The mermaid syndrome (sirenomelia): a rare case report

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Abstract

Mermaid syndrome is a very rare fatal congenital anomaly characterised by fusion and atrophy of lower limbs giving an appearance of human head and upper body with tail of a fish. Incidence of the condition is 1 in nearly 100,000 pregnancies with unknown aetiology. Here we present a case of mermaid syndrome which was a live born and delivered vaginally at term of third gravida tribal women of lower socio-economic status. We examined the baby clinically and found caudal dysgenesis having fusion of lower limbs, single leg with one foot and three toes. There were no anus and external genitalia. We report such a case due to its rarity and term live birth.

Key words: Congenital anomaly, caudal dysgenesis, mermaid syndrome, rarity.

Introduction

Mermaid syndrome is a very rare fatal congenital anomaly[1]. Its incidence is 1 in 100,000 pregnancies[2] and cases have been reported in all ethnic groups worldwide[3]. It is so rare that many medical men have not seen such a case in their medical professional carrier. The most prominent feature of this congenital malformative disorder is the complete or partial fusion of the lower limbs into a single limb[3]. The resultant infant bears a resemblance to the mermaid of ancient Greek mythology[4]. The underlying visceral abnormalities are usually such that the syndrome is incompatible with life[3] but there are number of reported cases of surviving infants with this condition in English literature[5-9]. Here we report such an interesting case due to its rarity and its term live birth.

Case report

A 25 years old tribal unbooked mother, P_{1+1}, admitted in the labour room of Malda Medical College and Hospital, India, with labour pain at term on 5.10.2013 at 1.15 pm. She had no history of diabetes mellitus and no history of intake of teratogenic drugs, neither had history of intake of any abortificient drug. There was no family history of diabetes or malformed baby also. Her first pregnancy was a full term live birth and second pregnancy underwent spontaneous miscarriage at 3 months. No ultrasonography was done in the present pregnancy.

On admission the patient was haemo-dynamically stable with term size fetus in cephalic presentation with regular fetal heart sound. On p/v examination os was fully dilated, cervix was fully taken up and liquor was clear. She delivered vaginally but surprisingly we found a congenitally malformed live born baby of birth weight 2.5 kgs with the following features (Figure 1-3).

1. Distended abdomen
2. Undetermined sex
3. Umbilical cord with single umbilical artery
4. Imperforate anus
5. Absence of urinary meatus
6. Fused lower segment into a single lower limb with one foot and three toes

Figure 1. **Front view of the baby.**

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Figure 2. **Single lower limb with one foot and three toes.**

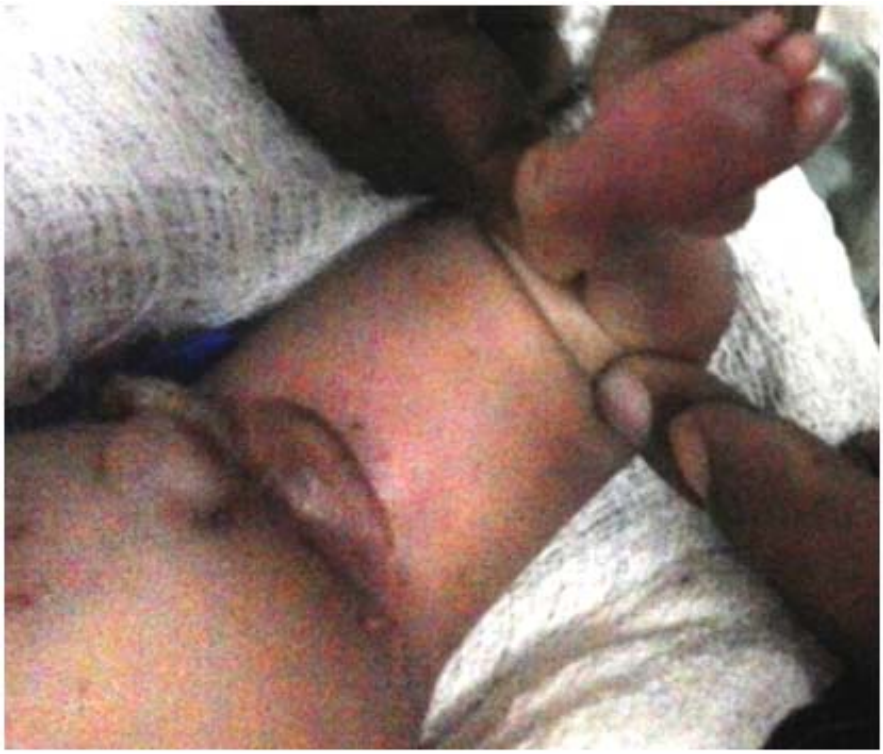

Figure 3. **: Back view of the baby.**

We did not have any radiograph and could not therefore classify our patient into any type of Stocker and Heifetz classification with certainty but through external examination was in favour of type IV (simpus monopus-fused lower limb with single foot).

Discussion

The first medical description of sirenomelia was by Rochens and Polfyr way back in sixteen century. Most of the mermaid babies were still born and dead usually due to renal agenesis. Cases of sirenomelia surviving beyond the perinatal period have been reported by Savadar et al for the first time in 1989[5] and later by Murphy et al in 1992[6]. Our case was live born and was referred to paediatric surgery department of higher centre.

Stocker and Heifetz classified the sirenomelia into seven types[10] as following:

Type	*Characteristic*
I	All thigh and leg bones are present
II	Single fibula
III	Absent fibula
IV	Partially fused femurs and fused fibula
V	Partially fused femurs
VI	Single femur, single tibia
VII	Single femur, absent tibia

Our patient belongs to type IV having partially fused femur and fused fibula as per clinical examination. Vascular steal theory has been proposed by Stevenson et al[11] as the primary mechanism due to diversion of blood flow away from the caudal region of the embryo through the abdominal umbilical artery. The single umbilical artery in our patient favours this theory. However Jaiyessimi et al[12] reported a case of sirenomelia without vitelline artery steal which indicates that factors other than vitelline artery steal could be responsible for sirenomelia in humans. In 1961 Duhamel[13] proposed

the theory of caudal regression resulting in a spectrum of malformation including anal imperforation and the mermaid syndrome. He called it the syndrome of caudal regression. But caudal regression is well differentiated from mermaid syndrome having features like two umbilical arteries, non lethal renal anomalies, non fused lower limb, abdominal wall defect, tracheo-esophageal defect, neural tube and cardiac defect and usually associated with maternal diabetes[14]. However with only about 0.5%-3.7% of sireno-melia cases occurring with diabetic mothers[15,16]. Association between sirenomelia and maternal diabetes has been described as weak[12]. Our patient was not known to be diabetic. Retinoic acid and heavy metal exposure are associated with sirenomelia[17, 18]. There was no history of heavy metal exposure or intake of retinoic acid in our case. Sometimes sirenomelia is regarded as a part of VACTERAL syndrome. VACTERAL syndrome involves vertebral, anal, cardiovascular, trachea oesophageal, renal and limb dysgenesis. There is major overlap in phenotypic manifestation of sirenomelia and VACTERAL[12]. But as our patient was a live born, autopsy could not be done and we have no radiographic proof to ascertain any relationship with VACTERAL as we referred the baby to higher centre for better management.

Conclusion

Sirenomelia remains a rare but peculiar syndrome and no definite etiopathogenesis is identified and sporadic in nature. It is usually not compatible with life and hence prenatal diagnosis by USG before 20 weeks is necessary for termination of pregnancy to free the mother from great psychological trauma and to free the family from economic loss if live born. ■

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