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Megalourethra: About a Case in Fetal Medicine

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Abstract

The purpose of this case report is to provide an overview of the current knowledge about megalourethra based on a case encountered in our fetal medicine center.

Megalourethra is a rare condition belonging to the group of Lower Urinary Tract Obstructions (LUTOs), which are more common. It can be diagnosed antenatally, especially in male fetuses. Megalourethra is a cystic dilation of the anterior urethra, mainly resulting in pulmonary hypoplasia and renal insufficiency in the neonatal period. Techniques are available (vesico-amniotic shunt, fetoscopy) to remove or overcome the obstacle and thus improve the neonatal and subsequent prognosis. It is essential to look for associated malformations, which are more common in megalourethra than in other conditions of the anterior urethra.

Introduction

Megalourethra is a cystic dilation of the urethra that can be identified antenatally. It results from an obstruction of the distal urinary tract that can affect the bladder and kidneys in the fetus and in the neonate. This condition belongs to Lower Urinary Tract Obstructions (LUTOs). It is a rare entity with few cases reported in the literature, and its descriptions have mostly been included in more general publications concerning LUTOs. Based on a case encountered in August 2021, this article is aimed at describing and better understanding this condition, providing avenues for its management, and addressing neonatal prognosis.

Clinical Case

A 31-year-old patient, G4P1 (one elective abortion, one full-term vaginal delivery of a healthy boy, one early spontaneous miscarriage), with no relevant medical or surgical history, was referred at 19+2 Weeks of Amenorrhea (WA) for megabladder, bilateral ureterohydronephrosis, and suspected hypospadias.

The first trimester ultrasound (12+6 WA) was unremarkable and noninvasive prenatal testing was normal. A *Cytomegalovirus* (CMV) seroconversion was noted in the second trimester of the pregnancy.

The ultrasound performed at 19+2 WA revealed bilateral nephromegaly associated with megabladder and cystic dilation of the penis (Figures 1-3). The amniotic fluid volume was low-normal (Phelan Index: 8.7 cm) and the fetus was eutrophic. There was no evidence of fetal CMV infection on ultrasound. A first vesicocentesis was performed at 19+4 WA and a second one was scheduled at 19+6 WA to evaluate the fetal renal function. The urinary ionogram was pathological and β2-microglobulin was increased in the urine, indicating fetal renal failure. The molecular karyotype performed on the amniotic fluid was normal.

The follow-up ultrasound performed at 21+0 WA showed the same abnormalities as previously described, but with the addition of oligohydramnios (index: 5.2 cm), and hyperechoic renal cortices.

After explanation of the situation and prognosis (see discussion) in our fetal medicine center, the couple requested medical termination of pregnancy. This request was accepted by the medical ethics committee.

The feticide (intracordal sufentanil and intracardiac potassium chloride) and expulsion (induced by mifepristone at D-2 and misoprostol) occurred at 22+0 WA. The patient gave birth to a male child, 520 g and 29 cm.

As the couple did not want an autopsy, no exhaustive post-mortem examination was performed. We noted an enlarged penis (Figure 4), no palpable erectile body and an unremarkable abdominal wall. Macroscopically, the urinary meatus appeared permeable (Figure 5). These findings, although

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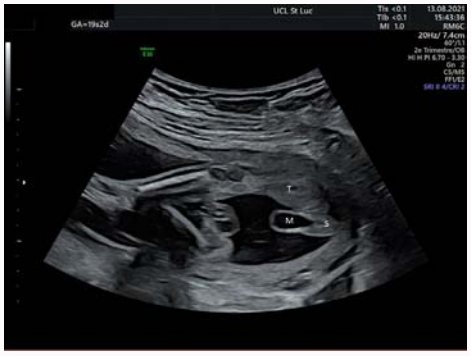

Figure 1: M: Megalourethra, S: Scrotum, T: Thigh.

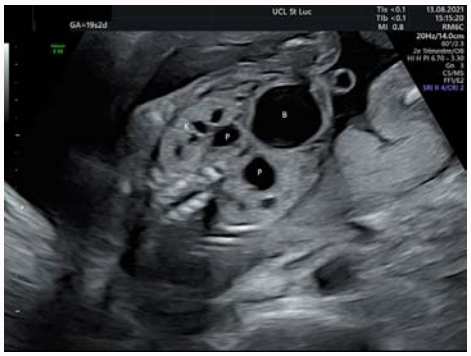

Figure 2: B: Bladder, K: Kidney, P: Renal Pelvis.

