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Mermaid syndrome

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Summary

Sirenomelia, also known as the mermaid syndrome, is a very rare congenital anomaly characterized by lower limb fusion and severe urogenital, gastrointestinal, cardiovasculer, central nervous system malformations. We report a case of sirenomelia who had a single umblical artery, renal agenesis, pulmoner hypoplasia, esophageal atresia, ventricular septal defect, anal atresia, intestinal atresia and who was lost at fifth hour of life. *(Turk Arch Ped 2013; 48: 65-67)*

Key words: Caudal dysgenesis, mermaid, sirenomelia

Introduction

The mermaid syndrome also known as sirenomelia is a very rare congenital anomaly. This syndrome is characterized by lower limb fusion which gives the baby the appearance of a mermaid and severe urogenital, gastrointestinal, cardiovascular, central nervous system malformations (1,2). These babies are usually not born live or are lost a short time after delivery (3). The degree of urogenital system anomalies determines the chance of survival and a very few number of living subjects has been reported in the literature (4,5). Here, a patient who was diagnosed as mermaid syndrome was presented because this syndrome is encountered rarely.

Case

A baby who was born at the 38th gestational week by cesarean section from the first pregnancy of a 19-year-old mother was referred to our hospital as intubated because of abscence of spontaneous respiration. In the pregnancy period, no pathology except for oligohydramniosis was found. No consanguineous marriage was present between the mother and the father. Respiratory support by mechanical ventilation was started in the patient whose general status was not well. A thoracic tube was placed, since pneumothorax was found in the right lung. The body weight was found to be 2 675 g (25-50%), the height was found to be 38 cm (<10%), the head circumference was found to be 34 cm (50-75%). A 3/6 systolic cardiac murmur was heard. The patient had esophageal atresia (Picture 1) and anal atresia. A single artery was present in the umbilical chord. The external genital structure had a male appearance, but the scrotum was not prominent and the testicles were not palpable. The lower extremities were fused (Picture 2,3,4), but the bony structures were not fused on radiologic examinations (Picture 1). Veventricular septal defect was found on echocardiogram. On abdominal ultrasonography, the kidneys and bladder could not be visulaized. The baby was lost in the fifth hour of life. On autopsy, it was found that both lungs were composed of a single lobe and were hypoplasic. Atresias were found in the enterance of the anus and 7 cm above this together with esophageal atresia. The kidneys, bladder and ureters were absent. While the right testicle was observed in the pelvis, the left testicle could not be found. With these findings the mermaid syndrome was considered. Chromosomal analysis revealed a 46 XY karyotype and that no structural disorder was present.

Discussion

The mermaid syndrome occurs with an incidence of about one in 60 000 births (4,6,7,8). It is still debated if it is a type of

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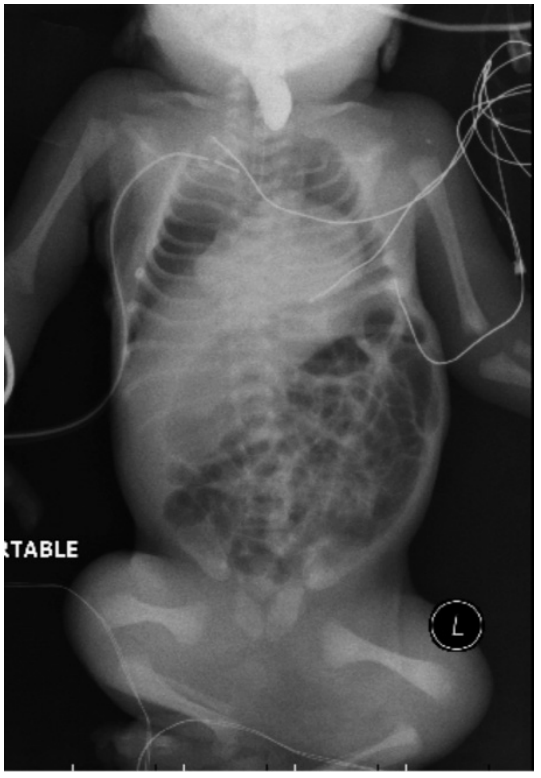
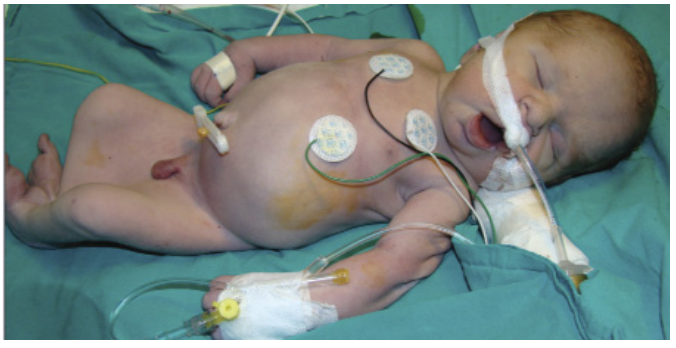
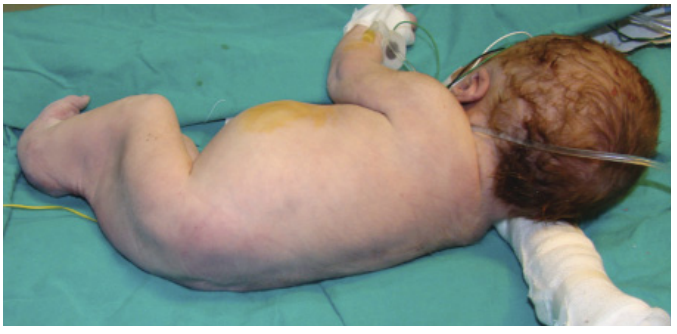


