Rare Case of Recurrent Fetal Familial Ureterocele

Abstract
We presented a rare case of familial recurrent ureterocele and duplex right kidney. The diagnosis was made at the anomaly scan at 19 weeks. Our patient is a case of first-degree consanguineous marriage with multiple miscarriages, two daughters with congenital ureteroceles and one healthy boy. Postnatal ultrasound confirmed the diagnosis. In spite of prophylactic antibiotics the patient developed severe urinary tract infection. Voiding cysterourethrogram (VCUG) revealed ureterocele without vesicoureteric reflux. The urinary tract infection was recurrent and severe and required urgent cystoscopy and puncture of ureterocele.

Keywords: Ureterocele; Prenatal; Ultrasound; Genetics

Case Report
Mrs. N A is a 35 year-old patient, multigravida. She was seen at the Fetomaternal Clinic for follow up of her pregnancy. She is married to her first-degree cousin. A prenatal ultrasound performed in the prior visit revealed a single viable female fetus. The follow up ultrasound in the 19th week of gestation showed normal amniotic fluid volume and left-sided unilateral hydrenephrosis with a full-distended bladder. Additionally, thinning out of the parenchyma and a dilated left ureter ending in a large left ureterocele. Douplex kidney was suspected. There was no any other abnormal scan findings. The patient was closely monitored.

In the 22nd week of gestation an ultrasound scan showed left renal anterior posterior (AP) dilation, which measured 9 mm with normal kidney function indicated by normal liquor and a distended bladder. The dilatation increased to 18 mm in the 26th week seen by ultrasound scan, and no other abnormalities were noted (Figure 1). Subsequent antenatal and ultrasound were showed no marked changes in these findings.

Figure 1: Antenatal Ultrasound: There is unilateral hydrenephrosis, with thinning of the parenchyma. A fullness is noted raising the possibility of Duoplex kidney (Panel A). A septum is seen across the bladder initial diagnosis was bladder diverticulum, subsequent scan confirmed Ureterocele (Panel B)
Postnatal Period

Mrs. NA gave birth to a baby girl by cesarean section at 37 weeks of gestation because of oligohydramnios. The birth weight was 2.1 kg, had a measured length of 49 cm, (7th percentile) and a head circumference of 33 cm (<3rd percentile).

She did not require admission to NICU. Although the baby was started on prophylactic antibiotics (amoxicillin) since birth, on the 16th post-natal day, she was admitted to the emergency due to dysuria noticed by her mother. She was given the diagnosis of urinary tract infection (Klebsiella pneumoniae, sensitive to cepotaxime). Further workup was done when the patient was admitted to the hospital. Abdominal ultrasound (Figure 2) of the kidney revealed cystic area in the upper part of the left kidney, upper moiety, which is an indicative of duplex kidney. Urinary tract ultrasound showed a right kidney size of 4.4 cm and left kidney size of 6.8 cm by length and a left-sided duplex kidney with hydronephrotic upper motility. The scan also showed a tortuous dilated left ureter measuring 8.6 mm, 14.6 mm and 7.6 mm at the proximal, middle and distal regions respectively (Figure 3).

The young patient was booked for Voiding cystourethrogram (VCUG), which was done uneventfully. VCUG revealed ureterocele without vesicoureteric reflux (Figure 4). Mrs. NA 7 years old daughter had a similar clinical condition at birth with left-sided ectopic duplex ureterocele, her condition was diagnosed only postnatally and corrected by surgery. Our young patient had the final diagnosis of a duplex right kidney and with large Ureterocele in the bladder: The patient is booked for cystoscopy and puncture of ureterocele. This was done laparoscopically and without any complications. The patient is undergoing further testing and genetic workup.
Sözübir et al. [4] document the largest series of familial cases of ureteroceles, giving evidence for genetic background. They have reviewed retrospectively the charts of patients with familial ureteroceles seen between 1992 and 2002 [4]. This is the largest series of familial ureterocele patients in the literature. Three of the families have twin siblings with ureteroceles. Our patient is a recurrent case of ureterocele in a single pregnancy. The first familial case of twin siblings with ureterocele was reported in 1936 by L. W. Riba, which was simple systemureterocele in identical twins [10].

We are reporting another case familial ureterocele, which is rare. In this case of first-degree consanguineous marriage there were multiple miscarriages, two daughters with congenital ureteroceles and one healthy boy. This mode of inheritance is most consistent with an autosomal recessive pattern. Autosomal recessive diseases are associated with consanguineous marriage, due to the method of inheritance requiring two homozygous affected alleles. This further supports the genetic basis of familial ureterocele in terms of ureteral development. There is a special interest in ureteroceles which ‘run’ in families which has prompted many studies. One such study published in The Turkish Journal of Pediatrics concluded that the link between the inheritance and ureteroceles was autosomal dominant as opposed to recessive [4]. This gives us a clue that there may be multiple types of inheritance pattern for this condition, which warrants further studies and analysis of genetics.

**Conclusion**

We have presented a rare case of familial ureterocele. The diagnosis was made during the morphology scan, however, the diagnosis could have been made earlier if she was seen at the feto-maternal center earlier based on her family history. Close collaboration between the attending obstetrician, feto-maternal specialist, pediatrician, radiologist and pediatric surgeon resulted in satisfactory outcome. In future pregnancies the patient may benefit from Prenatal Genetic Diagnosis (PGD), as it may possible to identify the affected gene by studying genetic work up of affected siblings and their parents.

**Acknowledgement**

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**Conflict of Interest**

None.
References