

Case Report

Caudal regression syndrome (sirenomelia) and its pathogenesis correlation: a case report

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ABSTRACT

Congenital anomaly characterized by an abnormal development of caudal region of the body with different degrees of fusion of lower extremities, bears resemblance of Sirenomelia or mermaid of Greek Mythology. This type of deformity is also known as Symmelia, Symposia, Sympus, Uromelia and Monopodia. It is associated with the single umbilical artery. It is associated with severe malformations of urogenital i.e. renal agenesis and absence of both internal and external genitalia. The aim of this study is to present, a rare congenital malformation Sirenomelia. In this context, we report a case of Sirenomelia illustrating the prenatal sonographic and pathological features.

In our study 25 years old primigravida gave birth to still born full term baby, sex could not be identified. The specimen was collected from private nursing home at Ongole, no relevant history of consanguineous marriage or tobacco smoking or drugs taking etc. Fetal USG, X-ray & CT were taken after birth. Fetal autopsy was done; the gonad, thymus and umbilical cord were sent to histopathological examination. The etiopathogenesis and characteristic features of Sirenomelia were discussed with the findings and review of the literature.

Keywords: Sirenomelia, Teratogenesis, Caudal regression syndrome (CRS), Congenital malformations, Vascular steal theory

INTRODUCTION

Sirenomelia or mermaid syndrome represents an extreme form of caudal regression. It is a rare malformation sequence characterized by fusion of the lower limbs¹ and as well as involving multiple organ abnormalities. This rare congenital malformation is incompatible with life. Some authors consider as one of the clinical manifestations of the "caudal regression syndrome" where in developmental disruption of structures occur, derived from the caudal mesodermal axis of the embryo during the primitive streak stage.²

It is regarded as a part of the caudal dysplasia sequence. It is a very rare syndrome, 0.8 to 4.2 / 100,000 births, commonly in males, with a sex ratio of 2.7:1³ The spectrum of lower limb anomalies ranges from simple fusion of soft tissues to the presence of single rudimentary limb.

Sirenomelia is not associated with chromosomal aneuploidy. The list of signs and symptoms for Sirenomelia includes the 9 symptoms:

- Fused legs.
- Absence of sacrum.
- Vertebral defects.
- Imperforate anus.
- Absence of rectum.
- Absence of external genitalia.
- Absence of internal genitalia.
- Renal agenesis.
- Absence of bladder.

In view of bad prognosis earlier intrauterine diagnosis allows less traumatic therapeutic abortion. Thorough antenatal ultrasound scanning for foetal anomalies is necessary for prompt diagnosis of this rare anomaly.

CASE REPORT

A 25 year old primigravida gave birth to still born full term baby, sex could not be identified. The specimen was collected from private nursing home at Ongole. There was no relevant history of consanguineous marriage or tobacco smoking or drugs taking. Her past medical history and family history were unremarkable. In particular, there was no history of maternal diabetes. A full-term foetus, small for gestational age and of undetermined sex was born by spontaneous vaginal delivery. The infant was still born. The infant had a flattened face and fused lower extremities with a single left lower limb. The foetus also showed absence of external genitalia.



Figure 1: Still born full term baby with fused lower limb, ventral view.

External features: Figure 1 & 2.

The Affected foetus presented with following Gross abnormal features:



Figure 2: Still born full term baby with fused lower limb – Dorsal View.

1. The eyes are amphibian-like and the ears are floppy and low set.
2. Upper extremity normal with abnormal fingers.
3. There was only one left lower limb and it was not completely formed.
4. There was Left foot and only little toe was seen.
5. Absence of external genitalia.
6. No urethra and anal openings.

Head & neck:

7. Normal hair growth.
8. External features of face normal, No cleft lip or cleft palate.

Back:

Normal –? No Spina bifida & Scoliosis is present.

