



## Sirenomelia - A Rare Case Report

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### ABSTRACT

Caudal regression syndrome is a rare congenital malformation described by various degrees of developmental failure. Its most extreme and rare form is known as sirenomelia or mermaid syndrome. The associated malformations include anorectal, vertebral, urological, genital, and lower limb anomalies. It invariably presents with lower limb fusion, sacral and pelvic bony anomalies, absent external genitalia, imperforate anus, and renal agenesis or dysgenesis. Here, a case of sirenomelia in a stillborn baby is being reported that was born by normal vaginal delivery at 34 weeks of pregnancy following an uneventful pregnancy. Physical examination at birth showed normal facies with fusion of the lower limbs and anomalies involving GIT & genitor-urinary tract. The study also included autopsy findings, radiological study & histology of different viscera to determine the aetiopathogenesis.

**Keywords:** Caudal regression syndrome, sirenomelia, malformations, aetiopathogenesis.

### INTRODUCTION

Sirenomelia is a condition was partial or complete fusion of lower limbs associated with VACTERL anomalies (Vertebral anomalies, Anal atresia, Cardiac defects, Tracheo-esophageal fistula, Esophageal atresia, Renal and Radial anomalies, Limb defects) is present. Sirenomelia or Mermaid syndrome was initially stated by Rocheas in 1542 and Palfya in 1553 and the name was given after the mythical Greek Siren.<sup>1-3</sup> Incidence being 1 to 4 in 100,000 with male: female ratio of 3:1.

It shows severe malformations of the gastrointestinal, genitourinary, cardiovascular and musculoskeletal system. However, oligohydromnios secondary to severe renal dysplasia is universal. Duhamel defined all the anomalies of mermaid syndrome in 1961 and described it as the most severe form of caudal regression syndrome.<sup>4-6</sup>

### Case Report

A case of intra uterine death from Obstetrics & Gynaecology department of Great Eastern Medical College, Srikakulam, Andhra Pradesh, presenting the features of sirenomelia was studied. The baby was born to non-consanguineous parents at 34 weeks of gestation having birth weight of 2.8 kg. There was no history of exposure to teratogenic agent during the antenatal period. Mother was non-diabetic, non hypertensive.

The external features of this case of sirenomelia showed the normal appearance above the level of umbilicus (Figure 1). All the abnormalities were seen below the umbilicus. Umbilical cord was short and had a single umbilical artery (Figure 2). There was a single fused lower

tapering extremity (Figure 3) with absence of external genitalia. There was a meningocele at sacral region.

Anal orifice was absent.

### Autopsy Finding

- Heart, lungs and esophagus were normal. Pericardial sac was large. (Figure 4)
- G.I.T. ended blindly at the level of the transverse colon.
- Adrenal gland was large in size, kidneys were hypo plastic.
- The ureter ended in the solid mass of tissue which could not be differentiated into a particular structure.

### X-Ray Findings

- X-ray revealed deformed pelvic and sacral region with single femur and tibia. (Figure 5)

### Histological

- Umbilical cord showed single umbilical artery and vein. (Figure 6)
- Villi were hypo plastic, with lot of fibrous tissue deposition. (Figure 7)
- In the placenta infarcted and hemorrhagic areas were seen. (Figure 8)
- Large fetal cortex of the supra renal was seen. (Figure 9)
- Hypo plastic kidney was observed. (Figure 10)
- Undifferentiated gonadal tissue was seen. (Figure 11)





**Figure 1:** Normal appearance above the level of umbilicus



**Figure 2:** Short umbilical cord



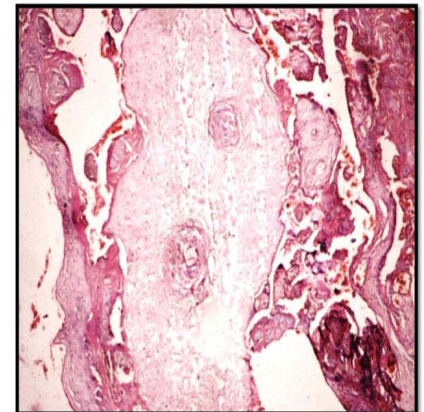
**Figure 3:** Single fused lower tapering extremity



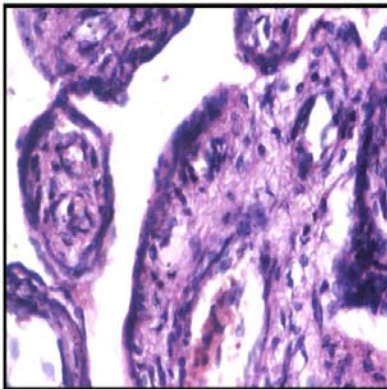
**Figure 4:** Heart, lungs and esophagus were normal with large pericardial sac



