

# Megalourethra: a case report managed with a single intrauterine bladder aspiration

## Abstract

Congenital megalourethra is a rare mesenchymal anomaly of the male urethra, characterised by severe dilatation of the penile urethra due to congenital absence of the corpus spongiosum and/or corpus cavernosum. Postnatal complications include voiding and erectile dysfunction as well as renal insufficiency and pulmonary hypoplasia. We present a unique case of congenital megalourethra diagnosed prenatally in the early second trimester. The parents opted to continue pregnancy and vaginal delivery of a live neonate occurred with a favourable outcome.

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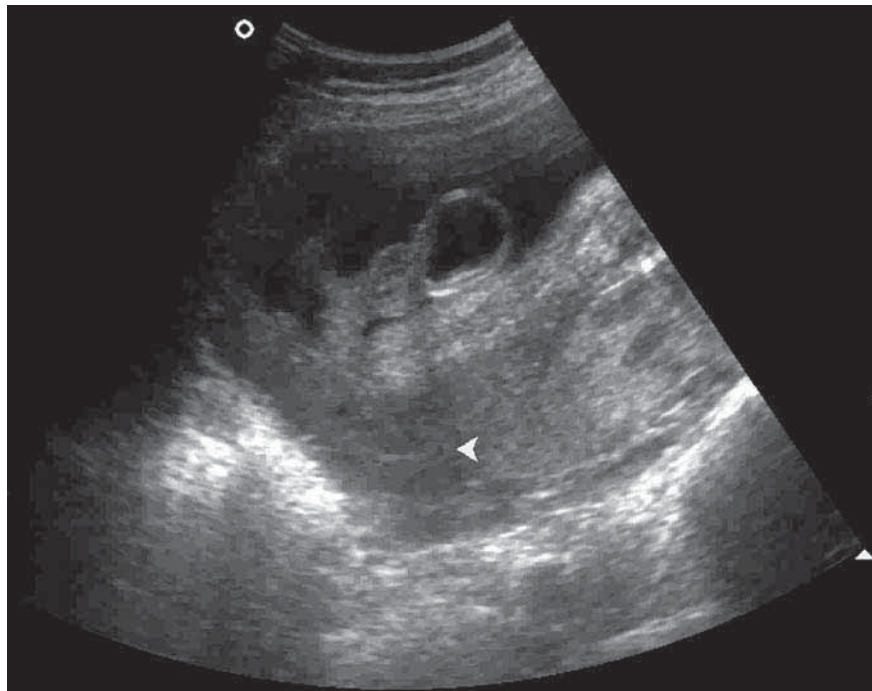
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**Figure 1:** Antenatal ultrasound image at 20 weeks gestation, illustrating megalourethra which is a cystic structure ballooning distally to the penile portion of the urethra.

