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Original Article

Sirenomelia: A Fetal Autopsy Study

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ABSTRACT

Introduction: Sirenomelia is a very rare congenital, evolutionary malformation in the caudal region with varying degrees of lower limb adhesion. Fetus looks like of Mermaid of Greek mythology, and hence, the synonym of Mermaid syndrome is also given to the defect.

Materials and Methods: The study was done in the Department of Anatomy in Government Medical College, Chandigarh, on fetuses sent for autopsy from Gynecology and Obstetrics department. The obstetric histories of all the mothers were taken thoroughly. The medical histories, drug history, qualification, and socioeconomic status of the parents were also noted. Fetuses were examined externally. Photograph and radiograph of each case were taken. Autopsy of all the fetuses was done for internal examination by following directions in Cunningham's manual.

Results and Conclusion: In our study, we came across fetuses with sirenomelia out of 1900 cases, which accounts for about 0.002%. In three cases, one femur falling in between simpus apus and simpus unipus. Case number one, with some variation in looked like simpus unipus, because it has two femur and two tibia and one foot. The etiology is unknown in all cases but hypothyroidism and anemia may have been a factor in the two reported cases.

Key words: Sirenomelia, Mermaid syndrome, Congenital malformations

INTRODUCTION

Sirenomelia is a very rare congenital, evolutionary anomaly, in which baby is born with defect in the caudal region of the body with varying degrees of lower limb adhesion. The defect may lead to complete absence of the lower limb to joining or adhesion of part of lower limb. It is also associated varying degree of defect in perianal region.[1] Fetus looks like of Mermaid of Greek mythology, and hence, the synonym of Mermaid syndrome is also given to the defect.[2] The prevalence of this anomaly is 1 in 100,000 births.[1] The anomaly is lethal in severe cases. Incidence of stillbirth is 50 %. Defect is much more common in identical twins.[3] In live born babies, head and trunk are such as humans and tail looks like fish.[2] Most live born cases die shortly after birth.[1] A spectrum of anomalies affects primarily the musculoskeletal, genitourinary, and gastrointestinal systems.[4]

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Kallen suggested that diabetes may be a cofactor that modifies the action of one or more unknown teratogens.[5]

Raabe *et al.* reported sonographic antenatal diagnosis of sirenomelia at 18 week' gestation as evidenced by (i) oligohydramnios, (ii) bilateral renal agenesis (non-visualization of the kidneys and bladder on serial prenatal examinations), and (iii) fusion of the lower extremities.[6]

Sirenomelia is closely associated with Potter's syndrome. It consists of (i) Potter's facies (large, low-set ears, prominent epicanthal folds, hypertelorism, flat nose, and receding chin), (ii) oligohydramnios, and (iii) pulmonary hypoplasia. This syndrome is almost invariably present with bilateral renal agenesis. Although a strong association between caudal regression syndrome and maternal diabetes has been described, no such relationship exists between Potter's syndrome and diabetes.[7]

MATERIALS, METHODS, AND OBSERVATIONS

The study was done in the Department of Anatomy in Government Medical College, Chandigarh, on fetuses sent for autopsy from gynecology and obstetrics department.

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The obstetric histories of all the mothers were taken thoroughly. The medical histories, drug history, qualification, and socioeconomic status of the parents were also noted [Table 1].

Fetuses were examined externally [Table 2]. Photograph and radiograph were taken. Autopsy of all the fetuses was done for observing internal examination [Table 3] by following directions in Cunningham's manual. Associated

