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CASE REPORT

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Mermaid syndrome: virtually no hope for survival

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Abstract Sirenomelia, also called the mermaid syndrome is a severe malformation involving multiple organs and characterized by partially or completely developed lower extremities fused by the skin. The birth of a ''mermaid'' is very rare (1.2–4.2 cases for 100,000 births); most are stillborn, or die at or shortly after birth. The case of a living female neonate with dipodic simelia (fusion of well-developed legs) is presented. No prenatal diagnosis was made and the newborn had an uneventful neonatal course following Cesarean section delivery. The complex and striking malformation was obvious at birth and further evaluation revealed very poorly functioning kidneys, associated with abnormal anorectum, urogenital tract, and external genitalia, as well as a pelvic malformation. Supportive care was applied because of the poor prognosis and the child died at 7 weeks of age, due to renal failure.

Keywords Sirenomelia · Mermaid syndrome · Renal dysplasia

Introduction

The complexity of Mermaid syndrome, a rare malformation, is illustrated by a case report, and the almost inevitable poor outcome is addressed, based on a review of the literature.

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Case report

In December 2000 a girl was born at 37 3/7 weeks of gestation by Cesarean section from a healthy IIIG IIIP 33-year-old mother following an uneventful pregnancy, where prenatal ultrasound had been performed and described as normal. Neonatal adaptation was excellent with an Apgar score of 9–10–10. The diagnosis of dipodic simelia was made at birth and the newborn was transferred to our institution for further evaluation and treatment. On admission, she weighed 3400 kg , measured 52 cm, and had a head circumference of 33 cm. Slight dysmorphic signs included a flat forehead, a wide nasal base, and wide hands. She was well perfused and presented normal cardiopulmonary function. Neurological examination was normal for age. The abdomen was soft and non-tender. There was one single umbilical artery and bilateral femoral pulses were palpable. Her lower extremities were well developed but totally fused by the skin from the hips to the heels (Fig. 1). Extension of the hip and flexion of the knees were impossible and painful. A grasping reflex was present on the sole of both well-developed feet. The perineum was abnormal (Fig. 2), dorsally displaced, with a small anus, long and partially fused external genitalia, and a single vulvar orifice with no uretral meatus. The caryotype was normal (46 XX).

Electrolyte analysis at 10 h of age revealed normal plasma levels of sodium and potassium, urea of 5.5 mmol/l, and creatinine of 120 μmol/l. Radiological studies revealed a narrow pelvis with a horizontalized sacrum. The bony structure of the lower extremities was normal, but both legs were externally rotated 180°. Ultrasound of the heart, and Doppler studies of the major arterial trunks, were within normal limits. Abdominal ultrasound revealed two 2.5-cm-long hypoplastic and diffusely microcystic kidneys. No uterus was

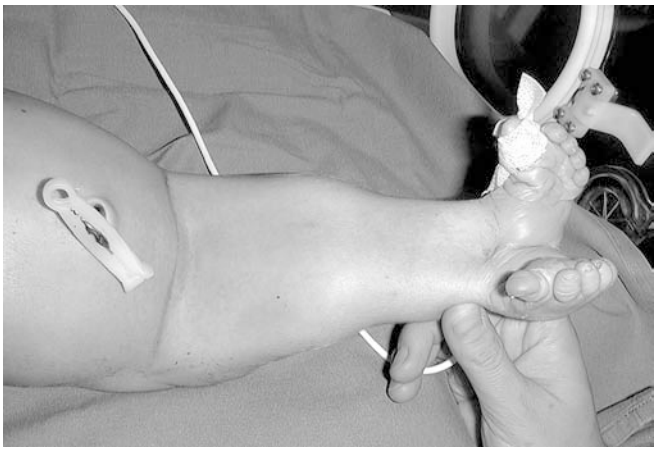

Fig. 1 Anterior view of the trunk and legs

seen. Anal opacification revealed a long and narrow anal canal without rectal dilatation. Vaginal retrograde opacification revealed a tortuous urogenital sinus with no well-defined bladder but multiple cavities leading to refluxing and tortuous ureters (Fig. 3).

Following several multidisciplinary discussions, including with a pediatric surgeon, an orthopedic surgeon, a nephrologist, a medical ethics specialist, and the nursing staff, it was decided in accordance with the family that purely supportive care would be the best attitude toward a child with such a poor prognosis. She was then transferred back to the district hospital close to her parents home. Her weight gain was never adequate, although feeding her presented no difficulty. Bowel movements were irregular but spontaneous with a formula-milk diet. At 2 months of age, she became progressively lethargic, then unresponsive and quickly comatose. She died soon after. Her last blood tests at 7 weeks of age included a sodium plasma level of 117 mmol/l, a potasium plasma level of 6.1 mmol/l, a creatinine plasma concentration of 227 mmol/l, and a base excess of –16.4 mmol/l.

