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A CASE OF SIRENOMELIA*

In one of a pair of identical twins, and in association with exomphalos

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Sirenomelia is a syndrome characterized by the fusion of the inferior limbs (symelia), by vertebral anomalies such as an increase in vertebra number or sacral or anorectal agenesis, by urinary tract anomalies such as bilateral renal agenesis or ureteral vesical and urethral agenesis, and by rectal anomalies[1,2].

The earliest known description of a human sireniform foetus, quoted by Kampmeier[3] in 1927, dates back to the first century A.D. The condition is likened to that of a mermaid.

Duhamel[1], in 1961, termed these anomalies "the syndrome of caudal regression". The incidence is one in 60.000 newborn and there is a male preponderance of 2.7:1[1-4]. It occurs more often in one of a pair of identical twins[2,4-8]. Due to the present anomalies some of the babies with this syndrome are stillborn and some die soon after birth[1,3].

As this syndrome is very rare and since, as far as we know, a case has so far not been reported from Turkey, we thought this one worth presenting.

Case Report

A one-day-old male infant was admitted to our hospital with multiple anomalies. He was one of a pair of identical twins. The twins were born of a nineteen-year-old mother by spontaneous vaginal delivery at thirty eight weeks of gestation. The other twin was normal. They had shared the same placenta and there was no oligohydramnios. The parents were not related.

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The patient weighed 1500 grams and was 35 cm long. Head circumference was 31 cm with normal fontanel and sutures. He had low-set ears, slight micrognatia and omphalocele. The lower extremities were fused and mermaid shaped. There were six digits on the left leg and there was syndactyly on the right leg. There was flexion deformity of the elbows and nonpitting edema of the hands. There was a primitive genital tubercule 0.5 cm in size, absence of testes and scrotum and imperforate anus (Figure 1).

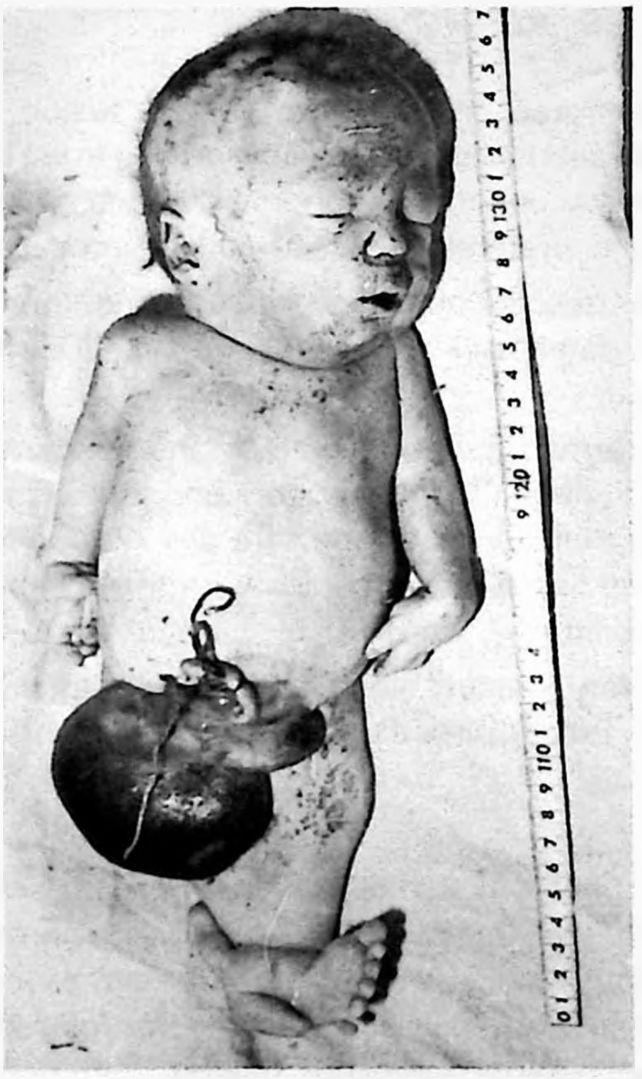

Figure 1

The appearance of the fusion of the lower extremities and exomphalos.

