Anaesthesia and Sirenomelia (Mermaid Syndrome)

A Bösenberg
Department of Anaesthesia, University of Cape Town, Cape Town, South Africa

Synopsis

A 2.4kg one day old, the product of a 36-week monozygotic twin pregnancy born to a 24 yr old primigravida by C-section for foetal distress, presented for laparotomy. The child's mother had attended antenatal clinic once during her otherwise uneventful pregnancy. No antenatal investigations were performed. The monozygotic twin sibling, a male weighing 2.65kg, was completely normal.

On examination of the abnormal twin, the most striking feature was the complete fusion of the lower limbs from buttock with no perineum to the heels, and a single 5th toe (sympus); there were also no external genitalia or urogenital orifice, and an imperforate anus. The feet were inverted, the popliteal region was dimpled and the sole of the feet faced forward. The feet were fused along the lateral border and resembled a flipper (dipus) (Figure1 and 2). The rest of the examination, from the umbilicus upwards, was completely normal.

Preoperative ultrasound of the abdomen revealed what was thought to be rudimentary kidneys. No bladder could be identified, but the liver and spleen were normally situated. X-ray of the lower limbs showed a normal looking pelvis, femurs, tibiae and fibulae, and abnormal feet. Full blood count, urea and electrolytes were normal.

Neither the paediatric surgeon nor anaesthesiologist had prior experience of this clinical problem and on a Sunday, access to a reference library was not possible. A colostomy, and if possible a stoma for urinary drainage, was planned.

Anaesthesia was induced with halothane and intubation achieved without muscle relaxants. In view of the easily palpable sacral hiatus, a caudal block using 2.5ml 0.25% bupivacaine was performed. A block to the level of T8 was achieved.

Laparotomy revealed bilateral renal agenesis, an absent urinary collecting system and no bladder. The rudimentary kidneys seen on ultrasound were in fact the adrenal glands. In view of hopelessness of these findings further surgery was abandoned. The gravity of the situation was explained to the mother. The infant was kept comfortable and discharged at mother's request to die at home.

A second case of sirenomelia was admitted to the paediatric surgical unit at King Edward VIII Hospital a year later. In view of the hopeless prognosis, no surgery was offered.

Figure 1. Monozygotic male twins. The typical features of sirenomelia are present in one – fused lower limbs (dipus) with flipper like feet, absence of external genitalia, imperforate anus and normal upper torso. Features of Potter's facies are not present.

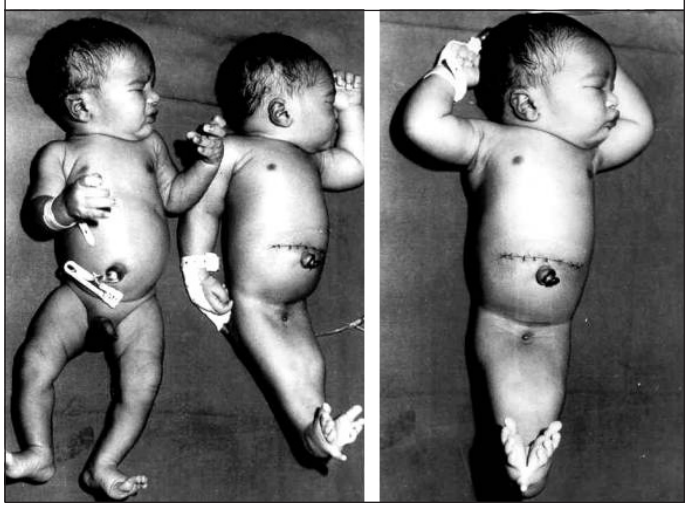
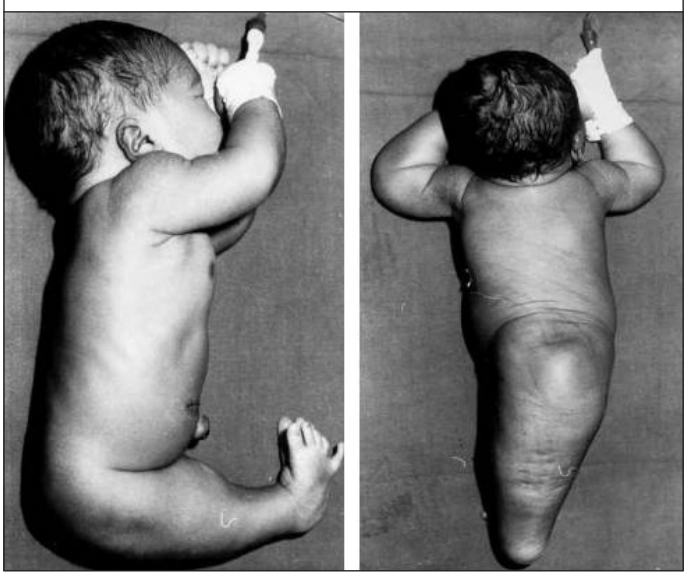


Figure 2. Lateral and posterior views of the sirenomelia. The sacral hiatus was easily palpable despite the abnormal buttock. The feet are rotated inwards and fused along their lateral borders with a single 5th toe.