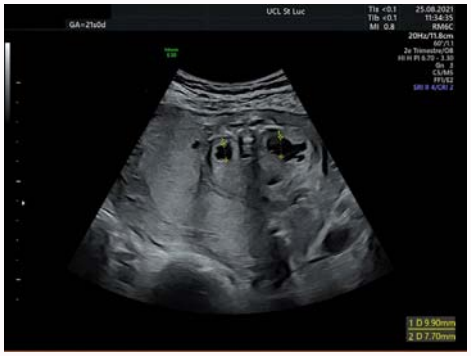

Figure 3: Bilateral kidney dilation.

not comprehensive, are consistent with a diagnosis of primary megalourethra (Table 1).

Discussion

Definition

The first clinical use of the term "megalourethra" dates from 1955 and the first prenatal ultrasound diagnosis from 1989 [2,3].

Congenital megalourethra results from an aplasia of penile erectile tissue causing abnormal dilatation of the anterior urethra due to the lack of supporting tissue [4-7]. Urinary stasis is therefore the consequence of the swelling of the urethra that lacks support. The swelling has a valve-like effect and prevents urine from flowing out [2]. These cases can be considered as primary megalourethras [6,8]. In other so-called secondary cases, urethral dilation is related to a distal urethral obstruction that may result from a valve phenomenon or urethral or preputial stenosis [2,6,8]. As a result, the urethra

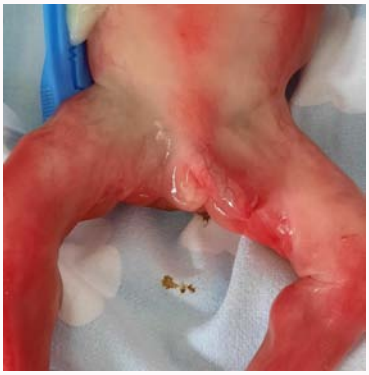

Figure 4: Penile Dilation.

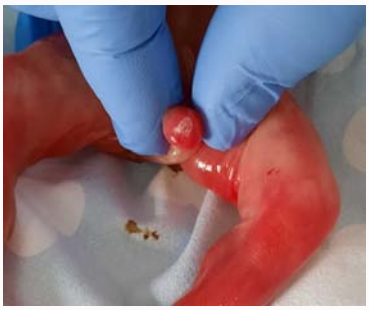

Figure 5: Urinary Meatus.

Table 1: Fetal urine biochemistry.

Vesicocentesis	19+4 WA	19+6 WA	Normal values [1]
Osmolality (mOsm/L)	252	245	<200
Sodium (mmol/L)	122	118	<100
Potassium (mmol/L)	5	5	
Chloride (mmol/L)	98	95	<90
Calcium (mmol/L)	2	2	<0.95
β2-microglobulin (mg/L)	>4	18.345	<0.3

Table 2: Differential diagnosis.

Cystic dilation of the penis - differential diagnosis
Anterior urethral valve
Preputial stenosis
Urethral stenosis
Primary megalourethra
Prune-Belly syndrome
Umbilical cord loop in the pelvic region

enlarges, its wall thins, and the erectile bodies become progressively hypoplastic [6]. Some authors describe the primary forms as fusiform, i.e. with absence of all the penile erectile bodies, and the secondary forms as scaphoid, where only the corpus spongiosum is a plastic [9]. This morphological distinction can be seen after birth [9-11].

Epidemiology

Urogenital tract anomalies are the third most common non-chromosomal anomalies. It is estimated that 3.8 children out of 1,000 births are affected by this type of conditions, which makes up approximately 20% of the anomalies diagnosed antenatally. Megalourethra is a rarity among these anomalies [12,13].

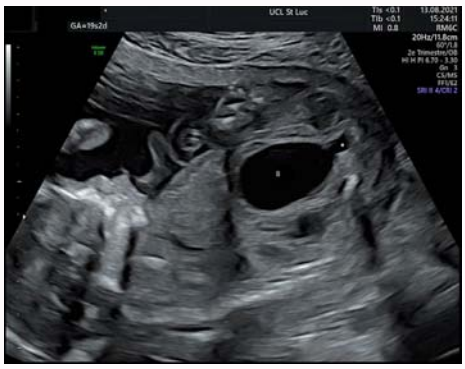

Figure 6: Keyhole Sign; B: Bladder, *: Posterior Urethra.

Embryology

Congenital megalourethra is characterized by the absence of penile mesodermal tissue. The cloacal membrane is composed of two layers, the endoderm and the ectoderm. The mesodermal mesenchyme migrates towards the cranial pole of the cloacal membrane and forms the genital tubercle between the cloacal membrane and the umbilicus. On either side of the cloacal membrane, the mesoderm forms the urogenital folds. More laterally, the labioscrotal swellings are formed. These different structures are the embryonic precursors of the male and female external genitalia.