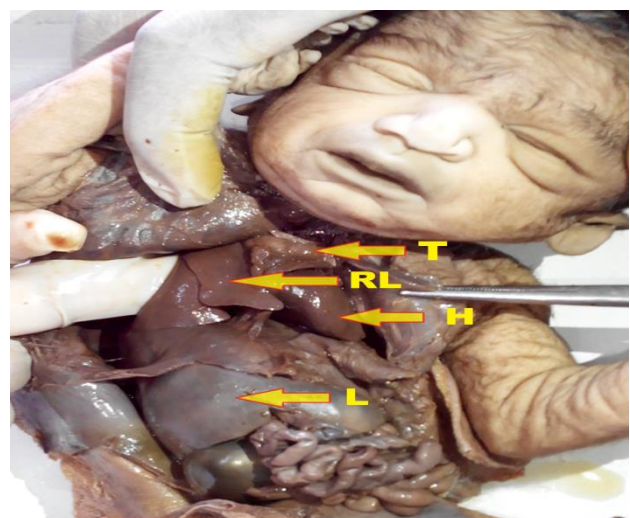


Figure 3: Opened Thorax and Abdomen showing viscera.



Figure 4: Opened Abdomen showing viscera.



Figure 7: Opened Abdomen showing Umbilical cord [U] with single Umbilical Artery and Umbilical Vein.



Figure 5: Opened Abdomen with reflected viscera showing Abdominal Aorta [AA].

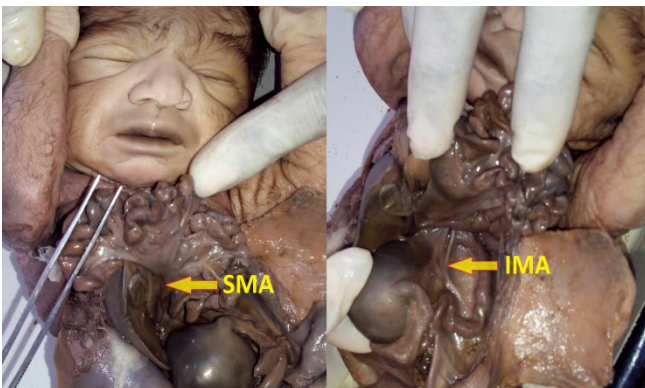


Figure 6: Opened Abdomen with reflection of viscera showing blood vessels Superior Mesenteric Artery [SMA], Inferior Mesenteric Artery [IMA].

Internal features: (Figure: 3-7)

Foetal autopsy was done and following features were observed.

1. The placenta and brain showed no remarkable findings.
2. Thorax: The heart and thymus were normal and lungs were hypoplastic (Figure-3)
3. Abdominal cavity showed multiple anomalies. The Gut rotation was complete. The Stomach, small intestine, liver and spleen were normal. The Caecum and Ascending colon distended with meconium and Rectum ended blindly.
4. There was complete renal agenesis, No kidneys, ureters and bladder.
5. Gonad (single) was seen in inguinal canal of right side (Figure-4)
6. Blood vessels of abdomen (Figure-5)

Abdominal aorta:

There was no division of Common Iliac arteries and complete absence of External & Internal iliac arteries. There was a small branch from abdominal aorta entering into the left lower limb.

Superior & Inferior mesenteric arteries were ill defined (Figure-6). A single umbilical artery was present, which was in direct continuity with the abdominal aorta. Umbilical cord showing single Umbilical artery and vein (Figure-7).

I. Investigations

X-ray findings: Figure-8

Skull normal (Figure-8)

Thorax: Shape deformed

- a. Right ribs are normal

b. Left ribs are thin and Fusion of 4, 5 and 6 ribs.

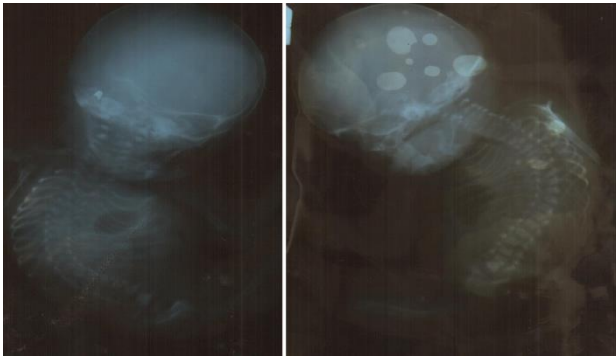


Figure 8: X – Ray showing Scoliosis [S], single Femur [F].