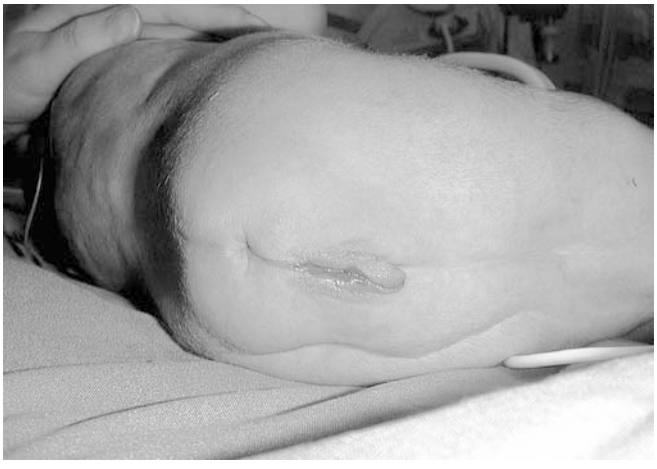

Fig. 2 Posterior view of the perineum and legs with the dorsally displaced anus and the abnormal external genitalia

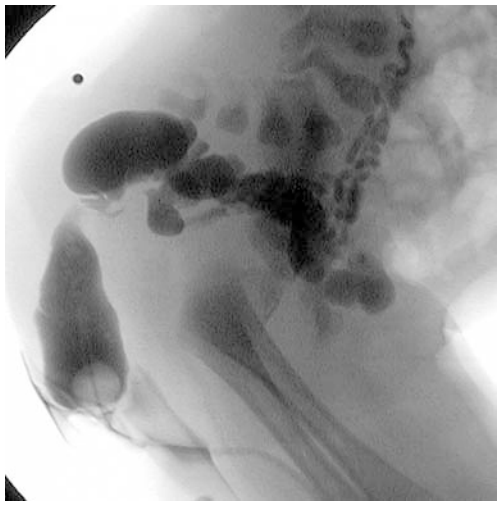

Fig. 3 Lateral view of a retrograde opacification of the urogenital sinus using a balloon catheter, showing multiple pelvic cavities filled with contrast, refluxing in tortuous ureters

Discussion

The Mermaid syndrome has been described since the sixteenth century and has almost always been fatal. One of the most significant studies on sirenomelia was published in 1987, describing 33 cases and reviewing the literature [1]. The various forms based on the extent of lower limb abnormalities and limb fusion have been well described and classified by Duhamel [2]. Frequency varies from 1.2 to 4.2 cases for 100,000 births [2, 3, 4, 5]. The etiology of this malformation remains unknown: the pathogenesis may be neural tube distention, cloacal malformation, or caudal somite destruction [1]. A deficit in the formation of the caudal region prior to the fourth week of gestation has been suggested by Duhamel [2] and has led to the commonly used term ''Caudal regression syndrome.'' More recently, Langer et al. suggested a defect of the blastogenesis following the observation of a case of sirenomelia associated with situs inversus [6].

Malformation of nearly every system has been described in the Mermaid syndrome, including urogenital, musculoskeletal, anorectal, vascular, cardiopulmonary, and central nervous systems [1,4]. Among these frequently associated abnormalities, renal dysplasia and a single umbilical artery are almost invariably described. The severity of renal insufficiency directly affects the prognosis of a surviving mermaid; therefore, as our case illustrates, prompt post-natal assessment of renal function is mandatory in order to direct future management of any child born with sirenomelia. There is no report of long-term survival. A review of the literature revealed three neonates with the mermaid syndrome, who survived up to 6 months of age [7, 8, 9]. Only in 1 case were the kidneys normal, although the child had a ventricular septal defect and an ectopic urethra ending 3 cm superior to the anus [8]. Extensive reconstructive surgery

could be anticipated but was not reported. The long-term quality of life in these children is not known.

Conclusion

In conclusion, the decision to operate on living newborns with the mermaid syndrome is influenced by the extent of its malformations, and mainly by the extent of renal dysplasia. A complete work-up assessing all major organs is mandatory in such children. Supportive care is almost always the most appropriate course of action.

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