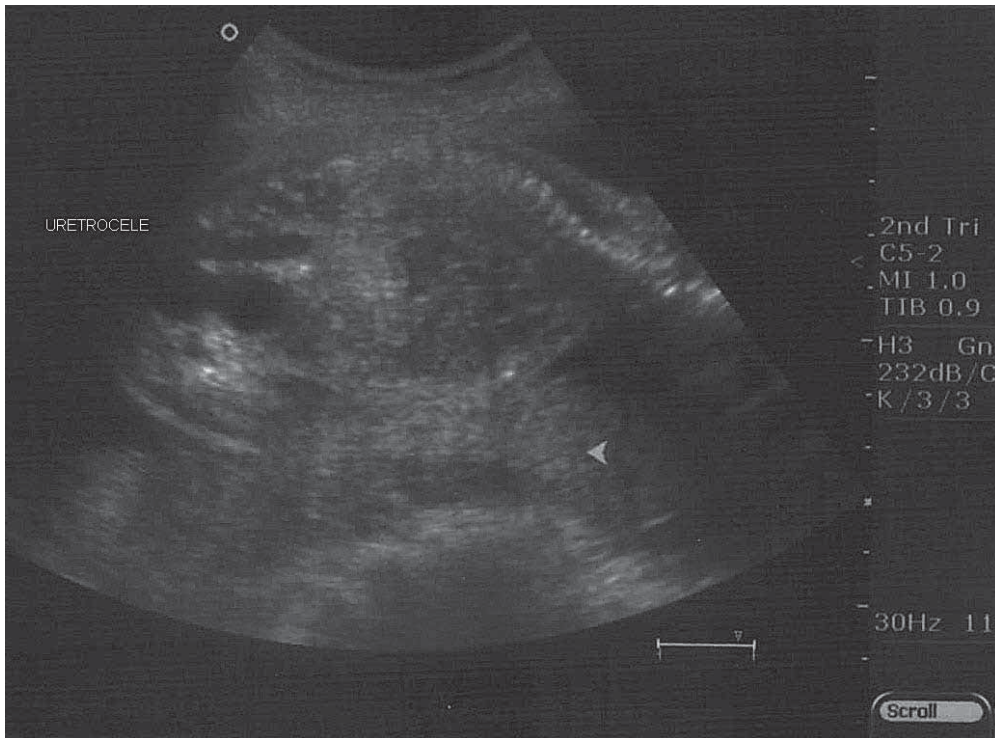
## Introduction

Congenital megalourethra is a rare form of functional lower urinary tract obstruction (LUTO).<sup>1</sup> It was first used in 1955 to describe an infant with renal insufficiency that had an enlarged penis which dilated during voiding.<sup>2</sup> It is defined as a diffuse dilatation of the anterior urethra due to absence of the development of erectile tissue of the penis.<sup>3</sup> Congenital megalourethra is classified into two variants: scaphoid and fusiform.<sup>4</sup> The first prenatally reported case was in 1989,<sup>5</sup> and after that, a limited number of cases have been reported. We present a unique case of congenital megalourethra diagnosed prenatally in the early second trimester. The parents opted to continue

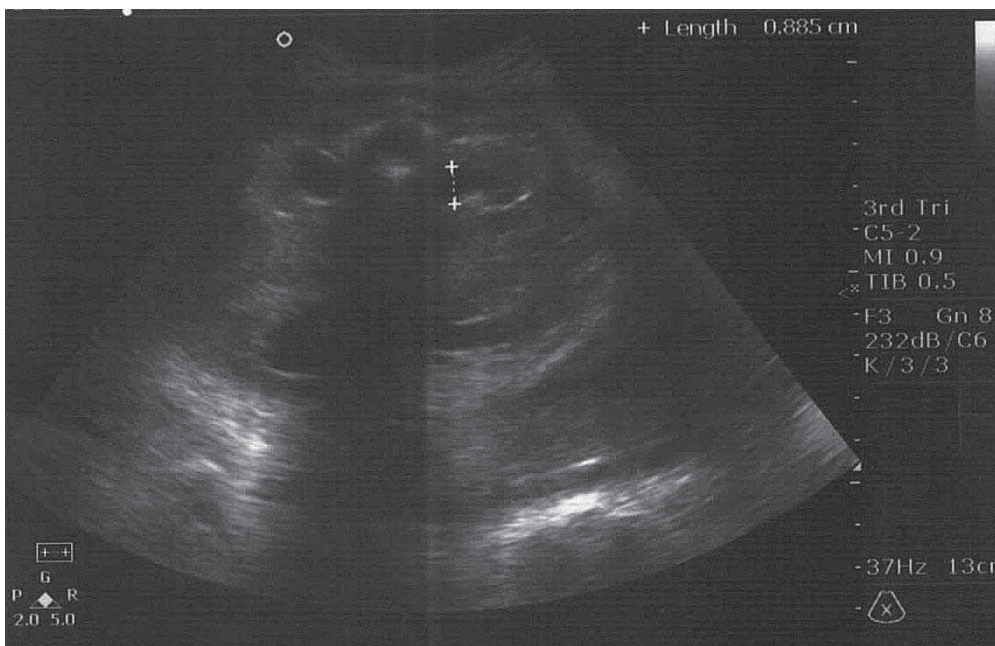
pregnancy and vaginal delivery of a live neonate occurred with favourable outcome. We report the antenatal and postnatal findings.

## Case report

A 27-year-old woman G2 P1 who was referred at 17 weeks gestation to our fetomaternal unit because of the presence of foetal abdominal cyst for further management. The mother's past medical and family history were non-contributory. Her first pregnancy was uneventful with a vacuum delivery; the outcome was a live healthy female baby with birth weight of 3.3 kg. In this current pregnancy, transabdominal sonography revealed a single foetus, measuring



**Figure 2:** Antenatal ultrasound image at 20 weeks gestation illustrating unilateral ureterocele with key hole sign of the urinary bladder.



