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Sirenomelia (Mermaid Syndrome): An Infant from Parents Who Used a Special Form of Snuff

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Abstract: We report the first case of a fetus with sirenomelia or mermaid syndrome, whose Afghanian parents were heavy user of a special form of snuff. The case was diagnosed as a mermaid syndrome but some of the features were common to both symmelia dipus and symmelia apus, for example, the single lower extremity had the normal femur, tibia and fibula, but the single foot was rotated medially. The digestive tube ended to a massive closed saclike structure and the anus was absence. The external genital organs were rudiment and the normal testes were undescended. Aorta was divided to branches, such that the external iliac arteries were very small in comparison to the internal iliac arteries. Inferior Vena cava was placed on to the left side of the aorta and unlike the majority of reported mermaid syndrome, the bladder was present. So, the researchers believe that the present case is a rare variant of the mermaid syndrome.

Key words: Sirenomelia (mermaid syndrome), lower extremity, snuff, case report

INTRODUCTION

Sirenomelia or the mermaid syndrome is a rare congenital malformation involving multiple organs. It is characterized by the fusion of the lower extremities (Lutz *et al*., 2004). There are some anomalies which accompany this syndrome like: imperforate anus, renal agenesis, absent bladder, sacral agenesis, colonic atresia, rectal atresia and absent internal genitalia. Usually, the gonads are present but remain ectopic in position like the abdominal cavity or the inguinal canal. The incidence of the sirenomelia has been reported as 0.8 in 100,000 (Stanton *et al*., 2003), to 1.5- 4.2 in 100,000 (Horikoshi *et al*., 2005). The condition occurs predominantly in males, with a sex ratio of 2.7: 1 (Murphy *et al*., 1992). Most of the cases of the syndrome are stillborn, or die during or shortly after birth. Until 2005, only four cases of a surviving infant with sirenomelia have been reported by Horikoshi *et al*. (2005).

Use of different forms of oral tobacco is dramatically increasing in the world, but the users are not aware of the health its health risks of some special forms (Soldz *et al*., 2003; Vander Weg *et al*., 2008; Zatterstrom *et al*., 2004). In addition, health effects of the special form snuff which used commonly in east South region of Iran and Afghanistan are not completely studied. Our report shows that use of this form snuff by parents may cause severe malformations in the developing fetus. It is suggested that more studies to evaluate the teratological effects of intake of the special form of snuff need to be elaborated.

CASE REPORT

A rare variant of Sirenomalia, Mermaid syndrome was obtained from the Niknafs Maternity Hospital of Rafsanjan City, who had died just after delivery. His parents were Afghanian in origin. His parents were heavy user of a special form of snuff (an oral form of tobacco product).

Examination of the perineal region and the lower limb exhibited malformations. The fetus had one medially rotated limb. Small rudimentary penis with no scrotal sac was seen (Fig. 1A).

The anus was absence (Fig. 1B). In the sacral region, there was an obvious spine deformity. Upon dissection, the following features were observed, the normal testes which had not descended, lying within the abdominal cavity at the deep inguinal ring.; the large intestine ended in a massive closed saclike in the left iliac region (Fig. 2A). The caecum and vermiform appendix lay in the left inguinal region. The abdominal aorta was divided into two common iliac arteries but on the both sides there were small external iliac arteries and large internal iliac artery. Abdominal aorta was situated left to the inferior vena cava (Fig. 2B).

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Fig. 1: (A, B) Anterior and posterior views of the case. Note to the single lower extremity with malrotated foot, the external genital organs with only a small rudimentary penis, the narrow radix of the first right finger (pollex) in the anterior view and absence of anus in the posterior view