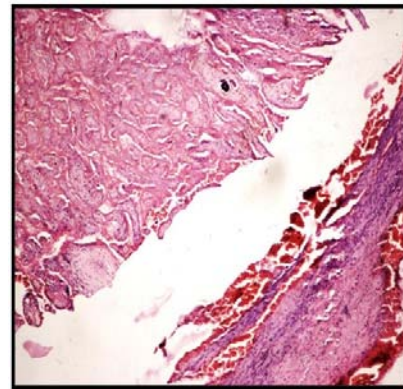
**Figure 5:** Deformed pelvic and sacral region with single femur and tibia



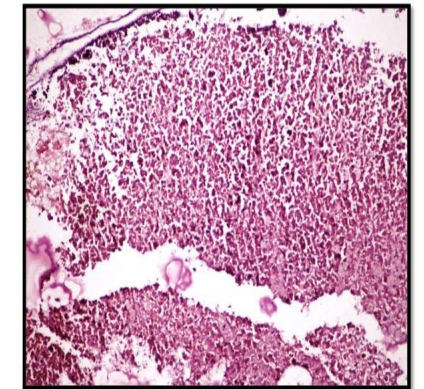
**Figure 6:** Umbilical cord showed single umbilical artery and vein



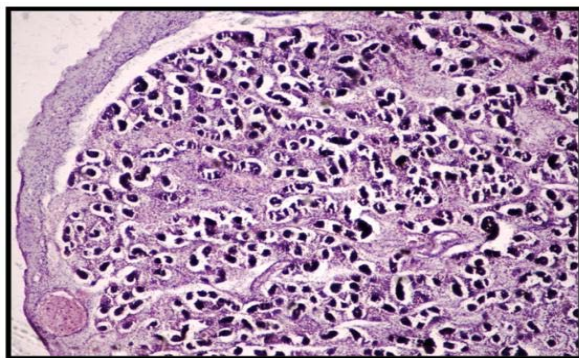
**Figure 7:** Hypoplastic villi with lot of fibrous tissue deposition



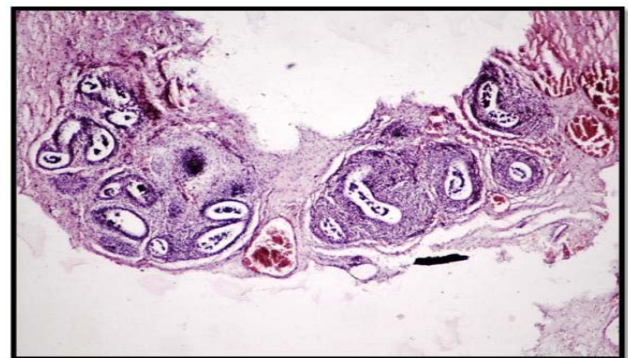
**Figure 8:** Infarcted and hemorrhagic areas in placenta



**Figure 9:** Supra renal showing large cortex



**Figure 10:** Hypoplastic kidney



**Figure 11:** Undifferentiated gonadal tissue



## DISCUSSION

Sirenomelia is a rare and lethal congenital anomaly with unknown etiology. Most of these newborns were still born or die immediately after birth. Most common cause of death is usually renal agenesis, which is incompatible with life. It involves abnormal development of the caudal region of the body resulting in varying degrees of fusion of the lower limbs. However, other visceral defects such as hypoplastic lungs, cardiac agenesis, absent genitalia, digestive defects, absent kidney and bladder, vertebral and central nervous system defect are also reported.<sup>7,8</sup> Duhamel stated that Sirenomelia and anorectal malformations represent the two extremes of a single comprehensive syndrome which arises from an embryonal defect in the formation of the caudal region. He called it the Syndrome of Caudal Regression.

There is a strong association between this syndrome and maternal diabetes; up to 22 % of foetuses with this anomaly are known to have diabetic mothers. But maternal DM is not present in this case. Several mechanisms have been proposed to explain sirenomelia and they included deficiencies in caudal mesoderm and trophic defects due to a deficient blood supply to the distal region.

### Classification

Stocker and Heifetz<sup>9</sup> classified the sirenomelia sequence into 7 types as below:

1. All thigh and leg bones are present
2. Single fibula
3. Absent fibula
4. Partially fused femurs fused fibulae
5. Partially fused femurs
6. Single femur single tibia
7. Single femur absent tibia

Our case is found to be **TYPE SIX**.

