Oeis Syndrome (Cloacal Exstrophy): About Two Cases Treated at the Mother and Child Teaching Hospital in Cotonou (Benin)

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Abstract

Introduction: OEIS syndrome, the most severe form of extrophy-epispadias complex, is characterized by the existence of an omphalocele, a bladder exstrophy, an anal imperforation and a spina bifida. Case Presentation: Two clinical cases admitted at the first day of life in neonatology department are reported; one was born by caesarean section, weighing 2500 g and the other one vaginally with a weight at 1890 g. The first child's mother was a primigravid, primiparous and the second mother was multiparous. Prenatal ultrasound was performed in one case. The clinical examination found two exstrophied hemi-bladders with two productive ureteral meatus, separated by a double intestinal orifice, a manifestation of anal imperforation, omphalocele, spina bifida and lumbar vertebral malformations; which corresponds to a typical form of OEIS syndrome. One of the babies had an associated clubfoot. The karyotype could not be performed. Both were dead; the first in the early postoperative period, the second one at 16 days of life without surgery. Conclusion: OEIS syndrome is a rare condition and represents a therapeutic challenge in developing countries. Promoting prenatal diagnosis is essential.

Keywords

OEIS Syndrome, Cloacal Exstrophy, Newborn, Prenatal Diagnosis

1. Introduction

OEIS syndrome or cloacal exstrophy, previously known as cloacal ectopia, vesi-
co-intestinal fissure or splanchnic extrophy is the most severe form of Exstrophy-Epispidias Complex [1] [2] [3]. The term OEIS complex was first used by Carey et al. in 1978 [2]. Subsequently, it was statistically demonstrated that OEIS complex is a clinically recognized non-random association that, in addition to the four classic malformations (omphalocele, extrophy of the cloaca, imperforate anus, and spine abnormalities) is variably associated with spina bifida, genital abnormalities, renal malformations, symphysis pubis diastasis, and limb abnormalities. The etiology of OEIS complex is thought to be multifactorial, based on the sporadic nature of most of the reported cases. We report 2 cases admitted, in the four months period of the year 2017, in the neonatology department of the University Hospital of Mother and Child Lagune of Cotonou.

2. Cases

2.1. Case 1

He was a male newborn, born vaginally at the 7th month of pregnancy and weighing 1890 g. The mother was 24 years old, primigravid and primiparous, and she has any risk factors, such as teratogenic factors. Genetic analyzes were not done in our context in search of mutations in Homeobox genes (HLXB9). The family and gestational anamnesis were without any particularities. No prenatal visit and ultrasound had been performed.

The newborn inspection, upon his admission at the first day of life, revealed two exstrophied hemi-bladders with two productive ureteral meatus, separated by a short intestine realizing the sign of the elephant’s trunk, an anal imperforation, an omphalocele and a spina bifida (Figure 1). A psychologically shocking mood could be observed in both parents and healthcare providers. The spinal X-ray showed lumbar vertebral malformations (Figure 2). The karyotype had not been done due to the lack of financial means. His mother strongly desired the parietal defect closure but she was abandoned by her family.

A laparotomy performed at the age of 03 months had revealed a common mesentery (Figure 3). The parietal defect closure, the hemi-bladders implantation in the distal colon followed by the opening of its orifice to the skin in the form of a proximal colostomy for urine derivation and a terminal ileostomy for fecal shunt had been carried out [4]. Death occurred three days after surgery in a context of respiratory distress.

2.2. Case 2

The baby was a male newborn, born by caesarean section after 8 months of pregnancy, weighing 2500 g. The 36-year-old mother got pregnant and gave birth 6 times. She had any risk factors, such as teratogenic factors. Genetic analyzes were not done in our context in search of mutations in Homeobox genes (HLXB9). Family and gestational history were without particularity. There was no prenatal diagnosis despite two (02) prenatal visits followed by ultrasound scans.
Clinical inspection of the newborn revealed two extrophied hemi-bladders with two productive ureteral meatus, separated by a short intestine, an imperforate anus, and an omphalocele (Figure 4). There was an equinovarus foot deformity. An abdominal ultrasound revealed a left kidney agenesis. The karyotype could not be performed due to lack of financial means.
The baby was breastfeeding and was waiting for an eventual surgery when he died after 16 days of life of a cardiac arrest.