Table 1: Case histories

	CASE I	CASE II	CASE III	CASE IV
Maternal history	24 years G_2A_1	30 years $G_3P_1L_1A_1$	26 years $G_3P_1L_1A_1$	24years, $G_3L_2A_1$
Gestational age and sex	20 weeks Un differentiated	18^{+4} weeks Un differentiated	19 weeks Appears to be Male	14^{+2} weeks Not determined
History of previous pregnancy	Spontaneous expulsion 1 year back	1 live female child 2^{nd} pregnancy history not available	Not available	Normal, 2 live female child
Past history	Hypothyroidism	Not suggestive	Not suggestive	Anaemia in mother
Family history	Not suggestive	Not suggestive	Not suggestive	Not suggestive
Antenatal history of present pregnancy	Regular intake iron, folic acid and vaccination	Not available	Medicines for hypothyroidism up to 2 weeks present gestation	T.T injection given to mother
Investigations	Multiple anomalies	Cystic hygroma	Multiple anomalies	USG not done
Termination of pregnancy	Spontaneous Expulsion	Spontaneous Expulsion	MTP	MTP

Observations: Four cases of sirenomelia were found in 1900 cases of fetal autopsies attempted

Table 2: Showing the findings on External examination

FEATURES	CASE I	CASE II	CASE III	CASE IV
Head, curvature of spine	Antero posteriorly lengthening of skull, different shape of pinna on both sides	Low set ears, prominent epicanthal fold	Broad nasal bridge, asymmetrical ear.	Normal
Limbs	Both lower limb joined till thigh. Below knee fused limb has appendage on the right side, left leg terminated in foot which has polydactyly	Fused lower limb, feet without toes	Only one limb in midline seen, foot degenerated	Only one limb present , deviated towards left by the extruded abdominal contents
Thorax	Normal	Normal	Normal	Broad and short thorax
Abdomen	Absence of anal and urinary opening	imperforate anus	Absence of anal, and urinary opening small male genitalia	Abdominal contents protruding out
Number of umbilical vessels	Two umbilical Vessels	Two umbilical Vessels	Two umbilical Vessels	Umbilicus could not be seen

Table 3: Showing the findings on internal examination

FEATURES	CASE I	CASE II	CASE III	CASE IV
Skeletal system	2 femur, 2 tibia foot bones not visible	1 femur, 1 tibia, foot bones not visible	1 femur, 1 tibia, foot bones not visible,	X-Ray could not be taken
CNS	Normal	Normal	Normal	Normal
CVS	Normal	Normal	Normal	Thorax has only great vessels Heart shifted to abdomen , Present in midline between abdominal lungs
GIT	Blind gut end, Imperforate anus	Blind descending colon, megacolon, imperforate anus	Blind sigmoid colon, No anal opening seen	All the gut, liver extruded from abdominal cavity. Contents covered by only a membrane. Gut opens in cloaca along with both the ureters
Respiratory System	Fluid seen in thoracic cavity	Normal	Horizontal fissure not seen in right lung	Both the lung lying in abdomen embracing heart. Left lung partially lying in thoracic cavity. Big gap in diaphragm
Urinary system	Bilateral renal agenesis along with urinary tract and bladder, No opening of urinary tract seen	Kidneys present (Left kidney ascended)	Both kidneys present with normal ureters No urinary opening.	Bilateral small kidneys, ureter opening in cloaca . no external urinary opening
Genital System	No genetalia visible as limbs are joined	No external genetalia seen	Small male genetalia seen	Undifferentiated internal gonads

congenital malformations were also observed and noted. Pictures of each fetus's external examination as well internal examination are given below (Figures 1-4).

DISCUSSION

Sirenomelia is a congenital structural anomaly involving abnormal development of the whole of the caudal region of the body. The fusion of the lower limbs varies from fusion of thigh only, thighs, and legs to whole lower limbs.[3]

The defect was first reported in 1542 by Rocheus *et al* and later by Palfyn *et al* in 1543.[8,9]

The incidence of male-to-female is 3 to 1.[10]

The anomaly may vary clinically from mild and severe varieties. In mild form, the baby has two limbs fused into one, only up to the extent of the skin. All the three main bones of the leg are fully and correctly formed. The feet are also formed and may be only attached at the ankles. A small surgical correction in these cases is required to correct the deformity. In severe variety, both limbs are completely joined and appear ill-formed. There is absence of foot structures and out of the three long bones; two bones may be present in the entire limb.[3]

