

Research Article

Congenital megalourethra: spectrum of presentation and insights in embryology and management

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ABSTRACT

Background: Congenital megalourethra is a rare urogenital mesenchymal anomaly of the male anterior urethra and erectile tissue of penis. The disease is characterized by severe dilatation of the penile urethra due to congenital absence of the corpus spongiosum and corpus cavernosum. It presents as two types, a milder scaphoid type and severe fusiform type. The prognosis is related to the degree of associated renal impairment and the severity of accompanying congenital anomalies. Some isolated case reports are available but very few case series have been published till date. This is one such rare case series.

Methods: Since July 2010, we have encountered three cases of congenital megalourethra in our department. All these cases had different patient profile, clinical manifestations and types. Two patients had isolated megalourethra which is very rare entity while one patient had megalourethra with associated prune belly syndrome. All the patients were investigated thoroughly. Micturating Cystourethrogram (MCU) is required to confirm the diagnosis.

Results: Reduction urethroplasty was performed in all the three patients successfully. All patients passed urine in single stream without any urethral dilatation. There were no complications.

Conclusions: Congenital megalourethra is a rare disease which warrants proper diagnosis and timely intervention for optimum outcome. Due to paucity of literature many cases are misdiagnosed and incorrectly treated the embryology, presentations and current management of this rare disorder are discussed herewith along with review of pertinent literature.

Keywords: Congenital megalourethra, Scaphoid, Fusiform, Reduction urethroplasty

INTRODUCTION

Megalourethra is a rare congenital mesenchymal anomaly of the male anterior urethra characterized by a non-obstructive dilatation of the penile urethra.^{1,2} It is defined as diffuse dilatation of the anterior urethra due to absence of development of the erectile tissue of the penis. This anomaly affects the anterior part of urethra, and usually cause abnormal size and shape of the penil shaft, especially while voiding.³ The first case of congenital megalourethra was reported by Obrinsky, who also described its association with 'prune belly' syndrome.⁴ Nesbitt in 1955 defined it as "a congenital dilatation of

penile urethra without distal obstruction".⁵ Less than 100 cases have been reported in literature till now.^{2,3,6} Dorairajan classified congenital megalourethra into two types based on findings of urethrography.⁷ The more common scaphoid type with a deficiency of the corpus spongiosum presents as bulging of ventral urethra. The fusiform type, there is deficiency of both corpus spongiosum and cavernosum which is seen as circumferential expansion of urethra.⁶ The prognosis is related to the degree of associated renal impairment and the severity of accompanying congenital anomalies. We are presenting our series of three cases with different presentations that were treated successfully. There have

been isolated case reports of this rare anomaly but case series are extremely rare. The aim is to provide a clear picture about embryology, manifestations and management which are of paramount importance in treatment of this often misdiagnosed rare anomaly.

METHODS

Since July 2010, we have encountered three cases of congenital megalourethra in our department. All these cases had different patient profile, clinical manifestations and types. Two patients had isolated megalourethra which is very rare entity while one patient had megalourethra with associated prune belly syndrome. These patients were being misdiagnosed and treated inappropriately.

After presentation to us, all the patients were investigated thoroughly by routine hemogram, renal function tests (RFT), Ultrasonography (USG), Intra venous urography (IVU) and Micturating Cystourethrogram (MCU). The details of clinical picture and investigations of all the patients are mentioned in Table 1.

A reduction urethroplasty was performed in all the three patients. Patients were given regional anesthesia in the form of caudal block along with sedation. After adequate preparation glans stitch was taken and penis degloved by circumferential incision. After complete degloving of the penis the urethra was opened longitudinally over the swelling. The excess redundant tissue was excised and urethroplasty performed using vicryl 6-0 continuous sutures over appropriate size infant feeding tube (IFT no 10 for case A and no 6 for cases B and C) (Figure 1C). The urethroplasty was strengthened by a second layer using pedicled inner prepuce skin flap. Subsequently skin flaps were sutured using vicryl 5-0 sutures. Adequate haemostasis was achieved. Light dressing was given so as to avoid any excessive pressure over the urethroplasty and skin flaps and at the same time avoid any hematoma formation. The catheter was removed on the 14th POD.