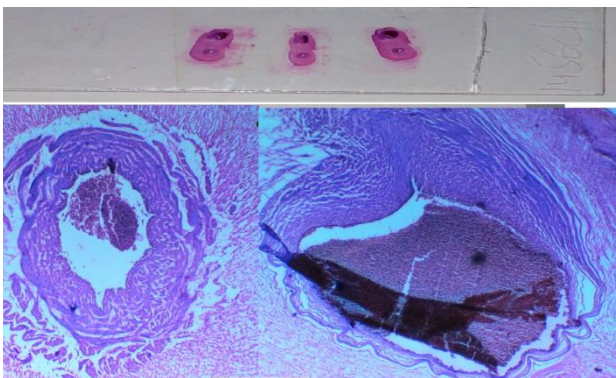


Figure 9: Microscopic Picture of Umbilical cord showing single Umbilical Artery [UA] and Umbilical Vein [UV].

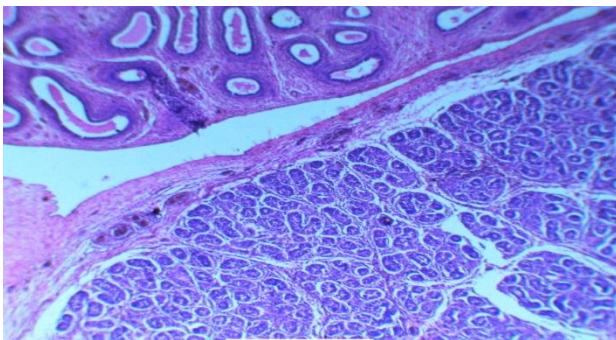


Figure 10: Gonad Microscopic Picture showing Seminiferous tubules [ST], Epididymis [E] and Retetestis [R].

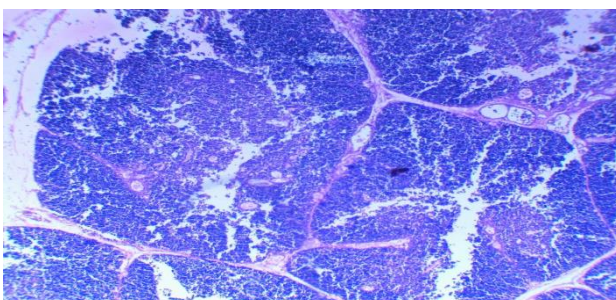


Figure 11: Thymus showing Multilobules with Hassal's corpuscles [HC].

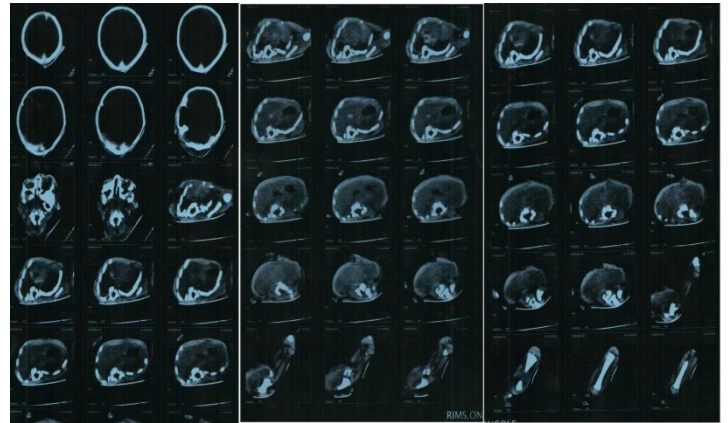


Figure 12: CT findings showing Scoliosis, single femur.

Abdomen: Stomach bubble seen.

Vertebral anomalies: Clefing seen in T3, T5 and T6

1. Scoliosis – convexity to left side +
2. Spin bifida at lumbar level.
3. Distal sacral segment not seen.
4. Partial agenesis of sacrum present.

Limb defects: Right hand: Polydactyly and Syndactyly of right thumb

Lower limbs: Single left lower limb was present, no fusion.

One femur, one tibia, no fibula, No tarsal bones, Only small (little toe) metatarsal present

Diagnosis: SIRENOMELIA

II. Histopathology:

Report of umbilicus, gonad and thymus done (Figure - 9, 10, 11).

Gonad was found to be testis, there was single umbilical artery and thymus was normal.

III. USG Abdomen

Liver and Spleen shows normal echotexture.

Right kidney absent with lying down adrenal sign.

Left kidney not adequately made out.

Urinary bladder bubble absent - ? Bilateral Renal agenesis

Well defined hypoechoic collection which was thick walled showing internal echos in lower abdomen – distended rectum secondary to imperforate anus.