Radiological survey revealed eight cervical and six lumbar vertebrae, a small pelvis, agenesis of the right ileum, a hypoplastic sacrum (Figure 2) and mandibulofacial dysplasia (Figure 3).

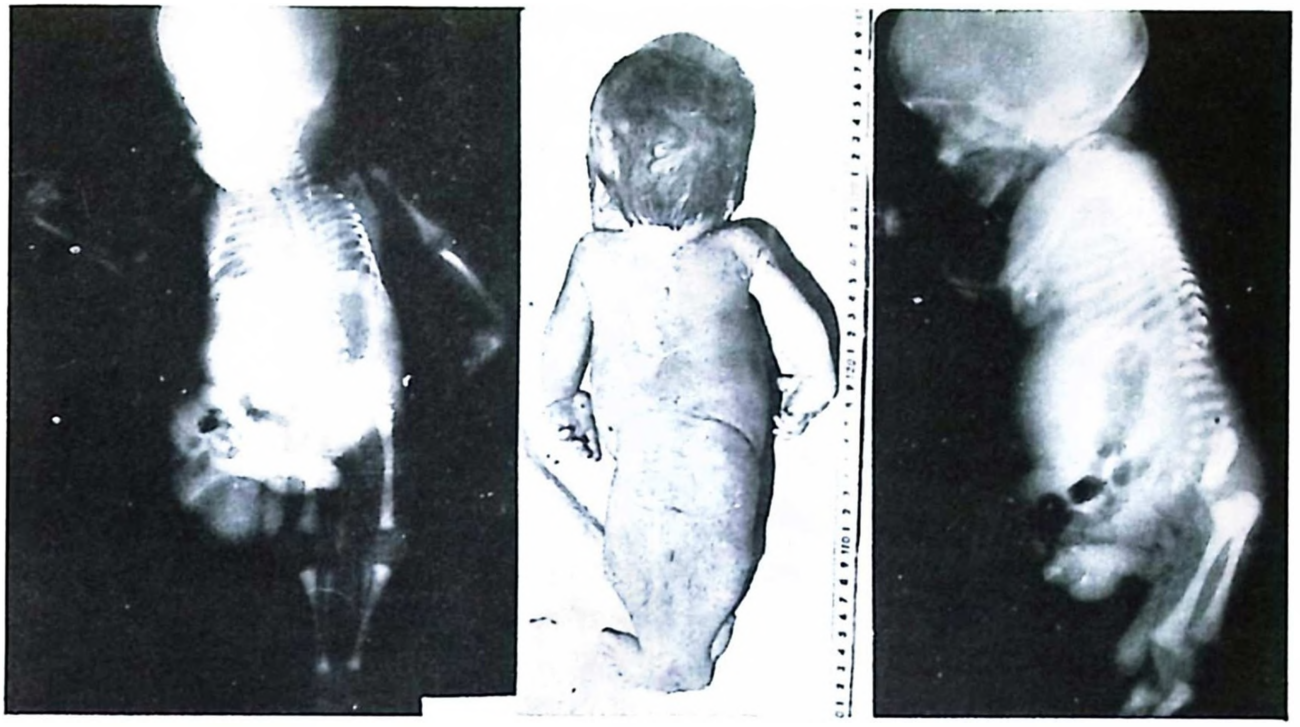

Figure 2

X-ray showing agenesis of the right ileum and six lumbar vertebrae.