RESULTS

Reduction urethroplasty was performed in all the three patients successfully by aforementioned technique. The redundant urethra was excised and urethroplasty was performed over a catheter. The catheter was removed after two weeks (14 days). All patients tolerated the procedure well. All three patients passed urine in good stream without any penile deformity. [Fig1d]. Regular follow-up up to 1 year showed no recurrence of penile swelling with normal voiding pattern and normal bladder and upper tracts on USG. There were no fistulas, infections or any other complications.

DISCUSSION

Congenital megalourethra is a rare form of functional lower urinary tract obstruction (LUTO) caused by primary or secondary agenesis/hypoplasia of the penile corporal tissues. The exact embryological cause of congenital megalourethra is not clearly understood. The most commonly held theories propose a defect in the migration, differentiation, or development of the mesenchymal tissues of the phallus.² Another hypothesis is that delayed or deficient canalization of the glandular urethra may be associated with maldevelopment of the corpus spongiosum and corpora cavernosae.^{2,8} Mild delays with earlier and more complete canalization may be associated with scaphoid while longer delays with later and less complete canalization and fusiform megalourethra.⁹ Owing to the poor development of supporting erectile tissue there is stasis of urine causing functional obstruction.⁶

Congenital megalourethra has been classified into two types:

(a) Scaphoid type

A milder form that involves the urethra and corpus spongiosum alone where during micturition the urethra dilates in a scaphoid (boat shaped) fashion as the dorsal aspect is well supported by the spongiosum.

(b) fusiform type

More severe form as corpus spongiosum and corpora cavernosa both are involved resulting in fusiform (spindle) dilatation of the phallus during voiding. However, this classification is neither pathological nor etiological and does not help in prognostication.

Recently, Amsalem et al classified the condition into (a) primary (or ex-vacuo), caused by absence or hypoplasia of the corpora spongiosa and cavernosa, associated with normal amniotic fluid volume, usually preserved renal function and better outcome, and (b) obstructive (secondary), which results in oligohydramnios with a higher incidence of renal failure, pulmonary hypoplasia and thus mortality.¹⁰ In both types the corpora are hypoplastic (either as a result of an initial absence of the corporal tissue in the primary type or secondary to pressure in the obstructive type). The 'primary' type can become obstructive if blocked by debris and can result in oligohydramnios later in gestation. A mortality rate of 13% in the 'primary/scaphoid' type and 66% in the 'secondary/fusiform' type has been reported in the literature.²

Various congenital anomalies are associated with megalourethra mostly related to urogenital system and sometimes with other organ systems as well. Jones et al in a review have reported, associated congenital anomalies are seen in 80% of scaphoid type and 100% of

fusiform type.² The commonly described genitourinary anomalies include renal dysplasia-hypoplasia, hydronephrosis, hydroureter, vesicoureteric reflux, prune-belly syndrome, urethral duplication, megacystis, hypospadias, posterior urethral valves, and undescended testes. Other system anomalies including VATER (vertebral, anal atresia, trachea-oesophageal fistula, and renal anomalies) and VACTERL (vertebral, anal atresia, cardiac, tracheoesophageal fistula, renal, and limb deformities) are described.⁶

