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Sirenomelia/mermaid syndrome without imperforate anus in a premature infant

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1. Introduction

Sirenomelia/mermaid syndrome, which was first described in 1542 and resembled to mermaid because of its characteristic phenotypic appearance, is a very rare congenital anomaly [1]. This disease characterized by the fusion of the lower extremities at different grades, can be accompanied by multiple system anomalies such as musculoskeletal, gastrointestinal system (GIS), genitourinary system (GUS) and cardiovascular system (CVS). The disease is seen in 1/600,000–1000000 rate [1,2]. In this article, we aimed to present a premature (26 weeks) patient with sirenomelia with very low birth weight (780 g) who could survive postnatal 8 h and had a normal anus, unlike expected in the anomaly.

2. Case report

The patient was born from the second pregnancy of 24-year-old mother as the second-vital and 780 g with a 26th week cesarean. When the patient was born, the 1–5, and 10th minute apgar scores were 3–5 and 6, respectively. The patient was intubated immediately after delivery. Mother had no prenatal follow up. There was only one lower extremity on physical examination (Fig. 1). On the lower extremity, there were two femurs, two tibia and one fibula, one foot and four toes. Both upper extremities were observed naturally. Single genital patency (Fig. 2) and single umbilical artery were present. In the patient's abdominal ultrasonography (USG), bilateral renal and bladder agenesis was detected. Any structure that could belong to the over, uterus or testicle could not be displayed in the patient. Echocardiography revealed aortic coarctation. Our patient had pulmonary hypoplasia at the same time and surfactant therapy was given 2 times to the patient. However, we lost the patient due to multiple anomalies on postnatal 8th hour.

3. Discussion

Sirenomelia/mermaid syndrome is accompanied by anomalies in different percentages. These are lower extremity, anorectal, renal, genital, lower urinary system, cardiac, esophageal atresia, other gastrointestinal defects and respiratory tract [3]. Most of the patients are still dying at birth or shortly after birth. The first patient with sirenomelia was reported in 1989 and, as far as we know, in the literature, six living sirenomelia patients have been described up to now [4,5]. While there were imperforate anus anomalies in living and lost sirenomelia patients, our patient had a normal anus [1–8]. Congenital anomalies accompanying the disease, lower extremity anomalies, GUS and CVS anomalies were also present in our patient. Sirenomelia is divided into 7 groups as anatomical type and according to this classification, our patient was accepted as type 2 sirenomelia because of having one fibula [5].

The etiology and pathogenesis of this malformation is still unclear. However, vasculature steal hypothesis and defective blastogenesis are accused in etiopathogenesis [6]. It is thought that the damage occurring in the caudal mesoderm of the embryo between 13th and 22nd days of gestation may cause malrotation, dysgenesis and lower extremity fusion [1,4,6]. The lower extreme fusion can extend from the superficial tissues to the bones. Also in our patient, superficial tissue and bone (femur) fusion was present (Figs. 1 and 3).

Under normal circumstances, there are two umbilical arteries originating from the iliac artery, there is a single umbilical artery originating from aorta in sirenomelia, and this condition is accused in the etiopathogenesis at the vasculature steal hypothesis [6]. In these patients, while the abdominal aorta is usually directly proceeded by bifurcation and iliac arteries, there is renal and inferior mesenteric artery absence [5]. This explains the patient's GUS and GIS anomalies. In these patients, the GIS anomaly was approximately 100%, but no GIS anomaly was detected in our patient [3]. The GIS anomalies most commonly described in these patients are rectal atresia, blind ending colon, imperforate anus, esophageal atresia and omphalocele [4,8]. The patient had a single genital patency just below the coccyx. In the contrast-enhanced graph, it was observed that the patency was linked to the colon (Fig. 3).

Oligohydramnios, bilateral/single renal agenesis and pulmonary hypoplasia which can be detected in the second trimester of fetal USG, are usually accompanied by all sirenomelia patients [1,3]. Sirenomelia

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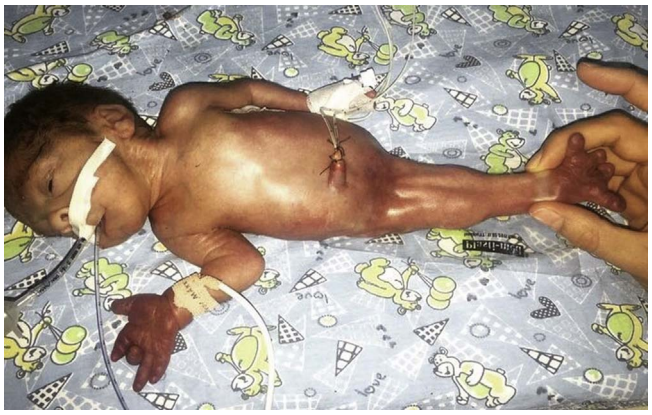

Fig. 1. Only one lower extremity was seen on physical examination.