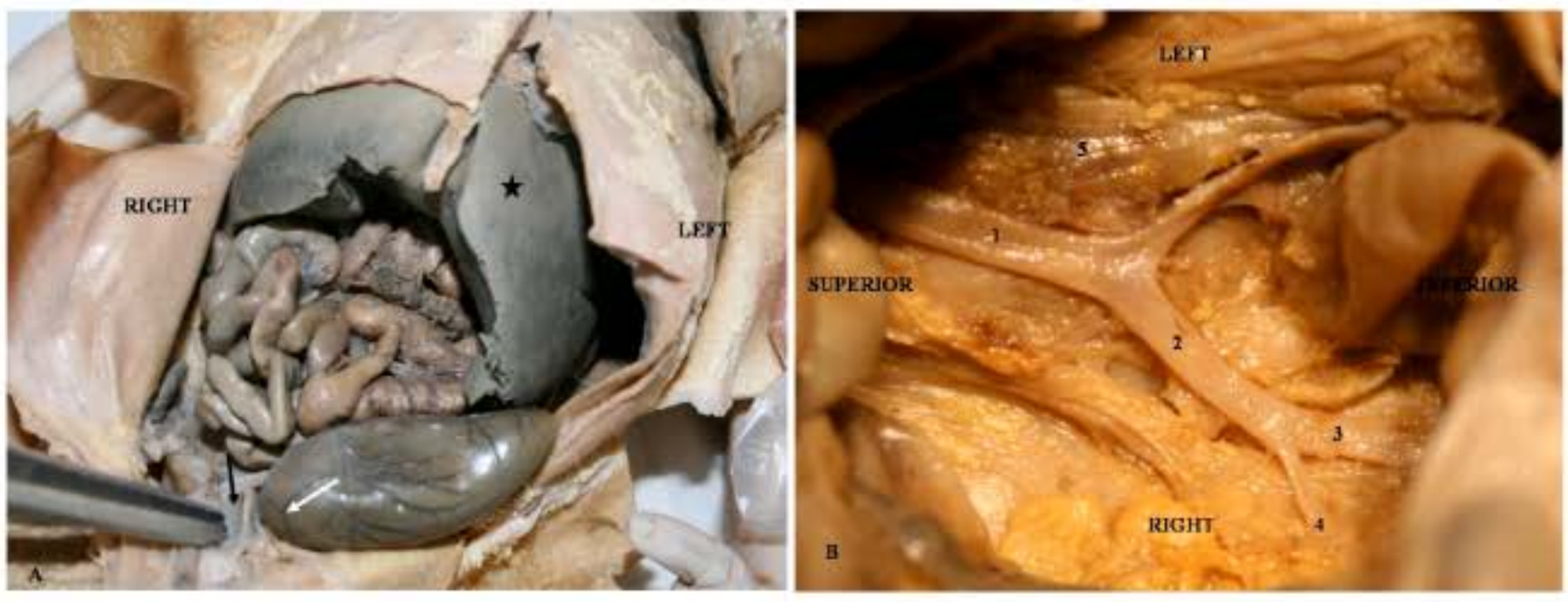

Fig. 2: Photographies of the dissected case. (A) Photograph of whole dissected abdominal cavity. The large liver, the massive closed saclike end of digestive tube and the testis are shown with star, white and black arrow, respectively and (B) photograph of the abdominal aorta and its main branches. Note to the small external artery and inferior vena cava on the left side of the abdominal aorta. 1: Abdominal aorta, 2: Common iliac artery, 3: Internal iliac artery, 4: External iliac artery, 5: Inferior vena cava

