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INFANTILE POLYCYSTIC KIDNEY
(Case report)

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Summary: A case of infantile polycystic kidney is presented. It is emphasized that the outcome of patients who survive the neonatal period appears not to be so grim as previously feared of. Not to underscore the importance of agressive supportive care, the need for physician education and early diagnosis of the type of Polycystic kidney disease (PKD) as a necessity for providing accurate genetic counseling is also stressed.

Key words: Infantile polycystic kidney

Infantile polycystic kidney is a rare autosomal recessive disorder characterized by microcysts in kidneys and liver. The causative mechanism of this disorder is still unknown, diagnosis is generally based on radiological findings and kidney and liver biopsies (4). But in the diagnosis of autosomal dominant polycystic kidney disease DNA probe is useful (7). It is usually caused by a mutant gene at the PKD-1 locus on short arm of chromosome 16 (2).In this article we presented a seven month-old boy with infantile polycystic kidney. His two brothers had died of unknown reasons at the age of 4 months and one day.

Case report

Seven month old male infant was admitted to the hospital with complaints of abdominal mass and restlesness. Intraabdominal mass had been discovered 15 days before, during a physical examination for unrelated complaints and he was referred to our hospital.

In family history parents were first cousins. First pregnancy had ended in abortion for unknown reason, second son at the age of 4 months and third son in his first day of life had died for unknown reasons. One daughter and one son were in good health. There was no reported history of renal disease in their family.

Physical examination revealed the child with a temperature of 36oC (axillary), pulse rate of 122/min., respiration rate of 24/min. and blood pressure of 140/100 mm Hg. His weight was 8700 g. and height 81 cm. His general condition was good. Liver was 6-7 cm palpable and both kidneys could be easily felt 2 cm below the posterior costal angle. The rest of the findings were normal.

Laboratory findings included Hb: 10.8 gr/dl., white blood count 9000 per mm3, and normal peripheral blood smear. Chest X-ray, ECG, serum Na,+ K,+ glucose, creatinin were in normal levels. Urinanalysis revealed pyuria and E.coli was grown. In abdominal ultrasonography both kidneys were enlarged, calyces were distorted, parenchimal echo was increased, but no cystic formation was observed.In IVP both kidneys were enlarged with minimal function, and poor calyceal definition. Pyelotubuler reflux and minimal calyxial distortion, was noted in retrograde pyelography (Picture 1).

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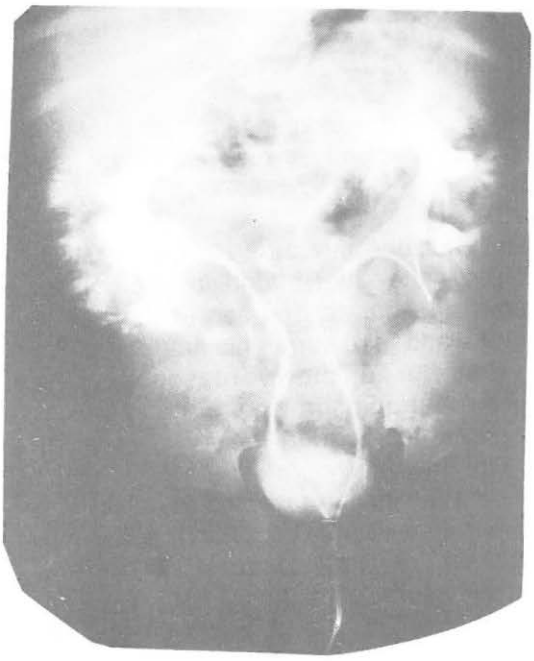
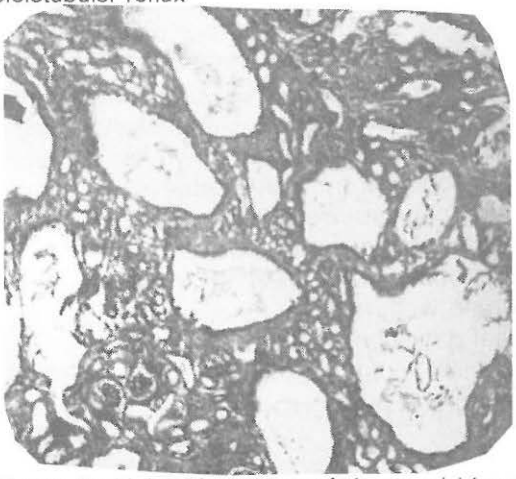


