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Sirenomelia (Mermaid syndrome): Clinical observation of a case at the University Teaching Hospital 'Pr Bocar Sidy Sall' of Kati, Mali

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Abstract **Case Report**

Sirenomelia is a rare form of caudal dysgenesis generally incompatible with life due to the severe renal malformations associated. In Africa, it is associated with mystico-religious considerations and witchcraft and sometimes exposes the family to a violent stigmatization. The transgressions of socio-cultural prohibitions by parents would be the cause of the occurrence of this malformation. Its etiology is still very controversial. We report the observation of a case born at the University Teaching Hospital 'Pr BSS' of Kati, this is the first Malian case reported in the literature.

Keywords: Mermaid syndrome, Sirenomelia, Caudal dysgenesis.

INTRODUCTION

Sirenomelia is a rare form of caudal dysgenesis first described by Rocheus in 1542 and by Polfyr in 1553 (Kampmeier OF., 1927). It is characterized by a variable degree of fusion of the lower limbs. Its prevalence is estimated at 1 per 100,000 live births (Valenzano *et al.*, 1999). More than 50% of patients die in utero, and those born alive usually die within the first 48 hours of life due to the severe renal malformations frequently associated with this condition. In Africa, the birth of these so-called "fish babies" is often associated with mystico-religious considerations and the mothers of these children are often accused of witchcraft. Very few cases have been reported in Africa (Morfaw & Nana, 2012; Ugwu *et al.*, 2011). We report the first Malian case of sirenomelia, observed at the University Teaching Hospital of Kati.

OBSERVATION

Mrs FM Malian aged 26 years primigravida nulliparous without particular medical history surgical, came of herself for painful uterine contraction on pregnancy of 37 weeks 02 days.

This pregnancy was contracted following a treatment given by an obstetrician gynecologist for primary infertility and the pregnancy was monitored by the same obstetrician with 6 prenatal consultations and 2 ultrasonographies, the 1st one at 6 weeks and 5 days and the 2nd one at 21 weeks, which showed dysplastic and polycystic kidneys. She received a dose of tetanus vaccine, a dose of sulfadoxine and pyrimethamine and iron + folic acid supplementation.

Her prenatal biological workup was normal. She did not present any notion of consanguinity with the husband and previous contraception.

On admission: general condition was good, conjunctiva were well colored, no jaundice, no cyanosis or edema. Blood Pressure: 110/60 mmHg, weight = 74 kg, capillary blood glucose = 5.2 mmol/l

Breasts were even, symmetrical and gravid in appearance, anodular without galactorrhea. Abdominal examination revealed the following: a uterus with longitudinal development measuring 32cm with painful uterine contractions estimated at one contraction every 10 minutes, breech presentation, fetal back to the right, fetal heartbeat = 140 beats/min.

Vulva was soiled with mucous plug. At the vaginal examination we noted: an effaced cervix dilated to 1cm, the water bag absent, breech presentation in

decomplete mode engaged at +1. The examination glove returns soiled with mucous plug.

Actions carried out:

- Taking a safe venous line.
- Indwelling urinary catheter.
- Emergency cesarean delivery for severe anamnios and breech presentation.

The cesarean delivery allowed the extraction by the podalic pole of a live baby with an Apgar: 4/10 at 1 minute, 6/10 at 5 minutes and 6/10 at 10 minutes.

The newborn was referred to pediatrics for neonatal distress and malformation.

Examination of the newborn:
Birth weight: 2500g, height = 48 cm, head circumference = 34 cm, chest circumference = 32cm, Heart rate = 132 bpm, SaO2 = 85%,

Examination of the newborn noted the presence of a single umbilical artery, absence of external genital organs, anal imperforation and fused lower limbs from their base to the feet and their external palpation gave the impression of probably having two femurs and two tibias. The two feet were connected by their soles with 9 toes distributed in two lines. The newborn had the appearance of a "mermaid".

We were unable to obtain parental consent to perform radiographs of the lower limbs in the newborn. Cardiac auscultation appeared normal. The newborn died 1 hour after birth.

