Ultrasonographic features of glomerulocystic disease in infancy: similarity to infantile polycystic kidney disease

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Abstract. Glomerulocystic kidney disease (GCD) is an unusual condition characterized pathologically by cystic dilatation of Bowman's space and the first portion of the proximal convoluted tubule. We report the serial ultrasound findings in an asymptomatic infant with GCD which were initially confused with infantile polycystic kidney disease. We emphasize the nonspecificity of ultrasonographic findings in cystic renal disease in early infancy and suggest a protocol for the diagnostic work-up of bilateral renal cystic disease in children.

Early diagnosis of cystic renal lesions in infants, many of which are hereditary, is essential for early management and appropriate genetic counseling. Multicystic renal dysplasia, adult-type polycystic kidney disease and infantile type polycystic kidney disease (IPCKD) have typical, though not diagnostic, findings, which ostensibly may allow a rapid, non-invasive diagnosis. The lack of specificity of the ultrasonogram in distinguishing renal cystic lesions was emphasized to us by a patient whose intrauterine and immediate postnatal examinations suggested IPCKD but who subsequently was found to have glomerulocystic disease (GCD) when the ultrasonographic features changed. The purpose of this report is to alert physicians to the potential confusion of the rare GCD with IPCKD when relying exclusively upon ultrasonographic examination and to emphasize the importance of histologic diagnosis in selected cases of childhood renal cystic disease.

Case report

A 1-month-old male infant was referred for a renal ultrasound examination. Fetal ultrasound imaging performed at 8 months gestation and a neonatal ultrasound examination had demonstrated bilaterally enlarged kidneys. The serum creatinine level was 0.7 mg/dl at birth. Physical examination revealed no dysmorphic features. The neonatal course was complicated by a spontaneous pneumothorax requiring oxygen therapy. Renal ultrasound examinations of both parents were normal.

An ultrasound examination performed at our institution at one-month of age (Fig. 1) revealed bilaterally enlarged echogenic kidneys (both > 2 SD above the mean for length), and a presumptive diagnosis of autosomal recessive infantile polycystic kidney disease was made. The infant continued to grow and develop normally with continued normal renal function and a decrease in renal size by physical examination.

Repeat renal sonography at 15 months of age demonstrated that the kidneys were no longer enlarged (both < 1 SD below the mean for length). There continued to be loss of definition of corticomedullary junctions and diffuse increased echogenicity of the medullary pyramids but of a lesser degree than on early examinations (Fig. 2).

Renal biopsy (Fig. 3) demonstrated subcapsular glomerular cysts up to 0.5 mm in size with moderate dilatation of the proximal convoluted tubules, interstitial fibrosis and lymphocytic infiltration. The pathological diagnosis was glomerulocystic kidney disease.

Fig. 1. Renal ultrasound at 1 month of age demonstrates an enlarged right kidney with diffuse increased echogenicity, loss of the corticomedullary junction, ill-defined renal margins and mild dilatation of the renal pelvis. The left kidney (not shown) demonstrated similar findings.
Fig. 2. Renal ultrasound at 15 months of age demonstrates resolution of nephromegaly. The kidneys (right one shown) now have well defined cortical margins and normal cortical echogenicity. The medullary pyramids remain hyperechoic.

Fig. 3. Renal biopsy. Subcapsular cysts containing glomerular tufts (arrows) are diagnostic of glomerulocystic kidney disease.

Discussion

Glomerulocystic kidney disease is a rare condition which may occur in otherwise normal infants [1] or in association with multiple malformations such as oral-facial-digital syndrome, [2] renal retinal dysplasia [3], trisomy 13–15 [4], Zellweger syndrome [5], and others [6]. The condition is always bilateral, consisting of cystic dilatation of Bowman’s space, therefore confined to the renal cortex, usually most severely affecting the more peripheral cortex [7]. The collecting tubules may also be dilated to a variable degree. In some cases, an inflammatory infiltrate or fibrosis is present in the interstitium. Reniform shape is usually maintained and the kidneys are frequently enlarged. The pathogenesis is generally unknown; however, medullary obstruction and gestational maternal phenacetin ingestion have been suggested as potential mechanisms [8]. While most cases appear to be sporadic, an autosomal dominant pattern of inheritance has been suggested in two kindreds [9].

This case demonstrates that the ultrasonographic features of GCD may be confused with infantile polycystic kidney disease. Although both GCD and IPCKD are characterized by small cysts in enlarged kidneys, the cysts are limited to the distal nephron in IPCKD rather than the predominant finding of dilatation of Bowman’s capsule in GCD. Sonographically both IPCKD and GCD show bilateral nephromegaly, indistinct renal margins, increased echogenicity of the renal cortex and loss of definition of the corticomedullary junction [10]. Renal macrocysts and hepatic cysts may also be found in GCD as well as in IPCKD [1, 11].

A previously unemphasized aspect of GCD is that renal size may diminish as the child grows. Lieberman and associates have also noted that some children with IPCKD have a reduction in renal size during childhood [12]. In our patient, the renal length returned to normal by ultrasonographic examination during the second year of life. Loss of corticomedullary differentiation persisted; however, the previously noted cortical hyperechogenicity resolved and the renal margins became well defined.

The natural history of GCD is uncertain due to the rarity of clinical recognition. Renal function may be normal though several previous reports have [2, 3, 9, 12] described decreased glomerular filtration rates with some patients requiring dialysis or renal transplantation. Our patient’s serum creatinine was normal and, had gestational ultrasonography not been obtained, this infant’s cystic renal enlargement may have escaped detection. The increased utilization of fetal ultrasound may, therefore, increase the detection of GCD as well as other cystic renal disorders in children and require our greater familiarity with the ultrasonographic features of this unusual disease.

The sonographic findings of cystic renal diseases in utero [13] or in young children are often typical, but are not diagnostic. Because of the importance of a correct diagnosis for genetic counseling and, perhaps, patient management, we believe histologic confirmation is required when clinical examination and family screening studies fail to determine the diagnosis. When a child is found to have bilateral renal enlargement with macrocysts and/or increased echogenicity, both parents should be carefully examined and investigated with renal and liver ultrasound. If neither parent has evidence of APCKD or tuberous sclerosis, there is no evidence...