Another classification of Sirenomelia has been divided into three types, simpus apus (no feet, one tibia, and one femur), simpus unipus (one foot, two femur, two tibia, and two fibula), and simpus dipus (two feet and two fused legs (flipper like) – this is called a mermaid).[3]

In 1961, Duhamel classified the mermaid syndrome as type 5 caudal regression syndrome (CRS) for the similarity with CRS anomalies.[11]

Nowadays, mermaid syndrome is considered a separate syndrome, and the diagnostic features are the presence of single umbilical artery and renal agenesis, while in the Caudal Regression Syndrome, there is dysfunctional kidneys rather than renal agenesis.[12]

In 1987, Stocker and Heifetz noted and reported that all patients with Sirenomelia had a large umbilical artery, separated from the upper abdominal artery aorta slightly below the celiac artery, and the branches of other aorta had not evolved. This suggested that due to the lack of blood supply and inadequate nutrition, the growth of the lower part of the body was stopped and led to sacral agenesis, lower limb fusion, imperforated anus, rectal agenesis, internal and external absence of genitalia, and renal agenesis.[4]

Although the main factor for mermaid syndrome is unknown. The other important hypotheses about mermaid syndrome are vitelline artery steal hypothesis and defective blastogenesis hypothesis.

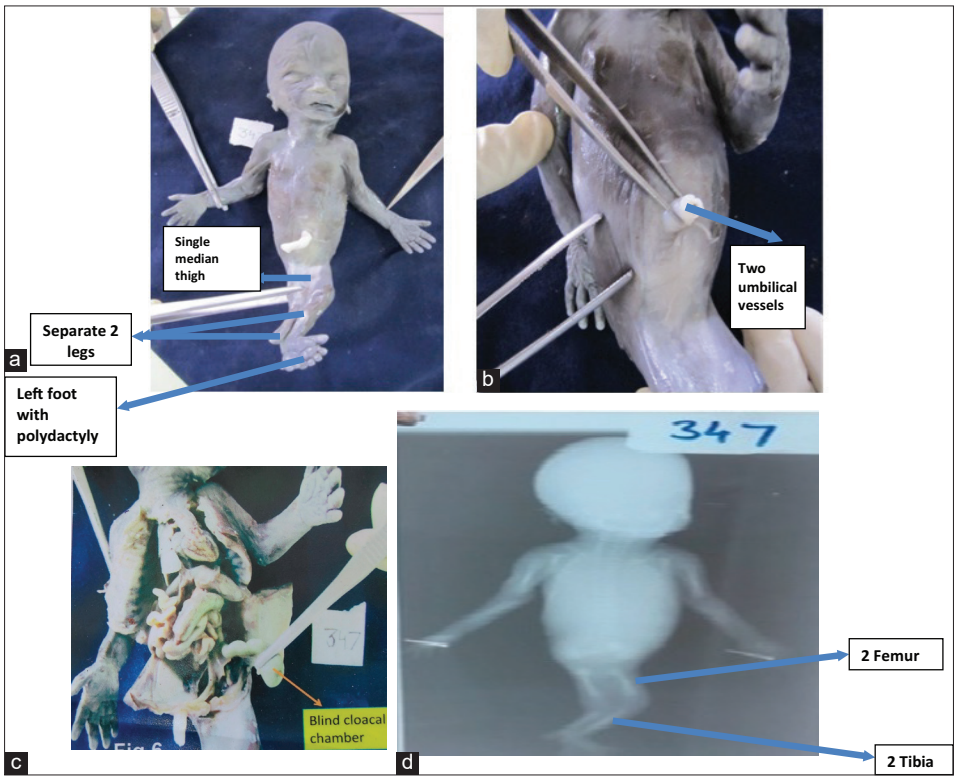

Figure 1: Case I - (a) Anterior aspect of fetus. (b) Representing single. (c) Internal examination of fetus. (d) Radiological examination of fused limb

