Prenatal Diagnosis of Familial Ureteropelvic Junction Obstruction: A Case Report and Review of the Literature

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Ureteropelvic junction obstruction, the most common cause of neonatal hydronephrosis, is well detected by prenatal sonography. It is usually sporadic in presentation, and familial occurrence is rare. The authors present one such case of likely familial hydronephrosis due to ureteropelvic junction obstruction in a mother and fetus that was detected on prenatal sonographic imaging. The importance of prospective fetal screening in mothers at risk for fetal obstructive uropathy is illustrated so as to facilitate timely postnatal management of the disease process.

Key words: ureteropelvic junction obstruction, hydronephrosis, familial

Congenital anomalies of kidneys and ureters are found in 3% to 4% of the population and account for 30% of all malformations detected prenatally on sonography (USG).[1] These include numerous anomalies such as dilatation of the urinary tract, renal agenesis, renal ectopia, bladder exstrophy, and so forth. A systematic approach is required in the prenatal detection of urinary tract anomalies. This approach includes assessment of the amniotic fluid volume (AFV), localization and characterization of urinary tract anomalies, and the search for other associated abnormalities.

The most common urinary tract anomaly detected prenatally is dilatation of the urinary tract, which may be due to both obstructive and nonobstructive causes. Ureteropelvic junction (UPJ) obstruction is the most common cause of neonatal hydronephrosis.[2] Although UPJ obstruction is a common entity, it is usually sporadic. Familial UPJ obstruction is very rare. It has an autosomal dominant mode of inheritance with variable penetrance in most families and complete penetrance in few.[3,4] A genetic interrelationship has also been suggested between UPJ hydronephrosis and a bifid pelvicalyceal system.[5] We diagnosed

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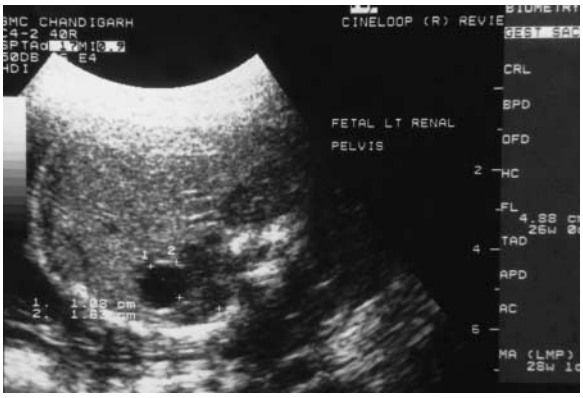

FIG. 1. Prenatal scan showing dilated left renal pelvis of fetus.

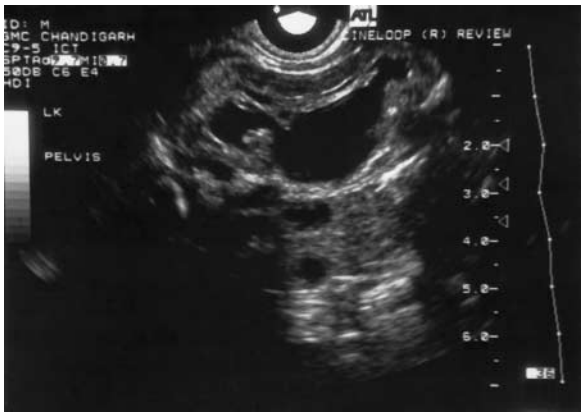

FIG. 2. Postnatal scan showing hydronephrosis on left side.

one such case of familial hydronephrosis due to UPJ obstruction on an antenatal obstetric USG.

Case Report

A patient in her 30s, gravida 3, para 2, was referred to us for USG at 28 weeks of gestation. She had bilateral renal masses with fever and deranged renal functions. She was discovered to have grossly hydronephrotic kidneys with ballooning of the pelvis and internal echoes. The ureters were not dilated, leading to the diagnosis of bilateral UPJ obstruction with pyonephrosis. Bilateral percutaneous nephrostomy was carried out under sonographic guidance, taking all aseptic precautions, following which the renal functions became normal.

On obstetric scan, the left fetal kidney showed a dilated pelvis with an (intrarenal) pelvic AP diameter (RPD) of 10.8 mm (Fig. 1). The right fetal kidney also showed dilatation of the renal pelvis with an RPD of 5.5 mm. However, the calyces and ureters were not dilated on either side. Fetal urinary bladder was also normal. No other major anomaly was found in the fetus. AFV was found to be adequate.

