

## **Prenatal Evaluation of Abdominal Vasculature of Sirenomelia with 3D Volume Rendering Ultrasound (HDlive Flow)**

**Aki Hayashi\*, Mikio Morine, Ayaka Tachibana, Akane Kondo, Kenji Hinokio and Kazuhisa Maeda**

*General Perinatal Medical Center, Shikoku Medical Center for Children and Adults, Kagawa, Japan*

**\*Corresponding Author:** Aki Hayashi, General Perinatal Medical Center, Shikoku Medical Center for Children and Adults, Kagawa, Japan.

**Received:** November 29, 2021; **Published:** December 27, 2021

### **Abstract**

Sirenomelia is a very rare congenital anomaly. We experienced a case of second trimester sirenomelia referred to our institution due to oligohydramnios at 18 weeks of gestation. Since fusion of lower limbs is the important findings for making a diagnosis, detailed skeletal survey of the fetal lower extremities was done by 3D-CT. In addition, sirenomelia presents variable visceral abnormalities in urogenital and gastrointestinal structure, so we focused on vasculature from descending Aorta. This is essential point to estimate other organ's development. In this case, we clarify abdominal vasculature by HDlive Flow (3D volume rendering ultrasound). This rendering mode was useful for the prenatal evaluation of complicated vasculature and blood flow in sirenomelia.

**Keywords:** *Sirenomelia; HDlive Flow; 3D-CT; Vascular Steal Theory; Vitelline Artery*

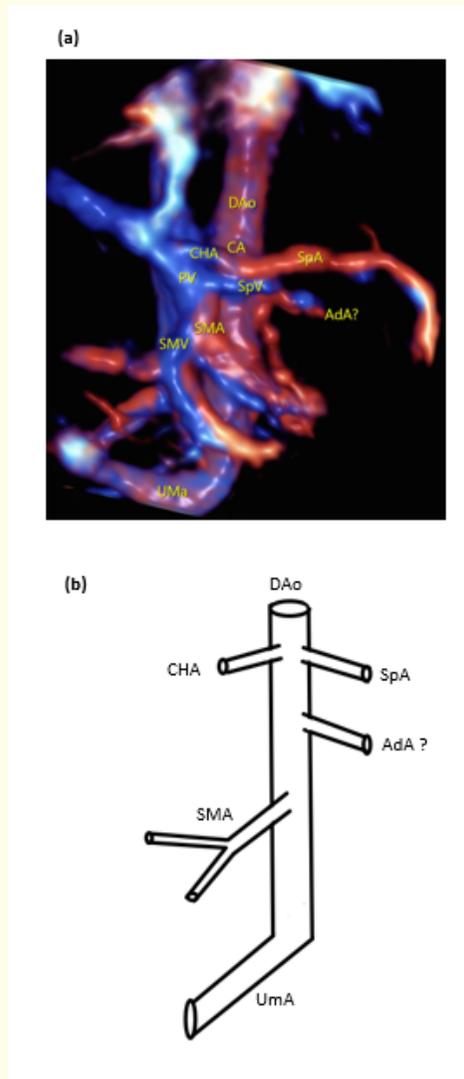
### **Introduction**

Sirenomelia, also known as mermaid syndrome, is a very rare congenital anomaly and occurs between 1.1 and 4.2 per 100,000 births [1]. This malformation is characterized by severe visceral abnormalities, most commonly urogenital and gastrointestinal, fusion of both lower limbs [1,2]. Sirenomelia is often accompanied by renal aplasia or hypoplasia, and oligohydramnios develops after the second trimester of pregnancy, resulting in a poor prognosis. Until now, the etiology and pathogenesis for sirenomelia is undetermined. However, the vascular steal theory with the vitelline steal artery diverting blood supply and nutrients from the lower body and limbs has been proposed as a hypothesis [1,3,4]. HDlive Flow is an appropriate application for assessing vascular blood flow because it reconstructs spatial three-dimensional (3D) images of fetal and placental blood vessels. Here, we investigated the usefulness of the evaluation of the abdominal vasculature by HDlive Flow and embryological considerations in sirenomelia.

### **Case Reports**

Our patient was a 31-year-old multiparous Japanese woman. Her medical and family histories were no problem. She had no previous history of smoking, habitual alcohol drinking, drug abuse, or radiation exposure. She was referred and admitted to our hospital at 18+1 weeks of gestation because of oligohydramnios. Ultrasound examination (Voluson E10; GE Healthcare Japan, Tokyo, Japan) at admission revealed severe oligohydramnios and a single umbilical artery (SUA). The kidneys, bladder and unilateral femur were not visualized. From the above findings, a diagnosis of sirenomelia was suggested. Three-dimensional computed tomography (3D-CT) showed only one femur and hypoplastic tibia. We confirmed that the celiac artery, superior mesenteric artery and umbilical artery were branched from the descending artery by HDlive Flow at 19+6 weeks. However, the inferior mesenteric artery, renal artery and iliac artery could not be visualized. A branch of the descending aorta was found between the celiac artery and the superior mesenteric artery, suggesting an adrenal artery [Figure 1a/b]. We informed the parents that the fetal findings and the suspected very poor prognosis. They then chose a termina-