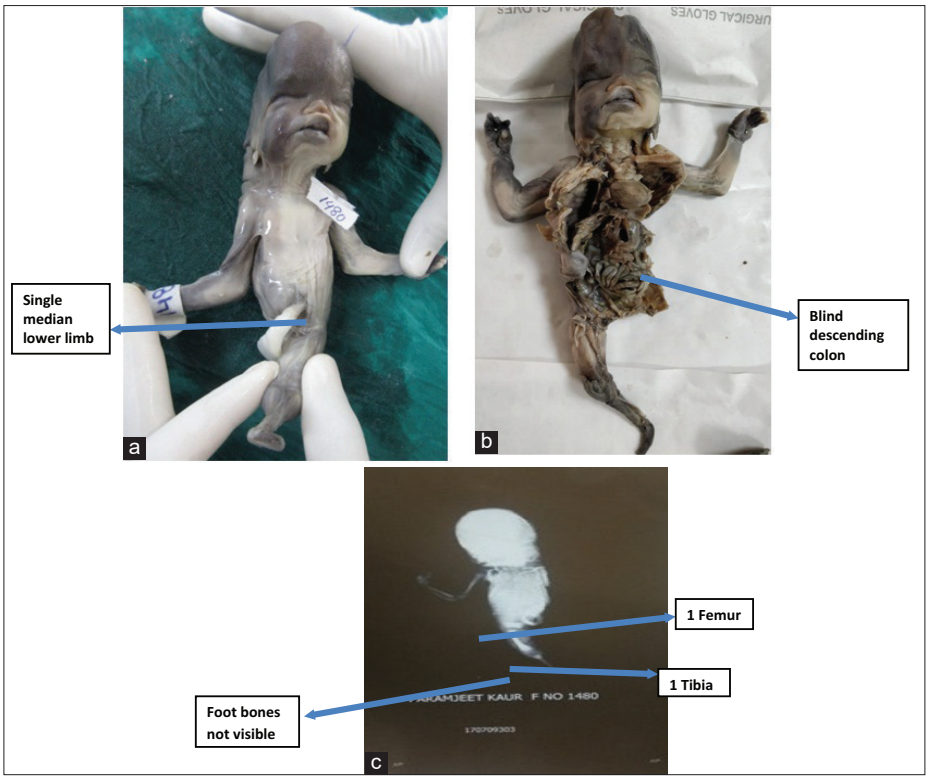

Figure 2: Case II - (a and b) External as well as internal examination of fused limbs. (c) showing X-Ray of fused limbs