**Figure 3:** Antenatal ultrasound image at 36 weeks gestation illustrating bilateral pyelectasis.

16 weeks 2 days with good foetal heart. The foetus had severe oligohydramnios, megacystis with key hole appearance and mild bilateral pyelectasis. The foetal gender was difficult to be identified. The family was counselled about the foetal condition and opted to delay foetal karyotyping. One week later, the foetus has anhydramnios, foetal bladder aspiration was performed as a temporary relief and as a source of foetal cells for karyotyping. The mother was booked in two weeks time for follow up. Surprisingly; transabdominal sonography revealed good amniotic fluid around a male foetus. The foetal penis was large and elongated and the urethra was dilated along the foetal penis and ballooned distally (Figure 1). Bilateral ureterocele with

mild pyelectasis were also seen. The bladder was not dilated, but had a sonographic appearance of a key hole sign (Figure 2). The diagnosis of congenital megalourethra was made. The family was counselled about the future urological problems and opted to continue with pregnancy regardless of the outcome. Foetal karyotype turned out to be a normal male foetus with 46 XY. Detailed foetal echocardiography was performed at 24 weeks gestation and revealed a normal heart. Mild bilateral pyelectasis, bilateral ureterocele and mild thickening of the urinary bladder wall were all seen during our follow up without significant changes. The cystic dilation of the megalourethra measured (2.5 x 3.0 x 2.0 cm) all through pregnancy, however the urethral wall





**Figure 4:** Just after birth, the neonate had a large phallus with redundant skin over its ventral surface with small scrotum.

started to thicken in the late third trimester.

The foetal growth was approximately on the 40% and the amniotic fluid volume was normal until 36 weeks gestation, then foetal growth started to slow. At 38 weeks gestation, the foetus was in cephalic presentation, the growth was on the 14%. The amniotic fluid index was 7.0 cm. Pyelectasis increased (the right kidney pelvis measured 1.2 cm and the left one 0.9 cm) (Figure 3). The foetal kidneys were not enlarged but started to be echogenic. The foetal abdomen was not distended. Delivery was induced, the mother had uneventful vaginal delivery of a male neonate with birth weight of 2.7 kg. The Apgar scores were 7 and 9 at 1 and 5 min. The neonate was transferred to the normal nursery for further follow up.

Physical examination of the neonate showed normal abdominal wall musculature without dysmorphic features. Examination of the external genitalia showed right inguinal hernia and a large phallus with redundant skin over the ventral surface, which ballooned during micturation (Figure 4). The neonate had poor urine stream in the form of dribbling from a normally located meatus, in addition compression of the ventral surface caused urine also to dribble.

His creatinine on the first day was 0.73 mg/dL. It increased to 1.4 mg/dL on the second day, and to 2.45 mg/dL on the third day. His abdominal ultrasound revealed echogenic kidneys with loss of corticomedullary differentiation and moderate hydro-ureteronephrosis. The urinary bladder appeared distended. Urgent vesicostomy was performed on third day of life due to the rapid rise in creatinine. Intra operative findings revealed a thickened bladder wall. The neonate then had a transient diuresis and his serum creatinine stabilized at around 2 mg/dL. Cystogram was performed through the vesicotomy which revealed dilated tortuous ureters (Figure 5). He had right herniotomy and right orchidopexy at the age of 2 weeks. His course was also complicated by two hospital admissions, due to pyelonephritis that were treated with intravenous antibiotics. His weight now is below the fifth centile. He is now followed in the clinic for his renal impairment, his creatinine blood levels are stabilized at around 1.6 mg/dL. He also developed acidosis, hypocalcaemia, hyperphosphataemia, and anaemia. He is now on sodium bicarbonate, calcium supplements, phosphate binder, erythropoietin and prophylactic antibiotics.



**Figure 5:** Postnatal cystogram. It showed bilateral vesicoureteral reflux with bilateral hydronephrer.

## Discussion

Megalourethra results from a congenital deficiency of the mesodermal tissues of the phallus due to defective migration, differentiation or development of the erectile tissue.<sup>6</sup> It is classified into two types; fusiform and scaphoid. The fusiform is more common and is due to maldevelopment of corpus spongiosum in the anterior urethra, causing bulging of the ventral urethra. The scaphoid type is a severe maldevelopment due to deficiency of corpus cavernosum as well as corpus spongiosum, causing circumferential expansion of urethra. Although these two types are described, intermediate forms have been reported and congenital megalourethra can be seen as a spectrum of disease.<sup>3</sup> The fusiform type has a higher incidence of oligohydramnios and a worse prognosis compared to the scaphoid type, which is consistent with a better survival rate.<sup>1</sup> This is supported by the literature which reports a mortality of 13% in the scaphoid type and 66% in the fusiform type.<sup>3</sup> This anomaly can be associated with other urinary tract anomalies including dysplastic kidneys, vesicouretric reflux, undescended testicles, prune belly, megaureter, hypospadias, posterior urethral valves, anorectal anomalies and other systems abnormality.<sup>7</sup> In babies who were born alive, the main morbidities complicating the postnatal life are not different from cases with lower urinary tract obstruction (LUTO) regardless of the aetiology; pulmonary hypoplasia as a result oligohydramnios which can result in neonatal death, and chronic progressive renal failure that can result in end stage renal disease.<sup>1</sup>