Picture 1: Retrograde pyelography on the patient showing bilateral minimal calyxid distortion and plelotubuler reflux



Picture 2: Light microscopy of the renal biopsy demonstroting markedly dilated elongated collecting ductus and tubules

On the basis of clinical and laboratory findings, the patient was diagnosed as having renal cystic disease and urinary tract infection, gentamycin 6 mg/kg/d. and propranolol 2 mg/kg/

d. were started. After hypertension and urinary tract infection were under control, renal and liver biopsies were performed.Evaluation of tissue specimens confirmed the clinical diagnosis (Picture 2).

Discussion

Although infrequent, this is the most common and characteristic form of polycystic kidney of infancy. Incidence is 1/10.000. Characteristically it is a bilateral process and is familial(9).As in our patient both kidneys are symmetrically enlarged sometimes to a severe degree and relatively smooth in outline. The cut surfaces of kidney resemble a sponge, being traversed by radiating fusiform dilated tubules that occupy the entire kidney and obscure the corticomedullary markings. There is always continuity of the lumen of the nephron from Bowman space to the bladder (3).

Every patient with recessive polycystic kidney disease has an affected liver regardlesss of age. Liver shows proliferation elongation,and cystic dilatation of the small bile ducts and ductules. Liver fibrosis is common but is not a prominent feature of the disease. There is no careful study of a patient with recessive polycystic kidney who shows histologically normal liver (5). In our case liver biopsy revaeled minimal fibrosis but not cystic dilatation.

The pathogenesis of this disease is stil unknown. One possibility is primary defect in peritubular, or periductal supporting structures, possibly due to an abnormality in collagen formation (4).

Most severely affected neonates with the oligohydramnios sequence (Potter face, pulmanary hipoplasia) die soon after birth. Patients whose disease develop in childhood may have an enlarged liver which, because of increasing fibrosis, becomes smaller and firmer. Older children tend to have less severe renal impairment and they more often have severe hepatic involvoment with portal hypertension. Those children come to the medical attention because of complication of hepatic fibrosis have firm livers, splenomegaly, hematemesis (due to eusophageal varices) cholangitis, growth retardation, anemia, leucopenia, thrombocytopenia and other clinical features. Infant with hypertension or extrarenal manifestation should be suspected of having of infantile polycystic disease(1). Our patient did not have any of the above mentioned extrarenal conditions.

In one study all patients had developed hypertension by 3 months of age (1).Urinary infection must be treated promply in these patients. The most two important problems of our patient were urinary infection and hypertension.

Classically IVP has been used to make the diagnosis of this disease, although renal function in these patients are so poor that delayed films usually were required to show the findings. Films obtained early in examination demostrate only a faint nephrogram and blatchy opacification of large kidneys. On delayed films, the radiating arrangement of dilated collecting tubules (sun burst nephrogram pattern) become apperent. On retrograde pyelography contrast material may reflux into dilated tubules giving the whole kidney a streaky appearance (8). Our patient's radiological findings were typical for polycystic kidney (picture1).

On ultrasonographic examination an enlarged kidney that maintane areniform shape and increased echogenity both renal cortex and medulla, poor calyceal definition and poor delineation of renal margine from surrounding tissue was seen. Because cysts are tiny it is diffucult to visualize the small fluid-filled spaces as such but they do provide a myriad of reflecting surfaces that produces the diffusely bright echogenity. Macrocysts on the other hand appear as anechoic rounded areas(6). It is suggested that with newer generation high resolution scanners a distinct sonographic appearence may be seen. We were not able to visualize cysts ultrasonographically in our patients.