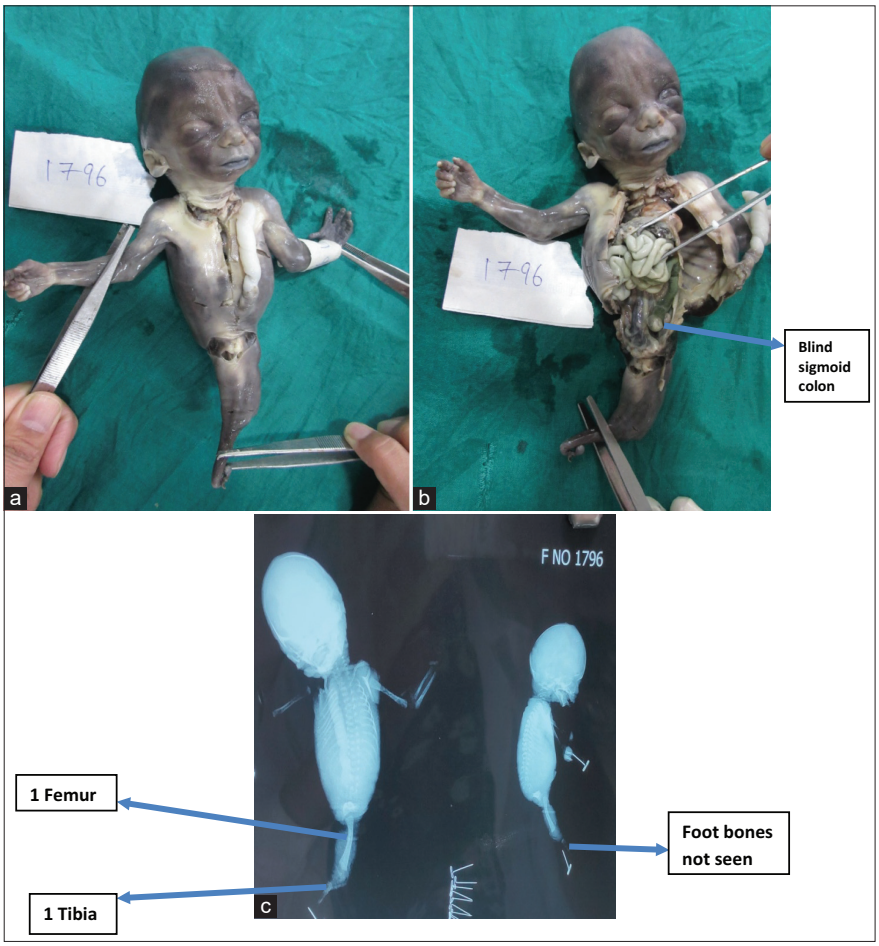

Figure 3: Case III - (a and b) Anterior aspect and internal features of fused lower limb, respectively. (c) Radiographic examination