tion of pregnancy, which was performed at 20+2 weeks. On the postnatal examination, the baby weight 290g and the sex was unknown. The external examination showed the fusion of both lower extremities, the bell-shaped thorax and imperforate anus [Figure 2]. Postmortem autopsy imaging (Ai) revealed the lower limb was aped leg, containing one femur and one hypoplastic tibia [Figure 3]. The bilateral kidneys and bladder were aplastic. Pathological autopsy suggested the blind end more than 15cm from the ileocecal region, the bilateral kidneys and bladder were agenesis and imperforate anus was observed. The chromosomal analysis revealed a 46, XY normal karyotype.



**Figure 1:** HDlive Flow (a) and schema (b) of the abdominal vasculature at 19+6 weeks of gestation. The celiac artery, superior mesenteric artery and umbilical artery diverged from the descending artery.

DAo: Descending Aorta, CA: Celiac Artery, SpA: Splenic Artery, SpV: Splenic Vein, AdA: Adrenal Artery, PV: Portal Vein, SMA: Superior Mesenteric Artery, SMV: Superior Mesenteric Vein, UmA: Umbilical Artery, CHA: Common Hepatic Artery.



**Figure 2:** Macroscopic image of the delivered baby. A single lower limb was noted.



**Figure 3:** 3D-CT image (VR (volume rendering) of the delivered baby. A single femur and hypoplastic tibia were revealed.

## Discussion

The diagnosis of sirenomelia is based the presence of fusion of the lower extremities, but detailed evaluation by ultrasound observation is usually limited owing to the sever oligohydramnios from renal agenesis [5]. Since 3D-CT can visualize fetal bone three dimension-

ally, the fetal lower limbs can be accurately evaluated even in the severe oligohydramnios. Furthermore, we suggest that the evaluation of the abdominal vasculature by HDlive Flow is useful for understanding of sirenomelia from an embryological point of view. Although the exact pathogenesis and etiology of sirenomelia remains unclear, the general theory is abnormal persistence of the vitelline artery, leading to vascular steal from the abdominal aorta and lower extremity [1,3]. This case, as with most sirenomelia cases, had a single umbilical artery (SUA). The SUA branched from a very high position on the abdominal aorta. And below the point, the abdominal aorta was highly hypoplastic and had few branches [Figure 1a]. These facts supported the vascular steal theory that the SUA diverts the normal blood flow to the placenta leaving the lower part of the body with a severely deficient circulation incompatible with normal development.

HDlive Flow is an appropriate application for assessing vascular blood flow because it can show spatial 3D images of fetal and placental blood vessels. Use of HDlive Flow to visualize the characteristic abdominal vascular pattern of a fetal sirenomelia has not been previously reported, but it may be very useful for the prenatal diagnosis. In our case, HDlive Flow provided spatial 3D images of the blood vessels branching from the descending aorta, and we were able to predict the presence of nourished organs. Spatial relationships of the vasculature, such as the descending aorta, celiac artery, superior mesenteric artery and umbilical artery were also visualized, and we were able to identify the level of the regressed aorta [Figure 1a/b]. As a result, we could estimate that the organs such as the kidneys and lower gastrointestinal tract, which normally should be supplied by the lower branches of the aorta, having various degrees of hypoplasia due to lack of blood flow and nutrients. And these findings were supported by the facts of pathological autopsy.

### Conclusion

Detailed evaluation of the fetal lower extremities by 3D-CT and the abdominal vasculature by HDlive Flow is considered to be useful for the prenatal diagnosis of sirenomelia and important step towards the understanding of its pathogenesis from an embryological point of view.

### Acknowledgements

None.

### Conflict of Interest

There is no conflict of interest.

### Bibliography

1. Garrido-Allepuz C., *et al.* "A clinical and experimental overview of sirenomelia: insight into the mechanisms of congenital limb malformations". *Disease Models and Mechanisms* 4 (2011): 289-299.
2. Kavunga EK., *et al.* "Sirenomelia associated with an anterior abdominal wall defect: a case report". *Journal of Medical Case Reports* 13 (2019): 213.
3. Stevenson RE., *et al.* "Vascular steal: the pathogenetic mechanism producing sirenomelia and associated defects of the viscera and soft tissues". *Pediatrics* 78 (1986): 451-457.
4. Martinez-Frias M., *et al.* "Does single umbilical artery (SUA) predict any type of congenital defect? Clinical epidemiological analysis of a large consecutive series of malformed infants". *American Journal of Medical Genetics* 146A (2008): 15-25.
5. Langer B., *et al.* "Sirenomelia and situs inversus: case report and review of the literature". *Fetal Diagnosis and Therapy* 11 (1996): 79-84.

**Volume 11 Issue 1 January 2022**

©All rights reserved by Aki Hayashi., *et al.*