In response to androgen production in the male fetus, the mesoderm of the genital tubercle elongates and enlarges to form the cavernous bodies and glans penis. At the same time, the urethra is formed by tubularization of the endoderm, while the skin and prepuce develop from the ectoderm. The genital swellings will fuse to form the scrotum.

In the case of secondary megalourethra, the embryological origin of the anomaly is thought to be a defect in the epithelial canalization of the glandular urethra [2,6,14,15].

Antenatal description

On ultrasound, the male fetus exhibits an ovoid dilation of the urethra extending to its distal end. LUTO signs can also be found on antenatal examination: megabladder, ureteral dilation, and possibly hydronephrosis with changes in renal echostructure and oligohydramnios [6,8,12,16]. In some cases, a posterior urethral valve with the "keyhole sign" Figure 6 is visible on ultrasound, reflecting dilation of the posterior urethra, and may be the consequence of a severe stenosis of the anterior urethra [8,12].

On ultrasound, cystic dilation of the penis should not be mistaken for a loop of the umbilical cord near the pelvic region of the fetus; the distinction is made by using color Doppler [7,17] (Table 2).

Associations

Megalourethra is a rare sign of the Prune-Belly syndrome that combines bladder hypertrophy, ureteral dilatation and abnormalities, hydronephrosis, bladder activity disorders, aplasia or hypoplasia of the muscles of the anterior abdominal wall, and cryptorchidism. Although the etiology is not clearly established, it seems that the primum movens is urethral atresia and the resulting obstruction of the distal urinary tract [18].

In their series of 10 cases, Amsalem et al. [8] described a VACTERL association; four fetuses had clubfoot and two had anal atresia. Some authors have associated megalourethra with anorectal malformations, esophageal atresia, VATER association, trisomy 21, or dental, intestinal, and musculoskeletal malformations [17]. Therefore, there might be some malformative associations to look for in case of megalourethra, while other anomalies of the anterior urethra are isolated in most cases [5].

Management

Vesico-amniotic shunt consists in placing a catheter between the fetal bladder and the amniotic cavity. This allows the bladder to empty, to artificially eliminate the lower urinary obstruction, and to decrease the pressure in the upper urinary tract [19,20].

It is also possible to perform a fetoscopy with cystoscopy to make the diagnosis and, if necessary, to perform a meatotomy or a valve removal, or to place a urethral stent [6,12,13,21]. While the technique is more complicated, its benefits compared to the shunt and its effectiveness cannot be ascertained due to the small number of cases [13,22].

These treatments are based on case series concerning LUTOs in general, some of which have included a few cases of megalourethra.

Neonatal consequences

The consequences of megalourethra are the same as those of LUTOs, namely: Pulmonary hypoplasia due to oligohydramnios, progressive renal failure due to vesico-ureteral reflux, micturition disorders, incontinence, recurrent urinary tract infections, and sexual dysfunction [2,5,16].

The prognosis is worse in case of bilateral ureterohydronephrosis, renal cysts, and oligo- or even anamnios. This is all the more true if the onset is early during pregnancy [4,12]. Some authors have described the primary forms as having a poorer prognosis [9].

Postnatal renal function is difficult to predict. Fetal kidney appearance does not seem to correlate with fetal urine biochemistry, and both ultrasonographic kidney appearance and renal biochemistry should be considered in assessing fetal renal function [23]. However, fetal urine biochemistry by vesicocentesis is not unanimously accepted for assessing fetal renal function, and even less for predicting it postnatally [24].

Long-term consequences

Studies tend to show an increase in overall survival in the case of prenatal intervention but they do not provide strong evidence of improvement in neonatal kidney function, let alone in the longer term [13,21,24,25].

Penile dysfunctions result in impotence, extensive dilation of the penis preventing correct vaginal intromission, and during micturition, urethral congestion and penile dilation. Few adult cases have been described in the literature. Surgical management is possible to obtain an acceptable aesthetic and functional result [26-28]. Antenatal removal of the obstacle allows recovery of a normal penile morphology, which would suggest that penile functions are better preserved and that reconstructive surgery is not necessary, but this could not be proven [9].

Conclusion

Megalourethra is a rare condition that can easily be detected in the antenatal period, although the definitive diagnosis is more akin to a diagnosis of exclusion for its primary form. In such cases, it is essential to perform a complete fetal morphological examination

in order to search for associated conditions or syndromes and repercussions on the urinary tract. As in all forms of LUTO, the neonatal prognosis depends on the severity of renal insufficiency and pulmonary hypoplasia, two parameters that are difficult to assess antenatally and even more difficult to predict postnatally. Therapeutic options exist to allow emptying of the urinary tree and hopefully preserve or improve postnatal renal and pulmonary function by maintaining sufficient diuresis and amniotic fluid volume.

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