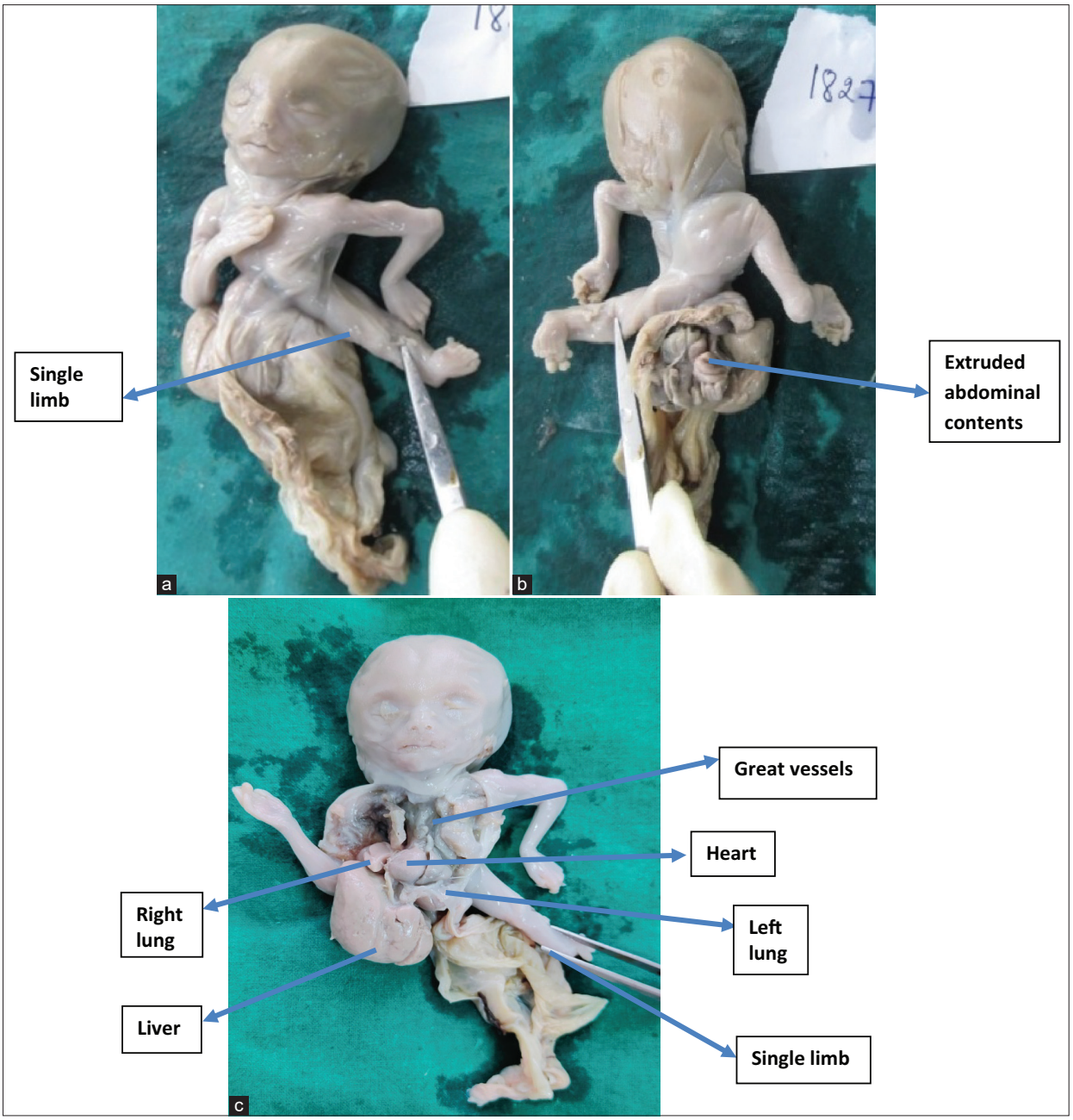

Figure 4: Case IV - (a and b) Anterior and posterior aspects of fetus. (c) Internal examination

According to the theory of vitelline artery steal hypothesis, the ventral branches of abdominal aorta other then coeliac are either absent or hypoplastic. This reduces the blood flow that feeds the caudal portion of the embryo by diverting blood flow from the embryo to the placenta. This occurs in the 3rd and 4th weeks of intra uterine life.[13]

Based on the theory of defective blastogenesis, an impaired blastogenesis, in which the lower body organs have inappropriate angiogenesis, leads to insufficient growth and incomplete development of the caudal region.[14]

Anomalies that are commonly seen with the mermaid syndrome include cleft palate, pulmonary hypoplasia, cardiac defects, omphalocele, pentalogy of Cantrell, and meningomyelocele.[15,16]

Neonates born with mermaid syndrome often have normal karyotype.[17]

The abnormalities of embryologic development, leading to the sirenomelia sequence, occur around 4th week of gestation and involve the medioposterior mesodermic axis and the caudal blastema.[18]

Most of the cases were observed in maternal age ranging from 20 to 30 years and all of them are multiparous. Fetuses were ranging from 14 to 20 weeks gestational age.

At this gestational age, the cloaca is formed; the kidneys are located in the pelvis, while the gonads are intra-abdominal. It seems logical that any damage to the caudal extremity of the embryo would affect the development of the external and internal genital organs (except the gonads, which are intra-abdominal), the terminal bowel, the bladder, the kidneys, and the pelvic bones.

CONCLUSION

In our study, we came across four fetuses with sirenomelia out of 1900 cases, which accounts for about 0.002%. In three cases one femur, one tibia was present along with foot like appendage. No foot bone was visible in X-ray. This case fall in between Simpus apus and simpus unipus. Though there are some variations in case number 1, which looked like simpus unipus because it has two femur and two tibia and one foot. This makes case fall in simpus unipus category. Associated anomalies observed were epicanthic folds, low set ears, absent external genitalia, blind sigmoid colon, and imperforate anus. The etiology is unknown in all cases, but hypothyroidism and anemia may have been a factor in the two reported cases. However, the emphasis should be both on prenatal diagnosis and genetic abnormalities to ensure an optimal management.

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