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Case Report

Relationship of Sirenomelia with Gestational Diabetes Mellitus and Hypothyroidism

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ABSTRACT

Sirenomelia is a rare congenital structural anomaly characterized by developmental disorders of the caudal region. Abnormalities observed in sirenomelia constitute the most severe form of caudal regression syndrome (CRS). It can be diagnosed by antenatal ultrasonography. The present study aimed to discuss a case with sirenomelia born from a mother with a history of gestational diabetes mellitus (GDM) and hypothyroidism. The present study is the first case report in the literature in which maternal hypothyroidism and sirenomelia were found simultaneously.

Keywords: sirenomelia, mermaid syndrome, caudal regression, hypothyroidism

INTRODUCTION

Sirenomelia is a rare congenital anomaly characterized by varying degrees of fusion of the lower extremities and genitourinary and thoracolumbar malformations and atresia of the anorectal region [1]. Also known as "Mermaid Syndrome", the incidence of sirenomelia is 0.8-1/100,000 and it is more common in baby boys [2]. Sirenomelia is the most severe form of caudal regression syndrome (CRS) and has a strong association with maternal diabetes [3, 4]. It can be diagnosed by ultrasonography during antenatal period. In these cases, mostly stillbirth or a very short life span is observed [5, 6, 7].

CASE

At the 36th week of the fourth pregnancy (G4P4Y3) of a 29-year-old woman who had no consanguineous marriage, substance addiction or fetal malformation history, it was detected in the ultrasonographic examination performed during routine controls that the lower extremities of the fetus were fused. She was screened for gestational diabetes mellitus (GDM) and hypothyroidism. After the diagnosis of GDM by Oral Glucose Tolerance Test performed in the first trimester, she was followed up. HbA1c measured in the third trimester was found to be 5.49%. Serum thyroid stimulating hormone and free T4 levels in the second trimester were found to be 33.9 IU/ml and 0.693 ng/dl, respectively. With these findings, she was diagnosed to have hypothyroidism and levothyroxine sodium was started for treatment (125 mg- 2 months).

During intrauterine follow-up, pulmonary hypoplasia, genitourinary and musculoskeletal system malformations and intrauterine growth retardation were detected in the