The patient was followed up at 35 weeks of gestation. The dilatation of fetal pelves was found to have progressed to 16 mm on the left side and to 8.6 mm on the right side. However, dilatation was not seen to reach the calyces or ureters, thereby confirming the diagnosis of fetal UPJ obstruction on the left side.

The parents, siblings, and other two children of the patient were also screened for the presence of any urinary tract anomalies. No abnormality was detected in any of them. Postnatally, on day 3, a sonogram of the newborn female child detected hydronephrosis (RPD was 1.5 cm) only in the left kidney that was reaching up to the calyces (Fig. 2). However, the ureter was not dilated, confirming the prenatal diagnosis of left UPJ obstruction. The right kidney showed no evidence of hydronephrosis at all, thereby suggesting a physiological etiology to the antenatal dilatation of the renal pelvis on this side.

Discussion

UPJ obstruction is usually sporadic in presentation, with familial UPJ hydronephrosis being very rare. Only 32 families have been reported thus far in the literature.

UPJ obstruction is the most common cause of neonatal hydronephrosis, and it is well detected prenatally as a dilated renal pelvis with or without caliectasis. The ureter is not visualized. It is usually unilateral but may be bilateral in 10% to 30% of cases.[6]

Generally, it is accepted that before 20 weeks of gestation, an RPD of 4 mm or more should be considered abnormal.[7] However, after 20 weeks of gestation, there is much controversy about the exact RPD that indicates significant hydronephrosis. Most studies, however, take an RPD of 1 cm or greater as abnormal.[8] An RPD of 5 to 9 mm is usu-

ally physiological, as it was in our case in the right fetal kidney (RPD was 8.6 mm), whereas the left-sided UPJ obstruction was pathological (RPD was 16 mm).

What is important is that if the renal pelvis appears abnormal, follow-up sonography is warranted after six to eight weeks. Serial sonographic scans are essential to assess the progression of hydronephrosis and AFV. When the contralateral kidney is normal, the prenatal detection of UPJ obstruction should not alter obstetric management, as the prognosis is good. However, in bilateral cases, prognosis depends on the severity and duration of obstruction, as well as the AFV. Early delivery/intrauterine shunting is rarely indicated, and only if there is marked progressive bilateral obstruction with severe oligohydramnios.

If the dilatation persists, postnatal follow-up studies are required. The timing of follow-up postnatal scans further depends on the severity of prenatal hydronephrosis. If there is severe dilatation of the renal pelvis, early USG evaluation should be performed to permit early intervention. Otherwise, USG examination should be postponed for three to seven days as a relative state of dehydration and physiologic oliguria exists in the first 48 hours of life. This can lead to an underestimation of the degree of hydronephrosis or even a false-negative renal sonogram.[9] If the initial postnatal sonogram is normal, it should be repeated in four to six weeks to exclude obstruction.[10]

Review of the literature shows that only 32 families with hereditary hydronephrosis have been reported thus far in research studies. In fact, major histocompatibility (HLA) complex typing has been done in a few studies and has led to the localization of the gene(s) responsible for genuine hereditary hydronephrosis in the 6p human chromosome.[11,12] Some studies have pointed to genetic heterogeneity (i.e., different loci of genes have been found in different families). Two main modes of inheritance have been proposed—an autosomal dominant mode of inheritance with complete penetrance. However, variable penetrance has also been found.[3-5]

Although genetic consultation of the mother and child was not obtained, it is unlikely that the two coexisting conditions could be a mere coincidence, and we feel that there are more chances of this being a familial rather than a sporadic UPJ obstruction.

No specific guidelines exist as such for prenatal or postnatal sonographic detection of familial hydronephrosis. Few studies suggest screening all first-degree relatives of patients with UPJ hydronephrosis for early diagnosis and management of this condition.[4,5] Only one study suggested prospective screening in the form of fetal USG as a further step in this direction.[4] However, to the best of our knowledge, this is the first case in which fetal screening was done in a mother with bilateral UPJ obstruction and familial hydronephrosis was found, which was confirmed on the postnatal USG.

Thus, we wish to highlight the importance of prospective screening and recommend fetal screening sonography as a mandatory procedure in all patients at risk for fetal obstructive uropathy so as to facilitate timely postnatal management of the disease process. This screening should be carried out throughout pregnancy because it may manifest as late as the third trimester of pregnancy, as in our case.

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