Pyelographic diagnosis of polycystic kidney disease may be misleading. The pyelographic appearence of recessive form may change to mimic that of autosomal dominant polycystic kidney.

Sonographie and computed tomoggraphy (CT) are more sensitive to detect paranchimal cystic changes so that polycystic kidneys can be easily differentiated from multicystic renal displasia.

The spesific diagnosis of the types of PKD is diffucult.During infancy, neither clinical signs nor radiographic diagnostic studies seem to be reliable(1). One or more of the following criteria is necessary to classify a patient in the "recessive PKD group" 1. Congenital hepatic fibrosis in liver biopsy or evidence of portal hipertension 2. Renal histologic studies consistent with collecting tubule ectasia or 3. A sibling known to have the disease (1).

The liver biopsy is essential in the accurate diagnosis of cystic disease in children in the absence of definitive family history of recessive or dominant PKD(1).
In most persons with risk of polycystic kidney disease imaging techniques are the

only mode of reaching a diagnosis before symptoms appear (2).

Prenatal diagnosis may be feasible with ultrasonography. Assesment is not completely reliable by week 22 of gestation. Excessive excretion of alfa-fetoprotein or trehalase into amniotic fluid are the markers remains to be evaluated. So distinction between adult and infantil type of polycystic kidney disease is based on clinicopathological and genetic grounds (5).

It is demonstrated that DNA probe is useful, in the diagnosis of autosomal dominant polycystic kidney disease (7). Autosomal dominant polycystic kidney disease is usually caused by a mutant gene at the PKD-1 locus on the short arm of chromosome 16 but in about 4 percent of families with this disorder it is caused by unknown mutations elsewhere in the genome (2). This way parents can elect to have the pregnancy terminated and this can be done by the twelfth week of gestation.There are no DNA probes to confirm recessive form

There is no spesific therapy for PKD. It is recommended that an agressive approach to medical management of children with PKD should be adopted rather than assuming that a dismal prognosis is inevitable and intensive therapy(for complications, hypertension, urinary infection..,) might be worthwhile. Renal transplantation is recommended because of end-stage renal insufficiency.

Prognosis is generally poor. Many affected infants die in the newborn period, or shortly after from respiratory failure, uremia or hypertension. Some patients probably the less severly affected ones live longer, sometimes several months or years with progressive renal insufficiency. Survival still depends to a large extent on the age of the patient at presentation. With improved respiratory support in the neonatal period, it is anticipated that more affected neonates will survive.

References

1. Barbara C,Canley SB, Stapleton FB : Polycystic kidney disease in the first year of life. **J Pediatrics** 111:693, 1987.

2. Birfrey SP: The diagnosis and prognosis of autosomal dominant polycystic kidney disease. **N Eng J Med** 18:1085-1090,1990.

3. Drummond KN:Devolopmental abnormalities of kidney and urinary collecting system.In:Vaughan VC, Mc Kay RJ, Behrman RE (eds). Nelson **Textbook of Pediatric** . WB Saunders Co. Philadelphia, London, Toronto, Sidney, Tokyo 1987, pp 845-846.

4. Hamburger J, Crasnier J : **Nephrology** . John Wiley&Sons Co. New York 1979, pp 1023-1041.

5. Kaplan BS, Kaplan P , Rosenberg HK: Polycystic kidney disease in childhood. **J Pediatrics** 115:867-890,1989.

6. Keith HC, Leonard S : **Pediatrics Ultrasonography**. Williams& Wilkins Co. London 1987, pp 291-300.

7. Reeders ST, Gal A, Propping P: Prenatal diagnosis of autosomal dominant polycystic kidney disease with DNA probe. **Lancet** 5:6-7,1986.

8.Silverman FN: **Caffey's Pediatric X-Ray Diagnosis**. Chicago, Year Book Medical Publishers 1985, pp1630-1637.

9. Straud WR, Rushton HG,Marhle BM: Autosomal dominant polycystic kidney disease in infants, **J Urology** 141:1151-1153,1989.