Promsonthi P, *et al.* reported a case in May, 2010 and reviewed all the previous prenatally reported congenital megalourethral cases.<sup>8</sup> There were 28 cases from 20 articles. The median gestational age at diagnosis was 20 weeks (ranges 12–34 weeks gestation). Most of the cases were between 18–24 weeks gestation (In one case, the gestational age at diagnosis was not available<sup>9</sup>). All cases had sonographic features of genitourinary abnormalities, lower urinary tract obstruction or prune belly syndrome. Twelve cases (42.9%) had anomalies of other organs. These included imperforated anus/anal atresia which were the most commonly associated abnormality (7/28), single umbilical artery, vertebral and limb abnormalities were found in 4/28 cases, three cases (3/28) had cardiac abnormalities (two hypoplastic left heart syndrome, and one case with ventricular septal defect) and three other cases (3/28) had esophageal atresia/tracheoesophageal fistula. Sixteen cases (57%) had normal male karyotype. Regarding the outcome of these cases, two cases (7.14%) resolved completely before birth without any residual finding at delivery.<sup>10,11</sup> Sixteen cases (44.8%) had favourable outcomes at discharge. There were four (11.2%) neonatal deaths and seven (19.6%) pregnancy terminations. One article did not report the postnatal outcome.<sup>10</sup>

A subsequent decade-long review by Amsalem H, *et al*<sup>1</sup> highlighted outcomes from 10 cases with foetal megalourethra. The median gestational age was 19 weeks (ranges from 13–24 weeks gestation). All cases presented with a distended bladder and megalourethra. Nine cases (90%) had dilated ureters and hydronephrosis, nine cases (90%) had normal amniotic fluid volume at the time of diagnosis, three cases subsequently developed severe oligohydramnios. Associated anomalies were found in 50% of the cases, (one case with VACTERL association, two cases with anal atresia and four cases with bilateral club

feet). All cases had normal karyotype. Three pregnancies were terminated and seven continued. Of the seven live born, one died in the neonatal period because of pulmonary hypoplasia. The other six infants who were alive, have a dysfunctional urethra, three of them have renal dysfunction, two of whom have end-stage renal disease.

Foetal bladder shunting has been used in LUTO to overcome the functional obstruction.<sup>13</sup> Our foetus presented with severe oligohydramnios due to megacystis (LUTO) in the early second trimester (17 weeks gestation), which progressed to anhydramnios in one week time. Foetal bladder aspiration was performed once at 18 weeks gestation as a source of foetal cells for karyotyping and as a temporary relief of the obstruction. In two weeks time the amniotic fluid volume was within normal range, and at that time we were able to visualise the foetal gender and to diagnose megalourethra. This is rather similar to only one case that was reported by Amsalem H, *et al.*<sup>1</sup> when bladder aspiration and vesico-amniotic shunt was inserted at 17 weeks gestation when oligohydramnios developed. This resulted in improvement of the of amniotic fluid volume and a favourable outcome after birth. However in our case, only a single bladder aspiration was enough to improve the amniotic fluid volume all through pregnancy without foetal bladder shunting and the neonate had a favourable outcome after birth. The improvement of amniotic fluid volume after bladder aspiration can be explained by the improvement of bladder muscle function with advancement in the gestational age. This may suggest that insertion of permanent vesico-amniotic shunt is not mandatory for the treatment of this condition. In summary, congenital megalourethra is a rare form of lower urinary tract obstruction. In approximately half of the reported cases, urogenital abnormalities were isolated. Congenital megalourethra should be considered in all male fetuses presenting prenatally with megacystis. The survival of those patients depends on the severity of renal damage and lung hypoplasia. In isolated cases, a single bladder aspiration could be enough to improve the amniotic fluid volume all through pregnancy and to get a rather good outcome after birth. However the quality of life in terms of urination and sexual function depends on the ability to correct the urologic dysfunction.

## Acknowledgment

We are grateful for the parents for allowing us to share their experience with the world, hoping for better future for all parents to be.

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