### Risk factors

About the risk factors, many different kinds of theories have been suggested such as maternal diabetes.<sup>10-13</sup> However, in this case, the mother was not known to be diabetic. Genetic and environmental factors also play a role.<sup>11,14</sup> Hibelink pointed out that teratogenic agents like cadmium element and lead may result in sirenomelia.<sup>15</sup>

Some studies showed that there is potential teratogenic effect of vitamin A<sup>16</sup>, cocaine<sup>17</sup> & irradiation exposure.<sup>18</sup> Sirenomelia is also associated with different new reproductive technologies, like ICSI (Intra Cytoplasmic Sperm Injection).<sup>19</sup>

Also, it is seen in twins. Studies indicate that incidence in monozygotic twins relative to dizygotic twins or singletons is 100-150 times higher.<sup>20</sup> But, in this case, no twin gestation or family history of twinning was found

out. Still, genetic counselling is suggested as the risk of recurrence is fairly 3– 5%.<sup>21</sup>

### Aetiopathogenesis

Five pathogenetic theories of about the causation of sirenomelia are described:

#### *An Embryological Insult*<sup>22</sup>

The sequence of events resulting in sirenomelia (sirenomelia sequence) starts from an 'embryological insult' involving the caudal mesoderm begins between 28-32 days of foetal life.<sup>22</sup> By this time, the cloaca is already formed, the kidneys are found in the pelvis while the gonads are intra-abdominal.<sup>21</sup> Hence, any developmental abnormalities of the caudal extremity affect equally the kidneys, the bladder, the terminal bowels, the pelvic bones as well as the genitalia.<sup>21</sup> In this sequence, there is renal agenesis, absent genital organs, anal imperforation, absent rectum and dysgenesis/agenesis of the sacrum.

#### *Vascular Steal Theory*<sup>14</sup>

Stevenson proposed the vascular steal theory which explains the development of abnormalities on the caudal extremity. It suggests that the shunting of blood via an abnormal abdominal artery which arises from high up in the aorta towards the placenta. This results in hypoplasia of the vasculature distal to the artery leading to nutritional deficiency of the caudal half of the body.<sup>18</sup> Hence there may be complete/incomplete agenesis of the caudal structures described above, but the gonads are spared as they are intra-abdominal. The single umbilical artery in this case favours the theory.

#### *As part of the Caudal Regression Syndrome (CRS)*<sup>6</sup>

Another study describes sirenomelia as part of the CRS which is a rare congenital defect characterized by a broad spectrum of lumbosacral agenesis. This syndrome was described by Duhamel & it includes genitourinary and vertebral anomalies mainly characterized by sacrum dysgenesis, altered spinal cord, urinary incontinence of variable intensity and misplaced lower limbs. Renal dysgenesis and imperforate anus is an inconstant feature. Some authors consider sirenomelia to be the most extreme form of this relentless condition.

#### *As part of the VACTERL syndrome (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies and limb abnormalities)*<sup>20</sup>

Another theory describes sirenomelia as part of the VACTERL syndrome. There is a major overlap in the phenotypic manifestations of sirenomelia and VACTERL.<sup>20</sup> In most cases, the distinction between sirenomelia sequence and VACTERL is based on the severity of the component defects. A case of sirenomelia with single lower limb can be regarded as an indicator of other severe malformations. In this case, autopsy reports tell that it is a part of VACTERL as here we can see presence of anal atresia, renal anomalies and limb abnormalities.



**External forces acting on the caudal extremity<sup>21</sup>**

This theory suggests that external forces acting on the caudal extremity of the embryo causes its hypoplasia.<sup>21</sup> This theory was supported by Gardner<sup>23</sup>, who suggested that excessive rotation of the neural tube at its caudal end provoked a lateral rotation of the mesoderm causing fusion of the lower limbs, and closure of the primitive bowel and urethra.

**CONCLUSION**

Sirenomelia is a rare but peculiar syndrome. Its antenatal diagnosis can be made by antenatal ultrasound. Controversies on its etiopathogenesis persist even though it is increasingly believed to be distinct from the caudal regression.

It is associated with many other visceral anomalies which are usually incompatible with life. However surviving sirenomelic fetuses have been described with costly conservative management.

All cases have been sporadic. More recent theory suggests a vascular pathogenesis resulting from a vitelline arterial steal, resulting in diversion of blood flow from caudal structures. Knowledge of this rare syndrome is important to dissipate cultural myths and free the family from stigmatization.

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