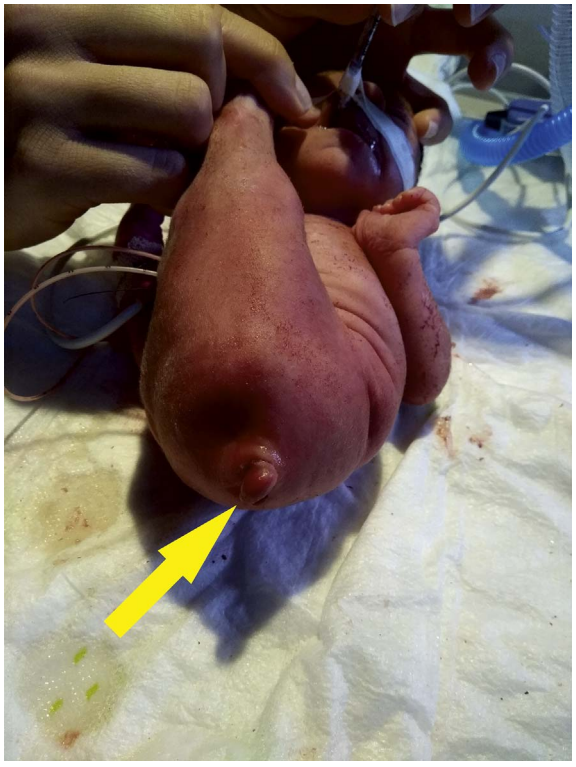

Fig. 2. Single genital patency was seen on physical examination (arrow).

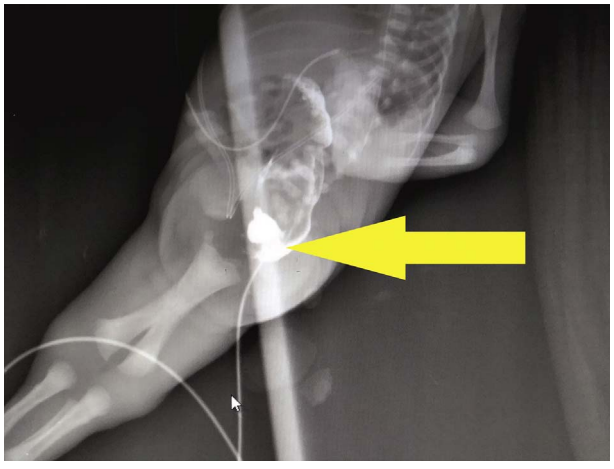

Fig. 3. In the contrast-enhanced graph, it was observed that the patency was linked to the colon (arrow).

is unfortunately fatal in many patients due to characteristic severe pulmonary hypoplasia that occurs due to severe oligohydramnios. However, since there was no prenatal follow-up in our patient, no oligohydramnios or other prenatal findings could be detected firstly in the 9th week. The diagnosis could be made postnatally in our 26-week baby born 780 g.

In addition, caudal regression syndrome (CRS), which was reported by Duhamel in 1961 and seen in 1/40,000–100,000 rate, is also accused in etiopathogenesis [6]. However, the presence of a single umbilical artery is expected in sirenomelia and is not expected in caudal regression syndrome [3]. In Caudal regression syndrome, mother diabetes mellitus (DM) association is close to 20%, while in sirenomelia this ratio is around 2% [3]. There was no history of diabetes mellitus and hypertension in our patient's mother. In addition, there was no use of drugs that could be teratogenic or alcohol which are accused in etiology.

In conclusion, sirenomelia is a severe and rare congenital anomaly, the patients can be born with a normal anus, just as it is with our patient. When detected early, due to fatal proceeding (bilateral renal agenesis - severe pulmonary hypoplasia, etc.), the family needs to be informed about the termination. Patients with no prenatal follow-up or no termination consideration are required a postnatal severe intensive care support and a multidisciplinary approach.

Conflict of interest

None.

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