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Case Report

Congenital megalourethra and posterior urethral valve in a patient with Down syndrome

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ABSTRACT

Megalourethra is a rare congenital anomaly of the urethra that develops as a consequence of dysgenesis of the corpus cavernosum and corpus spongiosum. Herein, we present three congenital abnormalities in one newborn ie; Down syndrome, posterior urethral valve (PUV) and scaphoid megalourethra. The patient was treated successfully by ablation PUV and reduction uretrophlasty. The postoperative period was uneventful and the renal functions could be preserved in the long term.

Keywords: Down syndrome; megalourethra; posterior urethral valve; urethroplasty.

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Introduction

Megalourethra is a rare congenital anomaly of the urethra that develops as a consequence of dysgenesis of the corpus cavernosa and corpus spongiosum. During the embryologic development, mesenteric congenital anomaly resulting in diffuse dilatation of the penile urethra may cause megalourethra. It has been demonstrated that it may lead to functional obstructive uropathy.[1] Among other urinary system abnormalities, megalourethra may be strongly correlated with other urinary tract anomalies. Uretrographic findings of megalourethra, which presents with diffuse dilatation of the anterior urethra, reveal two types of megalourethra: scaphoid-type megalourethra that is the only developmental disorder involving corpus spongiosum of the anterior urethra, and a more severe fusiform type of megalourethra characterized by a developmental disorder of the corpus spongiosum and corpora cavernosa.[1,2] Anomalies of other systems may frequently accompany megalourethra.[3] Renal dysplasia/hypoplasia, posterior urethral valve (PUV), hydronephrosis, vesicoureteral reflux, undescended testicle, hypospadias, and anorectal malformations have been demonstrated to be associated with megalourethra.[1,3] Bielek et al.[4] identified 48 patients with PUV and Down syndrome. On the other hand, as far as we know, there is no study in the literature demonstrating the association of megalourethra with PUV and Down syndrome. Herein, we present three congenital abnormalities in one newborn namely Down syndrome, PUV and scaphoid megalourethra.

Case presentation

One-month-old infant admitted to our pediatric urology clinic with symptoms of difficulty in voiding, penile swelling, fever, and vomiting attacks. Physical examination revealed elongated penis deformity and clinical features of trisomy 21 (Down syndrome). Dribbling-type urine flow was observed from the urethral meatus, which was apparent after manual milking of the distal penis (Figure 1). Complete blood counts were within normal limits and creatinine level was 1.4 mg/dL. Urine culture revealed the presence of 100,000 cfu/mL *Escherichia coli*.

Figure 1. Dribbling of the urine can be observed in the infant with megalourethra

Bilateral grade 2 hydroureteronephrosis was detected in the urinary system ultrasound. Retrograde urethrography revealed accumulation of contrast agent in the anterior urethra, and a narrow segment was observed at the level of posterior urethra. After achieving sterile urine by administration of antibiotics for one week, operation was decided. The operative steps of the procedure are demonstrated in Figure 2. An endoscopic evaluation using a 8-Fr cystoscope demonstrated the presence of type one posterior urethral valve, and ablation was performed using a holmium YAG laser. Reduction urethroplasty was decided during the same procedure. A circumferential incision was performed and the skin degloved down to the base of the penis. The dilated urethra was vertically incised and opened. The redundant urethra was excised on one side and urethroplasty was performed over an 8-Fr urethral catheter using 7-0 PDS running sutures. The other side of the redundant urethra was used as a second layer and fixed to the penis using 5-0 vicryl sutures. Circumcision was performed and skin was closed using 5-0 rapid vicryl sutures. The operation was terminated after fixation of 8-Fr urethral catheter to the glans penis. Coban bandage was used for the dressing of the penis. Patient was discharged at home 24 hours after surgery. The urethral catheter was removed on postoperative 10th day.