Correspondence:
Prof Adrian Bösenberg
email: bosie@cormack.uct.ac.za

Introduction

Sirenomelia is an extremely rare developmental malformation characterized by complete or partial fusion of the lower limbs and gastrointestinal and urogenital malformations.[1-3] Sirenomelia is seldom compatible with life. Most infants are stillborn and few survive beyond the neonatal period. The term sirenomelia is derived from Greek *seiren* (see footnote) and *melia* or limb. Descriptions of sirenomelia can be traced back to the 16th and 17th centuries, although these first reports were still considered a mixture of reality and mythical imagination.[2] Public interest has most recently been aroused by the successful separation of the limbs of an infant with sirenomelia in Lima, Peru.[4] This infant is one of few known survivors[8,14,18] beyond the early neonatal period.

The reported incidence is said to range between 1:60,000-100,000 live births with a male to female ratio of 2.7:1. Without genetic or post mortem studies the gender can often not be determined from the external genitalia. The occurrence in one of two monozygotic twins, as in our case, is not uncommon and is said to occur 150 times more than in singletons. Various classifications have been proposed but the division into three types is perhaps the most practical3 (i) *Apus* – no feet, only one femur and one tibia, (ii) *Unipus* - one foot, two femora, two tibia, two fibulae, and (iii) *Dipus* – two feet usually fused at the heel, two fused legs and two feet giving the appearance of a flipper. The latter is also referred to as the "mermaid syndrome".

Etiology

The cause of sirenomelia is not well understood and various mechanisms have been postulated. These include the following:

(i) Failure of the caudal mesoderm blastogenesis or an extreme version of the caudal regression syndrome has received most attention, fusion of the early limb buds with failure in varying degrees of development of intervening caudal structures.
(ii) A vascular steal phenomenon, causing relative ischaemia below a persistent vitelline artery that diverts blood from the abdominal aorta and caudal structures to the placenta.[5]
(iii) Mechanical compression by amniotic bands or oligohydramnios, or
(iv) Teratogens (cadmium, retinoic acid, irradiation)
(v) Genetic mechanisms- recent animal experiments have suggested that sirenomelia results from fusion of the hindlimb buds caused by a defect in the formation of the ventral mesoderm mediated by a bone morphogenetic protein (Bmp).[2]

Clinical presentation

Sirenomelia is difficult to diagnose antenatally by ultrasound because the oligohydramnios, usually severe, hampers proper examination of fetal extremities.[6] MRI has proved more valuable in this situation.[7,8] When sirenomelia is suspected, colour doppler imaging may be used to demonstrate an abnormal vascular pattern in the lower extremities.[9] Once diagnosed, termination of pregnancy is advocated in view of the poor prognosis particularly when associated renal agenesis is present.[10]

If the diagnosis is not made antenatally it becomes obvious at birth. The extent of the renal dysgenesis, which ultimately determines the prognosis, is less obvious. No two cases are alike. Classically the clinical features of sirenomelia include fused lower limbs, imperforate anus and complete absence of urogenital system (bilateral renal agenesis, absent ureters, urinary bladder, absent internal and external genitalia).[1,3,8-12] Invariably a single umbilical artery is present[1,9,11,15], but other life threatening anomalies may be associated.

Fetal urine production contributes significantly to the production of amniotic fluid. This in turn is essential for normal pulmonary development. The lack of urine secondary to renal agenesis is a major factor in the development of the "oligohydramnios deformation sequence".[1] Essentially, the effect on the developing fetus is one of compression caused by the lack of space in utero. The oligohydramnios may cause pulmonary hypoplasia (another factor in the early demise of these patients), and the "compression" may cause abnormal facies (Potter's) or abnormal positioning of the hands and feet. Oligohydramnios occurs in other renal disorders that interfere with formation of amniotic fluid and may also be seen in infants who have normal kidneys, but have had a prolonged amniotic fluid leak in utero.[17] The 'Potter facies,' which is considered typical of renal agenesis (but can occur with any cause of fetal compression), consists of wide-set eyes, 'squashed' nose, receding chin, and large, low-set ears deficient in cartilage.[16]