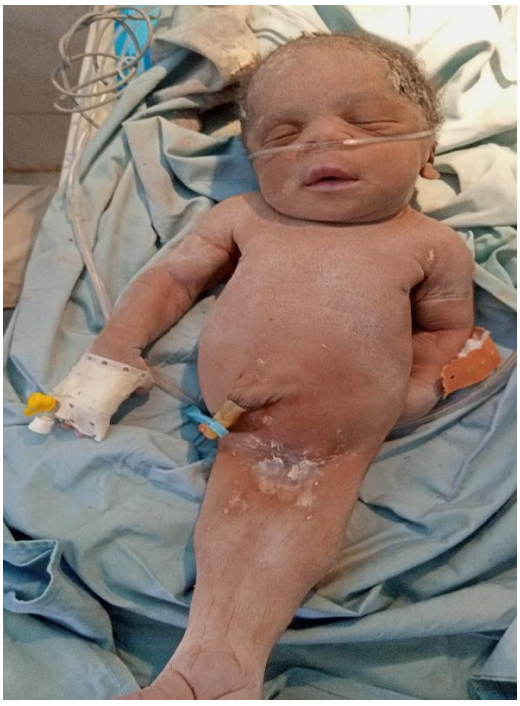

Figure 1: Anterior view

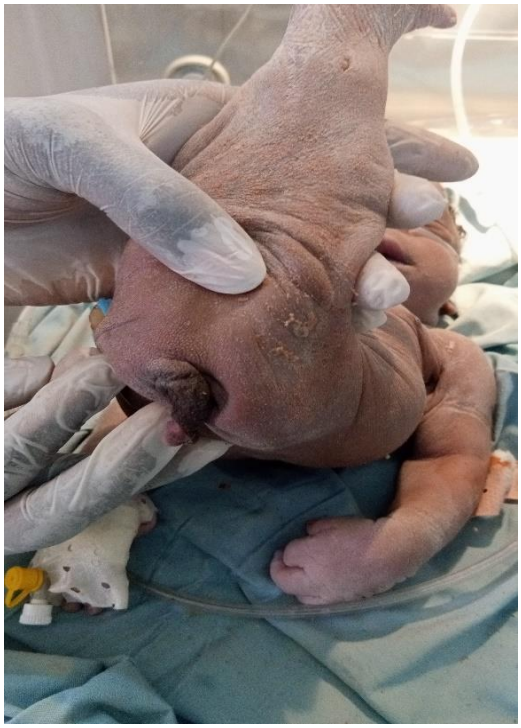

Figure 2: Posterior view showing the anal imperforation

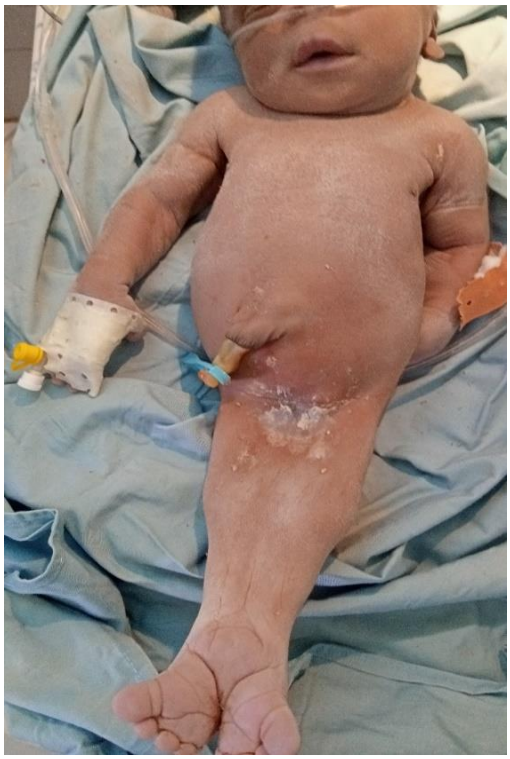

Figure 3: View of the lower limbs with the soles of the feet

DISCUSSION

The etiology of sirenomelia is still controversial. Generally sporadic, it has long been considered to be of environmental origin, with insulin-dependent maternal diabetes as the primary cause, given

that 10 to 15% of affected children are born to diabetic mothers. Some authors have suggested that oxidative stress induced by the accumulation of free radicals could have a teratogenic effect in the fetuses of diabetic mothers. However, only 0.5% - 3.7% of the cases reported in the literature have been associated with maternal diabetes. Maternal diabetes alone does not explain the occurrence of this malformative sequence. In our case, the mother was not diabetic. However, as the pregnancy was not followed up during prenatal consultations, it is not possible to formally exclude gestational diabetes. Other teratogenic factors such as vitamin A, cadmium and lead have been associated with sirenomelia in animal and human models. Since high levels of cadmium and lead have been found in the Lush population (Banza *et al*., 2009), the etiological hypothesis involving these heavy metals cannot be ruled out.

In 1986, Stevenson *et al*., proposed a "vascular flight" theory in which caudal regression results from a detour of blood flow to the caudal part of the embryo via a large vessel derived from the yolk artery originating in the abdominal aorta (Stevenson *et al*., 1986). This detour results in hypo-perfusion of the caudal mesoderm. However, it should be emphasized that this theory alone does not explain the cases associating craniofacial and cardiac malformations. Moreover, it does not give the cause of this malformative sequence.

The theory of blastogenesis defect has also been proposed to explain the pathogenesis of sirenomelia. According to this theory, sirenomelia is a primary defect in blastogenesis that occurs at the end of gastrulation around the third week of gestation. In 1961, Duhamel et al observed a frequent association of sirenomelia with malformations of the caudal region of the embryo (anorectal, genitourinary, lumbosacral), defining a malformative group that he named caudal regression syndrome but today known as caudal dysgenesis. It has recently been suggested that both hypotheses remain valid, as abnormalities of blastogenesis can affect both organs and vessels (Garrido-Allepuz *et al*., 2011).

The recent publication of familial cases has stimulated the search for genetic factors associated with sirenomelia (Gerard *et al*., 2012). Experimental studies in rats with loss-of-function mutations in the signaling sequences of the Bone Morphogenetic Protein (BMP-7) gene or gain-of-function mutations in the signaling sequences of the Retinoic Acid (RA) gene have shown the development of a phenotype similar to that observed in humans, associating lower limb fusion with severe pelvic malformations (Garrido-Allepuz *et al.*, 2012). Thus, it appears that there is a complex, polygenic genetic basis in the pathogenesis of sirenomelia.

In our patient's case, an ultrasonography performed at 21 weeks of amenorrhea during prenatal consultations showed dysplastic and polycystic kidneys. However, early ultrasonographic diagnosis is possible as early as the 9th week of pregnancy despite the severe hydramnios following bilateral renal agenesis observed in the vast majority of cases (Schiesser *et al*., 2003).

CONCLUSION

Sirenomelia is a rare and fatal congenital malformation. This is the first known and documented case in the Republic of Mali. The reluctance of parents about performing additional examinations on both the mother and the child has considerably limited our clinical description.

Conflict of Interest: None

Authors' Contributions

All authors contributed to the writing of this manuscript. All authors have read and approved the final version of the paper.

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