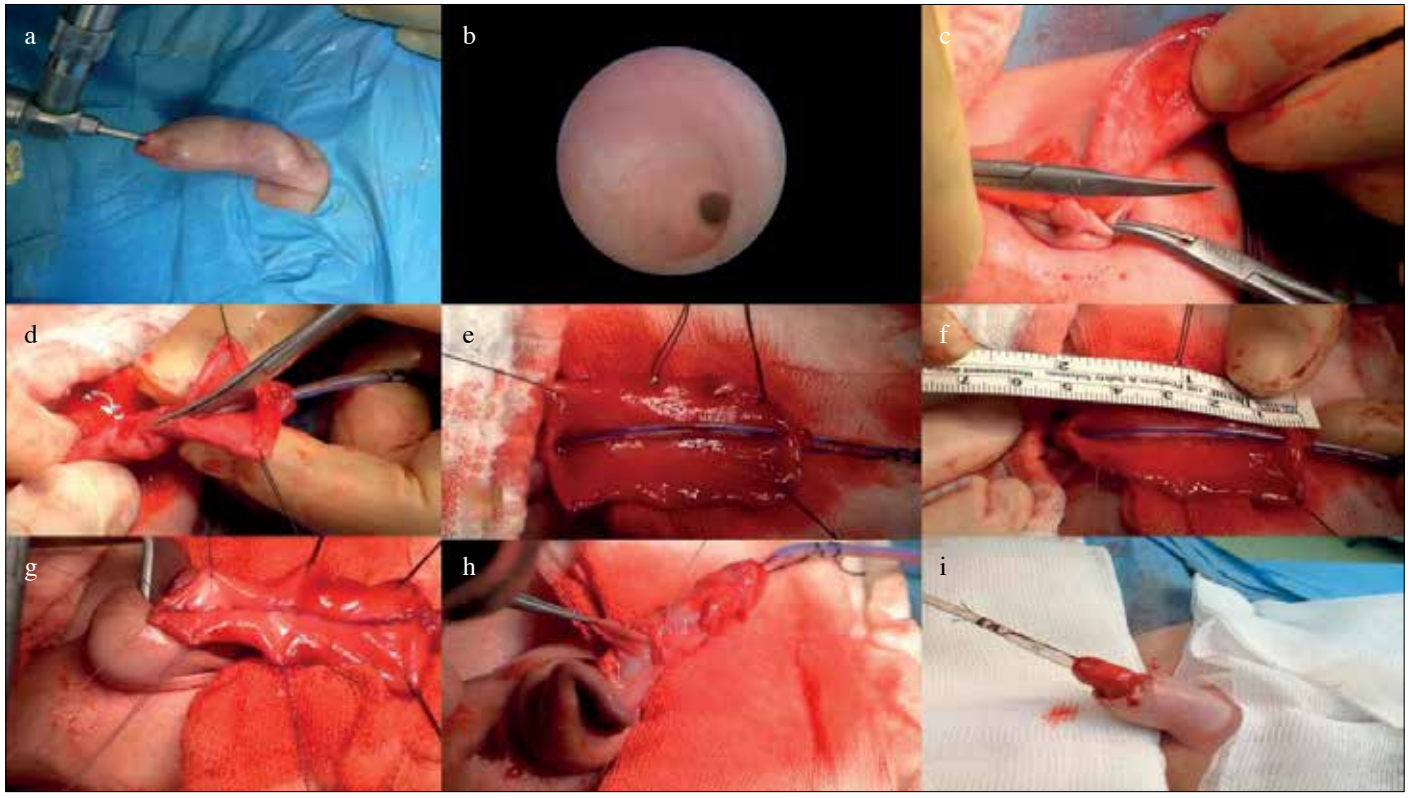

Figure 2. a-i. Operative steps of cystoscopy and reduction urethroplasty. (a) Cystoscopic appearance of the dilated urethra. (b) Cystoscopic view of dilated urethra. (c) Degloving of the penis after circumferential incision. (d) Vertical incision of the urethra. (e) Appearance of the whole dilated urethra after complete incision. (f) Measuring the dissection line. (g) Performance of urethroplasty before resection of the dilated segments. (h) Closure of urethral line as a second layer by dilated urethra harvested from one side. (i) Postoperative appearance of penis

Micturition was normal and postoperative creatinine value was 0.2 mg/dL. Bilateral hydroureteronephrosis completely resolved in the third month after surgery. The postoperative period was uneventful and the patient reported no symptoms. When we decided to write this case as a case report, we obtained an informed consent form from the parents of the patient.