Picture 1. Both leg bones are observed as seperate structures and an appearance compatible with esophageal atresia is observed via radiopaque material given into the esophagus



Picture 2. Appearance of the patient with a diagnosis of mermaid syndrome from the front

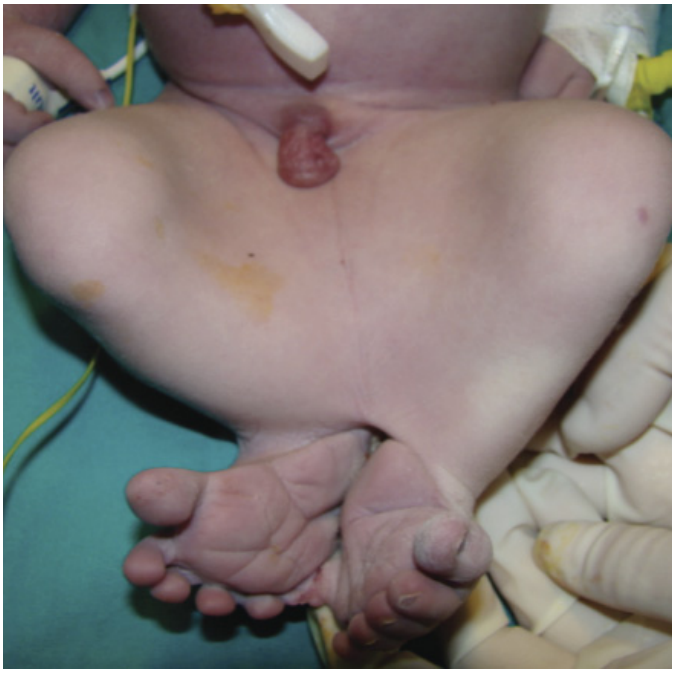


Picture 3. Appearance of the patient with a diagnosis of mermaid syndrome from the back

caudal dysgenesis or a completely separate condition (3,6,9,10,11,12,13). Althoug its pathogenesis can not be explained fully, some theories have been proposed. According to a view, inadequate mesodermal cell migration has been blamed (9,11). According to a more common view, inadequate blood supply to the tissues and organs in the lower part because of presence of a single artery in the umbilical chord causes fusion of the legs and other findings in the embrional period (6,10,11,14,15). Although the causes of this syndrom are not known exactly, risk factors including a maternal age of <20 or >40 years, being an identical twin, exposure to various harmful agents during pregnancy, being the baby of a diabetic mother and male gender have been reported (3,4,15). The maternal age in our case was 19 and the gender was male. Other risk factors were absent.

In the mermaid syndrome, the most notable characterstics is fusion of the legs and the legs may be fused together with the bone structures or only with the skin without fusion of the bone tissues (4,15). In our case, the legs were fused only with the skin. The most commonly reported findings in addition to lower limb fusion include single artery in the umbilical chord and renal developmental disorder (1,12,13). In our case, there was a single artery in the umbilical chord and the kidneys, bladder and ureters were absent. Other commonly reported findings include anal atresia and intestinal obstruction (10). Our patient had anal atresia and intestinal obstruction was found 7 cm above the anal entrance on autopsy examination.

In the mermaid syndrome, esophageal atresia, congenital heart disease, polydactilia, anencephaly, holoprosencephaly and myelochisis may accompany with a lower rate (9,10,16). Our



Picture 4. Fused legs and underdeveloped external genital structure

patient did not have polydactilia or central nervous system disorder, but esophageal atresia and ventricular septal defect were present.

In the mermaid syndrome, external and internal genital organs do not usually develop fully (3,9,10,11,12,14). Although our patient had a structure appearing like penis, the scrotum was not developed and the testicles were not palpable. On autopsy examination, the right testicle was observed in the pelvis, while the left testicle could not be found.

The mermaid syndrome is usually lethal. The degree of renal developmental disorder determines the survival of the baby (13). If a severe defect is present in renal development, oligohydramniosis and lung hypoplasia develop and stillbirth or mortality in the early period may result (3,11). A very few number of surviving patients has been reported in the literature and these usually have functioning kidneys (4,5). In our patient, a prenatal history of oligohydramniosis was present and autopsy revealed that the kidneys were absent and the lungs were hypoplasic. Although respiratory support was provided by mechanical ventilation, our patient was lost in the early period.

Prenatal diagnosis is possible by ultrasonography performed in the early period of pregnancy, but the diagnosis becomes difficult in the advanced periods of pregnancy because of oligohydramniosis (14,15). Termination of pregnancy may be recommended in severe cases diagnosed in the early period.

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