid baby

Examination of heart, liver,brain, adrenal and intestine revealed normal anatomy. Autopsy findings revealed absent kidneys, urinary orifice, imperforated anus and features suggestive of pulmonary hypoplasia(Fig.1).

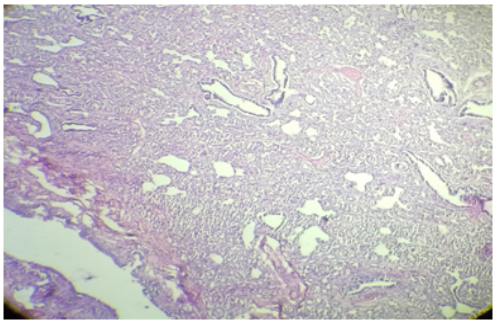

Figure 1 – Microscopy of lung(H & E 100X) showing pulmonary hypoplasia.

So the final impression given was Mermaid syndrome with bilateral pulmonary hypoplasia with pulmonary hemorrhages with bilateral renal agenesis.

DISCUSSION :

Mermaid syndrome is an extremely rare anomaly that was found in year 1542 by Rochel et al[3]. Mermaid means trunk looks like human and limbs look like a fish[3]. Anomalies that are commonly seenwith mermaid syndrome include : cleft palate, pulmonary hypoplasia,cardiacdefects, omphalocele and meningomyelocele[1]. The associated anomalies may include bilateral renal agenesis, complete or partial agenesis of genitourinary system, imperforate anus, absence or ambiguous external genitalia, single umbilical artery, lung hypoplasia, vertebral and cardiac anomalies.[1]Gestational DM, mothers younger than 20 year and older than 40 years, hyperthermia, exposure to teratogenic factors like cadmium, lithium, phenytoin etc are vulnerable factors to cause mermaid syndrome.

There are 2 hypothesis : vitellineartery steal hypothesis and defective blastogenesis hypothesis[4].In vitelline artery steal hypothesis there is shunting of blood via an abnormal abdominal artery that leaves the caudal end of embryo poorly perfused causing complete or incomplete agenesis of caudal structure.[5]In defective blastogenic hypothesis, the primary defect in development of caudal mesoderm is attributed to a teratogenic event during the gastrulation stage. Such defect interferes with the formation of notochord resulting in abnormal development of caudal structure.[6]

Sirenomalia is usually fatal within a day or two of birth because of complications associated with abnormal kidney and urinary bladder development and function. In literature around 300 cases are reported worldwide of which 14 are from India[7].

When features of Potter's facies are combined with oligohydramnios

and pulmonaryhypoplasia , it is known as Potter's syndrome which was present in our case[8]. Features of Potter's facies includes large, low set ears, prominent epicanthic fold, flat nose and recending chin which were present in our case. Horikoshi et al[9]reported two cases of sirenomalia.

Mermaid syndrome is fatal in most cases due to pulmonary hypoplasia and renal failure resulting from renal agenesis[10]. Pulmonary hypoplasia is calculated by taking ratio of lung weight to body weight. When it comes <0.22 pulmonary hypoplasia is diagnosed.In our case it was 0.01.

CONCLUSION :

Sirenomelia is a rare and lethal congenital anomaly and this case gives us valuable information about the clinical presentation of it at birth & its subsequent post mortem findings are helpful for the diagnosis. Regular antenatal checkups with optimum maternal blood glucose level with prevention from exposure to teratogenic drugs should prevent this anomaly. Early onset oligohydramnios is an alerting sign and detailed anomaly scan should be done so termination of pregnancy can be planned.

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