Discussion

Our case report demonstrated the first time three different congenital abnormalities in one patient: megalourethra, PUV and Down syndrome. The patient was treated by ablation of PUV and reduction urethroplasty at the same session. Postoperative period was uneventful and the renal functions turned to normal levels. Megalourethra is a rare congenital anomaly of the anterior urethra. Obrinsky et al.[5] reported the first case of congenital megalourethra in the literature. Stephens and Fortune[2] classified the megalourethra into two groups as fusiform and scaphoid type, in accordance with the urethrographic findings of dilatation in the anterior urethra. The defect is in the corpus spongiosum in scaphoid type, a relatively simple form of megalourethra; the fusiform type is a more severe form and the defect involves both corpus spongiosum and corpora cavernosa.[1,6] Megalourethra has an unknown etiology. Corpora cavernosa and spongiosum, which have roles in penile erection, develop in the mesoderm during the 7th week of fetal life. Megalourethra develops due to a developmental disorder of these erectile tissues in the mesoderm which are not supported by the urethral epithelium. Voiding cystourethrogram (VCUG) is used both for the diagnosis of of megalourethra for the differentiation between obstruction and stenosis.[7] Megalourethra does not lead to an actual anatomic obstruction, but creates a functional obstruction, which leads to stasis.[2] Megalourethra may cause oligohydramnios by creating an obstruction in prenatal period, and pulmonary hypoplasia may be detected in some cases. In addition, megalourethra may cause end-stage kidney disease by inducing chronic kidney damage. Jones et al.[3] reported that scaphoid, and fusiform -type megalourethra had respective mortality rates of 13%, and 66%, during the perinatal period. Megalourethra may be associated with gastrointestinal and other system anomalies in 85% of the cases.[7] These anomalies include vertebral anomalies, anal atresia, cardiac malformations (VACTERL), trachea-esophageal fistula (TEF), renal and extremity anomalies, PUV, urethral duplication, renal dysplasia, polycystic kidney, brachydactyly, unilateral renal hypoplasia, intestinal malrotation, hydroureteronephrosis, cryptorchidism, vesicocutaneous fistula, and prune-belly syndrome.[1-3] Researchers reported about cases of 100 megalourethra in the literature, and concomitancy between megalourethra and PUV has been rarely observed.[8,9] Down syndrome in association with upper and lower urinary system anomalies, and obstructive uropathies have been also rarely reported.[10] PUV is seen in 16.9% of every 10,000 newborns with trisomy 21.[5] However, concomitancies among trisomy 21, PUV, and megalourethra have been rarely reported in the literature. PUV is seen in one of every 5000 to 8,000 births.[11] In 1919, Young suggested a classification of PUVs in three groups according to the shapes and location of PUVs.[12] Most cases of PUV can be identified using prenatal sonography. The main imaging method used in the postnatal period with the suspicion of PUV is transabdominal or transperineal ultrasonography. Thickening of the bladder wall and prostatic urethral dilatation and key-hole sign may be observed in the evaluation. The major diagnostic method used in PUV is voiding cystourethrography (VCUG). During VCUG imaging, trabeculated bladder, dilatation in prostatic urethra, and inconsistencies between the dimensions of the prostatic urethra and of other parts of the urethra may be detected.[13] In our case the bladder was cystoscopically normal and no trabeculation was observed.

Megalourethra and PUV may cause hydroureteronephrosis, bladder dysfunction, vesicoureteral reflux, and renal dysfunction. We detected bilateral grade 2 hydroureteronephrosis in our case, and regression of bilateral hydronephrosis was observed after the ablation procedure applied for PUV, and urethroplasty for megalourethra.

Nesbit and Baum[14] first described the surgical repair of scaphoid megalourethra 50 years ago, and the procedure could be achieved with one or two sessions of urethroplasty, according to the patient's age and general condition. Prognosis of megalourethra depends on the degree of renal damage, and there is a possibility of renal dysplasia due to the association of PUV with megalourethra in these patients.[15] In addition, normal micturition and erection must be examined during the follow-up of these patients due to the absence of supportive structures of penile.[7,9]

Renal function test results were within normal limits, and micturition force was at adequate levels after postoperative examinations in our patient.

Taken altogether when megalourethra is diagnosed, physicians must keep in mind that urinary system or other system anomalies may accompany megalourethra. Early surgical management and multidisciplinary approach including pediatric urologist, pediatric nephrologist and genetics are critically important in the well-being of the patients.

In conclusion, megalourethra is a rare congenital anomaly which may be associated with other congenital abnormalities. Our case report demonstrated the first time three different congenital abnormalities in one patient namely megalourethra, PUV and Down syndrome. Early diagnosis and management is crucial to prevent the patients from deterioration of the renal functions.

Informed Consent: Written informed consent was obtained from patient who participated in this case.

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