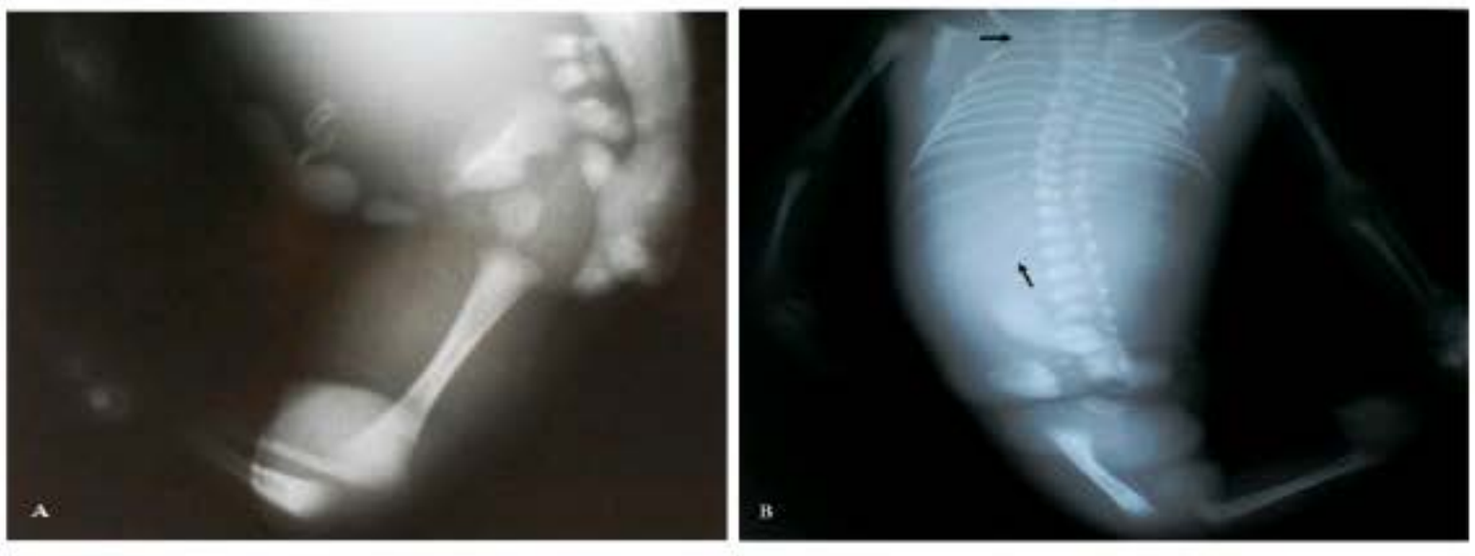

Fig. 3: Radiographs from the case. (A) X-ray from the single lower extremity showing normal size of tibia and fibula and (B) X-ray showing additional ribs in both sides and normal size of tibia bone. With regard to the single lower extremity and finform malrotated foot our case is classified to the symmelia apus and symmelia dipus groups, respectively. The first and thirteenth ribs are shown with arrow

Both the lobes of the liver occupied the upper abdominal cavity with a gall bladder. The urinary bladder was present (Fig. 2A). The radiographs showed that the femur, the tibia and the fibula of the single lower limb were normal. The femur bone articulated with the left acetabulum (Fig. 3A). He had 13 ribs on the both sides (Fig. 3B). The first right finger (pollex) was abducted and was narrow in the radix (Fig. 1A).

DISCUSSION

The first reported case of the mermaid syndrome was in the 16 century (Lutz *et al*., 2004). One of oldest studies was done by Malinger *et al*. (1987). Sarpong and Headings (1992) classified the syndrome into 3 groups according to the number of feet present. The first group is called symmelia apus, in which both legs are merged completely into a single lower extremity, with both the feet absent or rudimentary. The second group is symmelia unipus; Fetuses have 1 foot, 2 femora, tibiae and fibulae. In the third group that is called symmelia dipus, 2 distinct feet are present but are malrotated and resemble fins (Sarpong and Headings, 1992). This case belongs to the symmelia apus variant having one foot with 1 femur, tibia and fibula (Fig. 1A). The last feature (i.e., his foot rotated medially and was resemble fin) can set our case symmelia dipus, but unlike the majority of previous reported cases, the fibula and tibia in the single lower extremity is normal in size and form (Fig. 3). Although, some features were similar to fetus in unipus group, for example fetuses in the third group have two distinct feet but our case had only one finform malrotated foot.

Several etiological factors have been suggested as to the pathogenesis the syndrome such as: caudal somite destruction, cloacal malformation and neural tube distention. Duhamel suggested that a deficit in the formation of the caudal region prior to the 4th week of gestation leads to this malformation. Duhamel suggestion has led to the commonly used term, \caudal regression syndrome (Lutz *et al*., 2004). Rougemont *et al*. (2008) suggested that a defect in the blastogenesis leads to this condition. The theory as known, vascular steal was first proposed by Stevenson. This theory suggests that blood is diverted by a single large artery arising high on the abdominal aorta. However, it still remains unknown that sirenomelia is part of the spectrum of caudal regression or a distinct entity (Duesterhoeft *et al*., 2007). Relative ischemia to the caudal part of the developing embryo seems to be evident in the present case as the divisions of the aorta was not normal; the external iliac arteries were smaller in comparison with the internal iliac artery. In addition the aorta was placed on the right side of inferior vena cava (Fig. 2B).

The etiology of this malformation still remains unknown. Reports have shown that association between the syndrome and maternal diabetes, cocaine exposure and monochorionic twin exist (Van Zalen-Sprock *et al*., 1995; Di Lorenzo *et al*., 1991).

Snuff is a powder tobacco product which is consumed by placing it under the upper lip for an extended period of time (Vander Weg *et al*., 2008). The parent's in the present case used a form of snuff which is manufactured in East South region of Iran and Afghanistan. It consists of a complex of tobacco and little of lime [Ca $(OH)^2$]. The health effects of this form of snuff have not been completely studied, but it seems that it causes oral and esophageal cancers. We assume that the abnormalities in our case could be due to the use of snuff by his parent's. In certain areas of Iran and Afghanistan, usage of this form of snuff is very common but unfortunately, very few studies about its health effects have been conducted (Glenn *et al*., 2008).

CONCLUSION

At the first present results show that the presented case was a rare variant of the mermaid syndrome and he wasn't grouped in the famous classification of the syndrome. In addition, using the mentioned form of snuff by parents may cause serious malformations in their fetus. It is suggested that the consequence of snuff consumption by parents on the fetus deserve further studies.

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