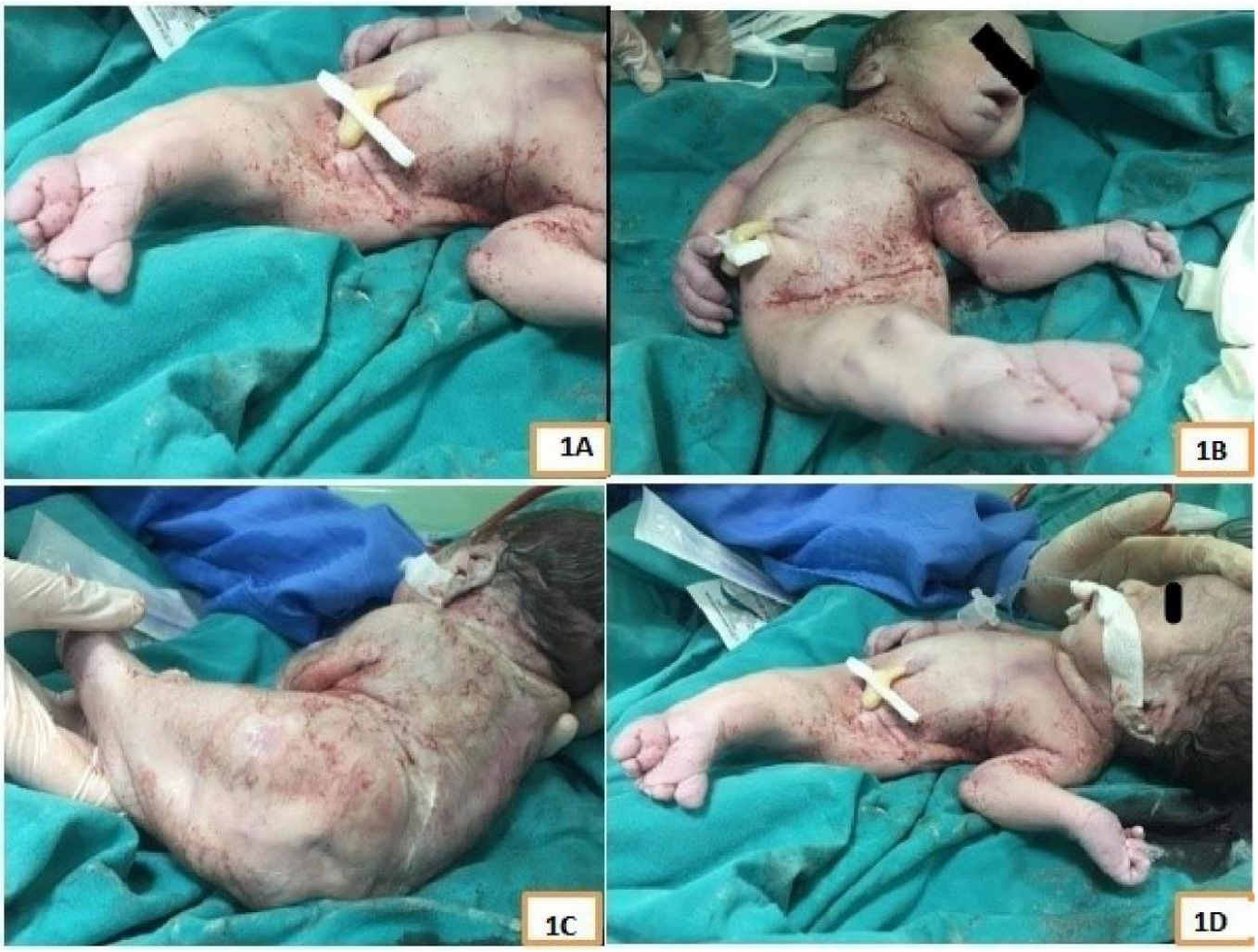

Figure 1. Postpartum images of the sirenomelia case

fetus. Due to anhydramniosis, premature rupture of membranes and breech presentation, cesarean was performed at the 33rd gestational week. The baby's birth weight was 1450 grams. The baby whose skin was cyanotic had a maximum heart rate less than 60 bpm. Therefore, resuscitation and intubation procedures were applied to the baby.

On physical examination of the case, Potter's phenotype, a typical facial appearance characterized by flattened nasal root, parrot beak nose, retrognathia, hypertelorism, prominent epicanthal folds and low-set ears, was detected. There was a single artery in the umbilical cord. The lower extremities were fused. It was observed that there were no urinary meatus, vaginal introitus or anus (**Figures 1A-1D**). The baby, who had respiratory distress since birth, died in the third hour of his/her life despite respiratory support. Present findings in the baby suggested the diagnosis of sirenomelia. Written consent was obtained from the parents for the present case report.

DISCUSSION

The etiopathogenesis of sirenomelia has not been fully elucidated yet, but several hypotheses have been proposed regarding this. While improper mesodermal cell migration has been emphasized in one of these hypotheses, another more common hypothesis claims that the tissues and organs in the lower part of the body have insufficient perfusion in the embryonic period due to the presence of a single artery in the umbilical cord [8-10]. In our case, there was only one umbilical artery in accordance with the common hypothesis. Diabetes mellitus, gestational age less than 20 or greater than 40, teratogen exposure during pregnancy are maternal risk factors, and being an identical twin and male gender are fetal risk factors for the development of sirenomelia [5,6,10].

Fusion in the lower extremities at various grades, the presence of a single umbilical artery, anorectal atresia and genitourinary system malformations are the main findings of sirenomelia. In addition, congenital heart and abdominal wall defects and Potter's phenotype, characterized by parrot beak nose, prominent infraorbital and nasolabial folds and

low-set ears are among the other accompanying findings [8,11,12]. Consistent with these findings, our case had Potter's phenotype, characterized by flattened nasal root, parrot beak nose, retrognathia, hypertelorism, prominent epicanthal folds and low-set ears.

Fusion in the lower extremities is the most striking feature in sirenomelia and legs may be fused with bony structures or only at the skin level [6,10]. Various grades of fusion in the lower extremities, called sympusdipus, sympusmonopus and sympusapus, may be observed. Foot-like structure is not developed in a sympusapus case and there is only one tibia and one femur. A symposmonopus case has only one foot-like structure and two femora, two tibiae and two fibulae. In the third group called sympusdipus, there are two foots and the V-shaped fused legs [11,13,14]. The legs of our case could be classified as sympusdipus in this categorization. Other commonly reported findings include intestinal obstruction and ambiguous genitalia [5,8,11,13,14]. In our case, anal atresia, ambiguous genitalia and the presence of a single artery in the umbilical cord were observed.

The degree of developmental anomalies of kidneys determines the life span of sirenomelia cases [11]. Few surviving sirenomelia cases have been reported in the literature and their kidneys were functional [6,7]. Recent studies have reported that a small number of cases with functional kidneys who underwent reconstructive pelvic and limb surgeries survived and their neurological development was normal [11]. Our case had renal anomalies and died in the third hour of his/her life. Sirenomelia is generally incompatible with life. Therefore, early diagnosis by ultrasonographic examination is important in terms of making a possible decision about termination of pregnancy.

Sirenomelia is thought to be the most severe form of CRS. Both syndromes are associated with maternal diabetes mellitus. It has been reported that the relationship of sirenomelia with maternal diabetes mellitus is lower than CRS (2% vs 22%) [3,4]. It is thought that free oxygen radicals that increase in maternal diabetes mellitus have teratogenic effects. The mother of our case was diagnosed with GDM in the first trimester.

Previous studies reported that GDM was the only maternal disease associated with sirenomelia that can be detected [15-17]. There was no known risk factor, except for GDM in our case. The mother of our case also had hypothyroidism. Therefore, the present study is the first case report in the literature in which sirenomelia and maternal hypothyroidism were found simultaneously. It is known that maternal hypothyroidism is associated with various congenital disorders. Based on this information, it should be taken into account that there may be a relationship between sirenomelia and maternal hypothyroidism.

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