3. Discussion

The OEIS syndrome’s incidence is between 1/200,000 and 1,400,000 live births [5]. Cloacal exstrophy is considered the most severe and rare ventral abdominal wall defect among the four components of the OEIS complex. It is due to defects of caudal mesodermal migration at around 4 weeks of gestation [6]. Simultaneous failure of convergence of cranial, caudal and two lateral body folds results in the defective ventral abdominal wall. Rupture of cloacal membrane before urorectal septum descent results in exposure of exstrophied cloacal membrane. This cloacal exstrophy prevents the development of proctodaeum manifesting as anorectal anomalies [7]. This caudal dysgenesis also affects the sacral vertebral development [8]. Spine malformations can occur more cranially than the normal lumbosacral level [9].

This may be higher, due to the fact, that many cases are misdiagnosed as omphaloceles, which are the most important component of this malformities complex [5]. In the sub-Saharan African context, and particularly in Benin, congenital malformations are not necessarily notified, and some cases are victims of infanticide [10]. The admission of two cases in the same year in our department, corroborates an underestimated frequency. According to the literature [2] [3], as in our observations, there is a male predominance.

Many risk factors such as teratogenic factors (diazepam, diphenylhydantoin), mutations in Homeobox genes (HLXB9), twinning, in vitro fertilization, alcoholism and gonococcal infection and inbreeding have been listed. Maternal alpha-fetoprotein is also reported to be elevated in OEIS complex [11] [12]. In our observations, genetic investigations could not be performed due to lack of financial means. According to Witters et al. [13], caryotypes are generally normal. Other factors, however, were absent from the mothers’ anamnesis of our cases.
Nevertheless, the young age of the mother in the first case and the advanced age of the mother in the second case are both risk factors for congenital malformation [12].

According to Mallikarjunappa et al. [11], prenatal diagnosis is already possible during second semester ultrasound by identifying the syndrome components. Those components are classified into major criteria (non-visualization of the fetal bladder, infra-umbilical abdominal wall lesion, omphalocele, myelomeningocele) and minor criteria (malformation of the limbs extremities, renal malformations, fetal ascites, narrow thorax, hydrocephalus, single umbilical artery) [7] [9] [14]. Cardiac abnormalities can also be associated [11]. Renal agenesis and a clubfoot deformity were diagnosed for our second observation.

Despite the ultrasound performed by the second mother, the prenatal diagnosis had not been established. The limits of conventional ultrasound are recognized [15].

Fetal MRI is considered an indispensable tool, for the prenatal diagnosis of the OEIS complex [15] [16], as it allows a better visualization of the main structural defects [17] and those associated with them. This type of imaging scan is inaccessible in our conditions.

Advances in prenatal imaging technology make it possible, to diagnose this complex malformation at the prenatal stage; and to benefit from the advantages of prenatal counselling, i.e. better knowledge of the malformation by the parents, preparation for treatment or possible therapeutic termination of the pregnancy, taking into account the severity of the pathology [16]. The discovery of the obvious structural defects of the OEIS syndrome at birth is a psychological trauma for both healthcare providers and parents [4] [18].

The management of this condition requires several major reconstructive surgical interventions, carried out by a multidisciplinary pediatric team including pediatric neonatologists, urologists, orthopedic surgeons, neurosurgeons, geneticists, and endocrinologists; the objective being the acquisition of intestinal and urinary continence on the one hand, and sexual function on the other [4] [16] [18]. This makes it a therapeutic challenge in developing countries with poor technical facilities. The first stages of these aggressive reconstructive steps are the fecal and urinary diversion [4] [16] as performed in the first case.

The prognosis is variable, depending on the severity of the structural defects. Mortality is very high due to obstruction of the urinary tract or a combination of renal, pulmonary, and cardiac complications [18]. These could explain the occurrence of deaths in our cases. According to Neel and Tarabay [18], patient survival also depends on the optimal management and level of advancement of pediatric surgery, and therefore pediatric health care in general [19]. In other words, OEIS syndrome patient survival is an indicator of the quality of care in pediatric surgery [16] [18]. In developing countries like ours, the prognosis is very poor [12] [14], because despite successful surgery, the risk of anal and urinary incontinence remains high [18]. There are views that the physical, emo-
tional, and financial burden is too great to justify correction [4]. On the contrary, Phillips et al. [20] reported 80 cases of OEIS complex treated for 35 years with good results.

4. Conclusion

OEIS syndrome is a rare condition. It is a social drama and a therapeutic challenge in developing countries. The promotion of prenatal diagnosis and genetic counselling is essential, for the management, of such a complex malformation, with a poor prognosis.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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