Gonads were not made out. Spina bifida was seen in lumbar region.

Diagnosis - Caudal Regression Syndrome

USG Thorax

Bilateral pleural effusion with pericardial effusion.

IV. CT scan (Figure 12)

Brain: Sulci absent, parenchymal hypo density.
Hypo dense cortical sinus secondary to stasis of blood.

Thorax:

Lungs: shows multiple air spaces with fluid.
Heart: normal, minimal pericardial effusion.
Bony thorax shows abnormal contour.

Abdomen: Liver, Spleen are normal
Stomach bubble +
Kidney – not seen (agenesis)
Distended rectum with meconium
Imperforate anus
Gonads- one gonad in inguinal canal
Urinary bladder not visualized.
Spina bifida in lumbar region.
Partial agenesis of sacrum
Scoliosis with left side convexity

Limbs: One left lower limb with femur, tibia and no fibula.

Diagnosis -Caudal regression syndrome (CRS).

DISCUSSION

Sirenomelia or the Mermaid syndrome is a rare congenital dysmorphic syndrome of the lower body segments and with fusion of the lower limbs. The first reported case of mermaid syndrome was in the 16th century. It is characterized by fusion of lower limbs with sacral agenesis and other anomalies like imperforate anus, colonic atresia and rectal atresia, renal agenesis, absent bladder. Usually gonads are present and external genitalia absent. (Lutz et al., 2004).^{1,8}

In 1961 Duhamel⁴ described the spectrum of CRS from lower limb fusion to imperforate anus.

Sirenomelia may be caused by abnormalities in blastogenesis that affect distribution of blood to the caudal region of the foetus. The “Vascular steal theory” indicates that a single large artery assumes the function of umbilical arteries (Stevenson et al).²

The diversion of the blood flow from caudal structures of the embryo to the placenta, in consequence with the caudal somites do not develop normally. The steal vessel derives from the vitalline artery complex that supplies the yolk sac i.e. Superior mesenteric artery. Arteries below the level of this steal vessel are underdeveloped and the tissues arrest in some incomplete stage of development.

One umbilical artery is commonly present which leads to vascular system anomalies of the lower part to cause abnormal development⁵ The current case presents single umbilical artery and single umbilical vein.

In 1980 Gardner and Breuer⁶ proposed a theory of a neural tube over distension in the caudal area may lead to a roof plate expansion of the tube leading to lateral rotation of mesoderm by 180°. This results in fusion of the lower limb buds, closing off the midline primitive gut and urethra.

Animal experiment have shown CRS like syndrome could be induced by agents like retinoic acid, diethylpropion, lithium, sulfonamide, cadmium, lead, vitamin A deficiency, radiation, hyperthermia, organic fat solvents, cocaine etc.⁷

Stocker's more detail classification into 7 Sub types according to the fused bones.⁹

Type I: Paired femora, Tibiae and Fibulae.
Type II: Paired femora, Tibiae & single fused fibula.
Type III: Paired femora, Tibiae & absent fibula.
Type IV: Partially fused femora, tibiae & single fibula.
Type V: Partially fused femora, tibiae & absent fibulae.
Type VI: A single femur, Tibia & absent fibulae.
Type VII: A single femur, absent Tibiae and fibulae.

According to Stocker's more detailed classification, our index case belongs to type VI, single Femur and Tibia and absent Fibula. Our index case was full term still born fetus of unknown sex, Sirenomelia, had severe internal abnormalities, like bilateral renal agenesis, associated severe pulmonary hypoplasia and severe GIT anomalies. It was also associated with multiple skeleton abnormalities with single limb deformity (left limb) with single femur, single tibia & no fibula. Therefore was incompatible with life.

CONCLUSION

The infant (Male) described in our case report has clinical and anatomical features consistent with Sirenomelia/Monopodia / caudal regression syndrome. It is fatal because of hypoplasia lung, bilateral renal agenesis and associated absence of urinary bladder and imperforate anus. There is currently no known Serum marker that may be used for antenatal diagnosis of this type of anomaly. Prenatal ultrasonography is used to diagnose Sirenomelia. Convergent femoral bones that lie in a side-by-side configuration without change over a time suggest the possibility of lower limb fusion. In view of the bad prognosis earlier intra-uterine diagnosis allows less traumatic therapeutic abortion.

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Ethical approval: Not required

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