

# A prenatally diagnosed case of sirenomelia with dextrocardia and omphalocele

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## Summary

**Sirenomelia (Mermaid syndrome) is a rare anomaly of caudal region of the body, presented with fusion of the lower limbs. Genito-urinary, gastro-intestinal, neural tube and vertebral anomalies are found in most cases. We present a case of sirenomelia diagnosed in the first trimester, associated with dextrocardia, and omphalocele.**

## Introduction

Sirenomelia, alternatively known as "mermaid syndrome" is a very rare and usually lethal sequence of congenital anomalies, involving the caudal region of the body, in which the fusion of the lower limbs, gives the appearance of a mermaid's tail. It was firstly described by Rocheus in 1542 and Palfyn in 1553 and named after the mythical Greek sirens (1).

The sequence of sirenomelia is characterized by a variable degree of atrophy or hypotrophy of the lower limbs with secondary merging and rotation. Genito-urinary, gastro-intestinal, neural tube and vertebral anomalies are found in most cases, among them agenesis or dysgenesis of the kidneys, absent external genitalia, anus imperforatus or blind ending colon, lumbosacral and pelvic bone abnormalities and a single umbilical artery (2-6).

Sirenomelia has an incidence ranging between 1/24,000 and 1/67,000 births (7), with a range of 0.1 to 1% of all malformed infants. It occurs 100-150 times more frequently in monozygotic than in dizygotic twins or singletons (8). Males are three times more often affected than females (9).

Prenatal diagnosis of sirenomelia has been made by ultrasound mostly during the first trimester or early second trimester, in which the amniotic fluid volume is usually normal, unrelated to the fetal urine production. Conversely in the second trimester, oligohydramnios due to renal agenesis makes the diagnosis of sirenomelia more difficult.

In this report, we present a case of sirenomelia associated with dextrocardia and omphalocele diagnosed on a first trimester ultrasound at 11 weeks' gestation and 2 days.

## Case

A 31-year-old primigravida, with unremarkable medical and family history and no history of drugs or substance abuse, underwent a sonography at 11 weeks' gestation and 2 days according to the last menstrual period. On transvaginal ultrasound, a living singleton fetus with a crown-rump length of 46 mm (corresponding to the gestational age) with a normal head, trunk and upper extremities could be visualized. However, only one lower extremity on the median could be identified. Color Doppler imaging showed a single umbilical cord artery. The amniotic fluid volume was normal. Dextrocardia and omphalocele were identified. Presence of lower extremity fusion and single pelvic bone were further supported by transabdominal three-dimensional (3D) ultrasound imaging. After the parents were counseled about the poor prognosis of the syndrome, they opted for termination of pregnancy (Figs. 1, 2 and 3).



Figure 1



Figure 2



Figure 3

## Discussion

Caudal regression syndrome is a deficit in the formation of the caudal region before the fourth week of gestation and consists of a spectrum of anomalies that range from ectopic anus to sirenomelia.

Unlike caudal regression syndrome, sirenomelia seems to be less likely associated with gestational diabetes and commonly has severe renal tract abnormalities and fusion of the lower extremities, which is different from caudal regression syndrome, in which there is just hypoplasia of the lower limbs (10).

The etiology of sirenomelia is mostly unknown. Several theories have been proposed, but the hypotheses of vascular disruption or caudal embryo damage between 28 to 32 days' gestation are felt to be responsible for most of these malformations (11, 12).

An early antenatal diagnosis of sirenomelia may be su-

spected in the presence of lower fetal extremity fusion, oligohydramnios, bilateral renal agenesis, and a single umbilical artery.

In the second half of pregnancy, the prenatal diagnosis of sirenomelia is usually hampered by oligohydramnios or anhydramnios secondary to renal agenesis or dysgenesis.

This case report illustrates that sirenomelia can be reliably detected at routine first trimester ultrasound and early diagnosis gives the parents the option of early pregnancy termination.

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