Benacerraf et al, in 1989, were the first to report this condition prenatally.¹¹ Congenital megalourethra can be diagnosed prenatally if a detailed examination of the fetal genitalia is performed specially in fetuses with urinary tract dilatation. The earliest diagnosis reported was at 13 weeks in a fetus with multiple malformations.¹² However, most cases are detected in the second trimester, when

signs of lower obstructive uropathy viz. distended bladder with or without hydroureter and hydronephrosis and a cystic structure in the perineal region that may be detected in the presence of normal, decreased, or even increased amniotic fluid volume.¹³ It is important to be cautious on USG as a thin-walled distended urethra can easily be mistaken for a loop of umbilical cord. Color Doppler is very helpful in differentiating these two entities.^{10,16} fusiform megalourethra is more likely to be detected ante-natally than scaphoid megalourethra as the former is invariably associated with signs of urinary tract dilatation.¹³ Prenatally diagnosed megalourethra has to be thoroughly evaluated for associated congenital anomalies and followed up serially by ultrasound for amniotic-fluid index, urinary bladder volume, and upper tracts assessment along with fetal echocardiography.⁶

Table 1: Clinical and radiological picture of all three cases.

Serial no	Patient A	Patient B	Patient C
Age /Sex	11 years/ Male	18 months /male	2 years/male
Presenting complaints	Dribbling micturition after the normal act of micturition with wetting of underpants	history of dribbling micturition	ballooning of distal half of penis during micturition dribbling of urine for some time after micturition
Misdiagnosed as	nocturnal enuresis and urinary incontinence	-	Phimosis with nocturnal enuresis
Clinical examination	cystic swelling in the distal part of the penis with urine expressible from the swelling [fig 1a]	cystic swelling over the anterior part of the urethra	meatal stenosis with widening of distal penis. Prominent ballooning of penis while micturition[fig 2]
Associated anomalies	-	Prune belly syndrome	-
RFT	Normal	Elevated Serum Creatinine	Normal
USG Abdomen	Normal	Renal dysplasia with HN + HU	Normal
IVU	Normal	HN+HU and dilatation of pelvicalyceal system	Normal
MCU	Massively Dilated anterior urethra suggestive of megalourethra with grossly trabeculated bladder.[Fig 1 b]	Megalourethra with grossly trabeculated bladderwith bilateral grade IV VUR	Mildly Trabeculated bladder
Surgery	Reduction urethroplasty	Reduction urethroplasty	Reduction urethroplasty
Remarks	Good outcome	Surgery for bilateral UDT bilateral VUR performed later Satisfactory outcome	Good outcome

In the post natal period, the diagnosis can be confirmed by a micturating cystourethrogram (MCU), which shows a massive dilatation in the anterior urethra. In view of

associated anomalies, the workup of megalourethra should include renal function tests and imaging of upper and lower urinary tracts like intravenous Urography

(IVU) and USG.⁶ The status of upper urinary tract determines the ultimate outcome.¹ Oligohydramnios reflects substantial renal impairment and portends poorly, especially in the second trimester with the risk of pulmonary hypoplasia.¹³



Figure 1A: Cystic swelling in the distal part of the penis.

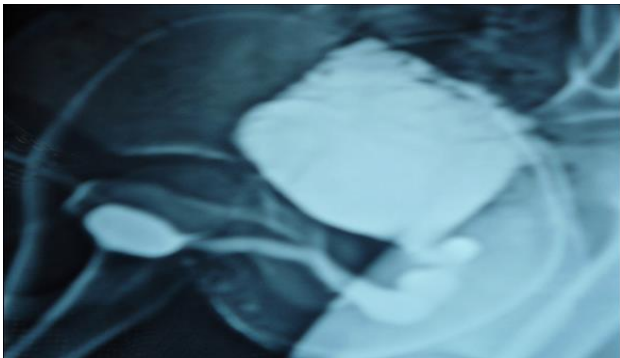


Figure 1B: Congenital megalourethra with a grossly trabeculated bladder on MCU.