A variety of other sporadic abnormalities have been reported in association with sirenomelia.[15] These include congenital heart defects, lower sacral/vertebral anomalies[14,18], abdominal wall defects, gallbladder agenesis, upper extremity defects[15] and a vestigial tail.[11]

Anaesthesia considerations

There are no published reports of the anaesthetic management of sirenomelia. The major considerations center around the renal and pulmonary function and the type of surgery (colostomy, urinary stoma, division of limbs) planned. But, as with any neonate born with a congenital defect, associated or incidental abnormalities that could affect the conduct of the anaesthetic must be excluded. In our patient an exploratory laparotomy was performed, partly through ignorance of the usual findings in sirenomelia, and partly to provide a stoma for both the urinary and gastrointestinal tracts. The ultrasound report proved misleading.

Provided placental function prior to delivery is normal, electrolytes and the fluid status in a newborn can be completely normal even in the presence of bilateral renal agenesis. This can be misleading, as in our patient. Metabolic disturbances (e.g. hyperkalaemia, hyperglycaemia, acidosis), fluid overload, haematological abnormalities (anaemia, platelet dysfunction), and cardiovascular and gastrointestinal changes develop later. The rate at which these changes develop, depend on the severity of the renal dysplasia present.

Ideally, in the absence of renal function, anaesthetic agents should be used that do not require renal elimination for termination of their action, nor should potential nephrotoxins be used. Inhalation agents can be used for induction and maintenance of anaesthesia. Desflurane and isoflurane are theoretically the best choice. Muscle relaxants are best avoided but newer agents such as cisatracurium can be used safely in renal failure. Remifentanil requires nonspecific tissue esterases for the termination of its action. These are well developed at birth. Remifentanil may also be useful since absent renal function does not result in a prolonged effect.

The decision to perform a caudal block in our patient was based on the presence of an easily palpable sacral hiatus, the presumption that some renal function was present, and a desire to avoid muscle relaxants and opiates. The advantages of a caudal block include a reduction in anaesthetic requirement, haemodynamic stability, a reduction in the stress response, and a reduced need for postoperative ventilation. In view of the intraoperative findings in this case, a continuous epidural infusion was not used.

Local anaesthetic agents (bupivacaine), are metabolized by the cytochrome P450 enzymes in the liver, although a significant amount is excreted unchanged in the urine. In the absence of renal function, it is presumed that bupivacaine is entirely dependent on hepatic metabolism. Despite renal agenesis, no untoward effects from the caudal block were noted in our patient.

Conclusion

The antenatal diagnosis or delivery of a neonate with sirenomelia presents a difficult clinical and ethical dilemma. Not only is the event a medical curiosity, but the prognosis is usually hopeless. In the rare event of survival, the subsequent surgical and management is fraught with difficulty. The final management decision should be addressed with compassion and understanding and preferably out of the limelight of the media.

A newborn infant with sirenomelia should prompt further investigation of the family for the presence of hereditary renal adysplastic anomalies. There is a higher incidence of dysgenetic renal anomalies amongst siblings and offspring.[19-23] Bilateral renal agenesis is associated with a recurrence risk of about 1% in siblings.[19]

The likelihood of providing anaesthesia for an infant with sirenomelia outside a tertiary care center is remote. However, armed with this information, practitioners can make a more rational decision with regard to future management of these infants, and whether referral to a tertiary center is in the best interests of the child or their family.

Footnote: *In Greek mythology, the Sirens are creatures with the head of a female and the body of a bird. They lived on an island (Sirenum scopuli; actually three small rocky islands) and with the irresistible charm of their song they lured mariners to their destruction on the rocks surrounding their island before devouring them (Virgil V, 846; Ovid XIV, 88). On a journey passed the Sirens, Odysseus, on the advice of the enchantress Circe, tied himself to the mast of his ship in order to hear the sirens' beautiful voices and ordered the sailors to stuff their ears with wax thereby preventing them from hearing their voices. The Sirens sang when they approached, their words even more enticing than the melody. They would give knowledge to every man who came to them, they said, ripe wisdom and a quickening of the spirit. Odysseus' heart filled with longing, but the ropes held him and the ship quickly sailed out of earshot to safer waters (Odyssey XII, 39).*

In early art, the Sirens were represented as birds with the heads, and sometimes the chests, of women. Later, they were represented as female figures with the legs of birds, with or without wings. Birds were chosen because of their characteristic beautiful voices. However, later in history Sirens were sometimes also depicted as beautiful women (whose bodies, not only their voices, are seductive), or even as mermaids (half woman, half fish). The fact that in some languages (such as French) the word for mermaid is Siren adds to this confusion.

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