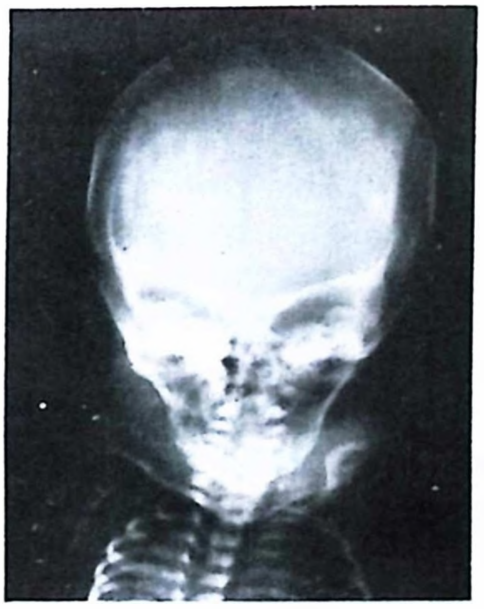

Figure 3

X-ray showing mandibulofacial dysplasia and eight cervical vertebrae.

Chromosomal examination of the patient was carried out by counting fifty metaphases from peripheral blood lymphocytes and the 46, XY caryotype was identified.

The baby died of renal insufficiency during the first day of life. Postmortem examination revealed a single umbilical artery, arising as a midline continuation of the aorta, aplasia of the right and hypoplasia of the left kidney, absence of the renal artery, both the ureters, the bladder, urethra and rectum. The testes were located in the pelvis. The lungs, the heart and the other viscera were all normal.

The histologic examination of the hypoplastic kidney showed a well-defined cortex and medulla, immature glomeruli and poorly-formed microcystic tubules and collecting ducts.

Discussion

The primary defect of the caudal regression syndrome is in the midposterior axis mesoderm and/or caudal blastema[4]. The most severe form of this defect, sirenomelia or sympodias, is presumably the consequence of a wedge-shaped early deficit of the posterior axis caudal blastema, leading to the fusion of the early limb buds and their fibular margins, and to the absence or incomplete development of the intervening caudal structures. The embryonic defect apparently dates back to the third week, prior to development of allantois. For this reason there is usually absence of allantoic vessels. There is a single umbilical artery arising directly from the aorta[4,9,10]. Sirens occasionally occur in the offspring of rats treated with trypan blue[11].

This case had fusion of the lower limbs and a single umbilical artery. An increase in vertebra number, sacral agenesis, imperforate anus, absence of external genitalia, renal agenesis, horse-shoe kidney, ureteral, vesical and urethral agenesis, oligohydramnios and amnion nodosum are commonly encountered features of this syndrome[1-4,10]. This case had imperforate anus, additional vertebrae, only one iliac bone and a primitive genital tubercule 0.5 cm in size. There was also an absence of testes and scrotum, aplasia of the right and hypoplasia of the left kidney, together with absence of the renal artery, both the ureters, the bladder and the rectum. This case also had the uncommon association of exomphalos, so far reported in only four cases[12-14].

In connection with sirenomelia, radial hypoplasia, esophageal atresia with tracheoesophageal fistula (TEF), congenital heart defects such as a high ventricular septal defect and cor triatrium, flexion deformity of the elbows, bilateral cloudy cornea and hamartoma in the neck region, have all been reported[3,4,14]. In this case there was no congenital heart defect, TEF or radial hypoplasia, but there was flexion deformity in both elbows.

The Say syndrome (imperforate anus, polydactyly and hemivertebra) should be taken into consideration in differential diagnosis[15]. In the Say syndrome vertebral anomalies such as bifid vertebra, hemivertebra and sacral agenesis are frequently seen, but epistasis is not encountered; also genital anomalies and oligohydramnios are rare and fusion of the lower extremities is absent.

Moreover cases of Potter's syndrome in association with sirenomelia have also been reported[16,18]. Although in this case the ears were low set and there was mandibulofacial dysplasia, there were no prominent epicanthal folds, nor a depressed nasal bridge, flat nose, pulmonary hypoplasia or oligohydramnios[19]. As a consequence Potter's syndrome was not considered in our case.

Summary

A case of sirenomelia with exomphalos in one of a pair of identical twins is described. Radiological examination revealed eight cervical and six lumbar vertebrae, a small pelvis, agenesis of the right ileum, a hypoplastic sacrum and mandibulofacial dysplasia. Autopsy findings included a single umbilical artery, aplasia of the right and hypoplasia of the left kidney, absence of the renal artery, of both ureters, the bladder, the urethra and the rectum.

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