The etiology of renal dysfunction associated with megalourethra is not certain. True megalourethra is characterized by the absence of an anatomic urinary obstruction. Albeit, there is a functional obstruction to urine outflow as the urethra “balloons” with fetal voiding, with increasing pressure eventually resulting in efflux of urine.¹³ This functional obstruction may also act as a valve like flap mechanism in some cases, producing intermittent mechanical obstruction to the urine stream through the glandular urethra. Few cases associated with posterior urethral valves (PUV) have also been reported, but this seems more to be a concomitant anomaly rather than etiology.^{14,15} Some authors believed that fetal uropathy due to megalourethra may carry a better prognosis than PUV due to a non-obstructive form of urinary tract dilatation and mildly elevated intrarenal pressure, which probably will not result in permanent renal fibrosis and dysplasia.^{16,17} Spulveda et al suggests that the concomitant obstructive uropathy, regardless of its etiology, may severely damage the fetal kidneys and lead to renal insufficiency in most cases.¹³

The differential diagnosis of congenital megalourethra includes a cystic abdominal wall mass, an umbilical cord

loop, or a cyst, the latter 2 entities can be distinguished with color Doppler sonography.⁹ Urethral anomalies mimicking megalourethra like urethral atresia, a web, duplication, and diverticulum are also considered in the differential diagnosis.⁹

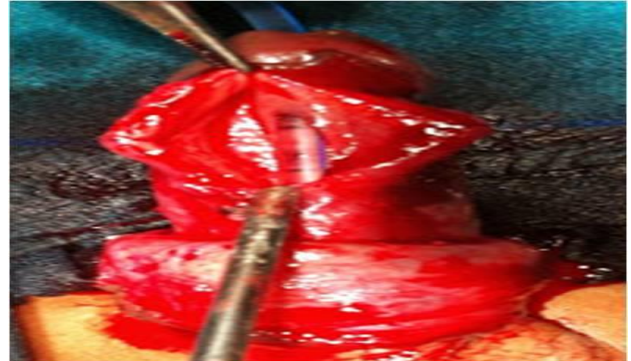


Figure 1C: Reduction urethroplasty.

The goal of antenatal diagnosis is to provide parents with accurate diagnosis and prognosis allowing them to take decision regarding continuing the pregnancy. Termination of pregnancy may be advised in cases with other severe congenital anomalies and renal impairment.⁶ In some milder forms of megalourethra spontaneous resolution has been observed in fetuses at 19–34 weeks gestation.⁹



Figure 1D: Post-operative picture.



Figure 2: Ballooning of distal half of penis during micturition and dribbling of urine.

The treatment of megalourethra may be one stage or two stage urethroplasty depending on the age of presentation and general condition of the patient. For scaphoid type, Nesbitt first described a longitudinal reduction urethroplasty as was performed in these cases.⁵ Heaton and colleagues described a technique of urethral plication for some cases of scaphoid megalourethra.¹⁸ However, urologic repair is almost impossible when there is a lack of supportive corporal tissue, although successful cosmetic and functional repairs have been reported. The management of fusiform type is complicated ranging from sex reassignment to major phallic reconstruction.² Patients of fusiform type may require placement of penile prosthesis in the adult period.⁶ Long term follow-up is required in these patients to see for the impaired continence, erectile function and fertility potential.⁹

CONCLUSION

Congenital megalourethra is a rare and surgically correctible malformation of the anterior urethra. It can be detected antenatally by ultrasound. The condition should be specifically looked for in all fetuses with suspected lower obstructive uropathy and oligohydramnios. Detailed examination of the fetal perineum and genitalia will provide clues to the correct diagnosis. It is essential to consider megalourethra in the differential diagnosis of all children with penile or penoscrotal swelling. Management has to be individualized depending on type and the presence of associated congenital anomalies. The presence and severity of associated anomalies determines the quality of life. In isolated scaphoid type of megalourethra reduction urethroplasty gives excellent results as seen in our cases. Due to paucity of literature many cases are misdiagnosed and incorrectly treated. It is therefore imperative for pediatric surgeons, physicians and urologists to be aware and correctly diagnose, evaluate and manage such patients.

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