Case Reports

Cystic Kidney Disease

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The term polycystic kidney disease (PKD) has inappropriately been used to describe any kidney riddled with cysts(l). Polycystic kidney disease is a rare disorder and was seen in 16 patients whereas renal cysts occurred in 136 of 6521 consecutive autopsies in infants and children(2). The matter has been further complicated with the recognition of juvenile presentation of autosomal recessive(3) and infantile presentation of autosomal dominant polyc-ystic kidney disease(4). Moreover it seems possible to differentiate most cases of these two conditions on the basis of history, his-tological findings in kidney and other organs(5,6) and radiological examination(7). It is important to establish the exact diagnosis as this helps in genetic counselling. Review of the Indian literature shows only a few reports of this condition(l,8,9). The present report is based on autopsy findings of infants admitted to our nursery with a clinical diagnosis of polycystic kidney disease in order to define the underlying disease and associated anomalies.

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Material and Methods

We reviewed the records of the patients admitted to our nursery with the diagnosis of polycystic kidney disease. The diagnosis of polycystic kidney disease was based on presence of bilateral renal lumps on clinical examination and presence of multiple renal cysts on ultrasonographic examination. Only seven cases, in whom autopsy were done, were analyzed. Gross examination findings were recorded. Microscopic study of slides prepared from different organs and stained with hematoxylin eosin stain was done and findings were recorded. Patients were classified into four groups (z) autosomal recessive polycystic kidney disease, *(ii)* autosomal dominant polycystic kidney disease, *(Hi)* indeterminate polycystic kidney disease, and *(iv)* other cystic disorders. One or more of the following criteria was necessary to define a patient as autosomal recessive polycystic kidney disease(6): *(a)* congenital hepatic fibrosis in liver histology or evidence of portal hypertension, *(b)* renal histological studies consistent with collecting tubule ectasia, and (c) a sibling known to have autosomal recessive polycystic kidney disease. Inclusion into autosomal dominant polycystic kidney disease necessitated *(a)* positive parental history, *(b)* macroscopic cysts in liver or other organs, and (c) presence of Berry aneurysms. Patients who could not be classified into any of the above were labelled as indeterminate polycystic kidney disease. The pathologic diagnosis of polycystic kidney disease was based on presence of multiple radially arranged cysts with absence of dysplastic tissue in the kidneys(lO). The diagnosis of renal dysplasia was based on presence of disorganized parenchyma, presence of primitive ducts tubules, fetal glomeruli or metaplastic cartilage(ll).

Results

On the basis of autopsy findings two patients were classified as autosomal recessive and two as indeterminate polycystic kidney disease. None was classified as autosomal dominant polycystic kidney disease. These were three cases of multicystic renal dysplasia. The clinical and autopsy findings are shown in *Tables I & II,* respectively. All seven children died of respiratory failure, although raised blood urea (more than 40 mg/dl) and serum creatinine (more than 1.5 mg/dl) were seen in four of the seven cases. Case 3 and case 7 had a history of death of previous sib with the diagnosis of bilateral polycystic kidney but autopsy was not done in these babies.

Discussion

Polycystic kidney disease in neonates is suspected on the basis of presence of bilateral renal lumps. These babies may or may not have hypertension during neonatal period(6). It is on these grounds that these

TABLE I–*Clinical Features of Infants with Cystic Kidney Disease*

S. No.	Birth weight (kg)	Gestational age (wks)	Antenatal history	Delivery	Apgar score 1",5", 10"	Musculoskeletal system	Other Systems
1.	2.0	38	Normal	Breech	2,4,5	Skin, Soft tissue contracture	RD
2.	1.8	34	Toxemia	Cephalic	4,7,7	Normal	RD
3.	1.9	40	Normal	Cesarean section	4,8,8	Bilateral CT EV	RD
4.	1.5	33	Normal	Breech	6,8,9	Normal	RD, thick umbilical cord, large placenta
5.	1.1	30	Normal	Breech	1,1,1	Bilateral CTEV	RD, rudimentary pelvis and scrotum
6.	1.3	31	Maternal fever at 2 months, APH at 7 months	Cephalic	1,1,1	Normal	RD, liver 4 cm, pansystolic murmer
7.	1.8	34	Oligohydramnios	Cephalic	1,3,5	Bilateral CTEV upper limb deformities	RD, Potter's sequence

APH=Antepartum hemorrhage, CTEV=Congenital talipes equino varus, RD=Respiratory distress.

children were suspected as cases of infantile polycystic kidney disease. However, three of the seven cases turned out to be cases of renal dysplasia. Two could be classified as autosomal recessive and remaining two as indeterminate polycystic kidney disease. Congenital hepatic fibrosis, which is considered a must for the diagnosis of autosomal recessive polycystic kid-ney(5), was not seen in these two cases.

On the other hand histology of these two cases did not suggest a diagnosis of autosomal dominant polycystic kidney disease but case 4 has cystic lesions in both lungs which are seen in autosomal dominant polycystic kidney disease(12). Case 6 had Meckel's diverticulum and multiple gastrointestinal stenosis. Although pyloric stenosis is known to be frequently associated with autosomal dominant polycystic kidney disease, multiple intestinal stenosis has not been reported with any type of polycystic kidney disease. Gastrointestinal lesions are known to occur with renal dysplasia, but our patient 6 had no evidence of renal dysplasia.

The urinary tract and other anomalies seen in our cases of renal dysplasia are well recognized(ll). None of our cases was clearly a case of autosomal dominant polycystic kidney disease. However, it is important-to classify the cases of polycystic kidney disease in autosomal dominant or autosomal recessive varieties using genetic probes as the mutation of the gene that gives rise to autosomal dominant polycystic kidney disease has been found to disrupt a gene on the distal third of the short arm of chromosome 16(13,14). This gene locus is now knows as "PKD 1".

However, some workers have not been able to confirm this association between "PKD 1" and autosomal dominant polycystic kidney disease(15,16) and, therefore, it may still be considered a genetically heterogenous disorder despite the fact that no other gene locus has been described so far. Autosomal recessive polycystic kidney disease can be diagnosed on the basis of exclusion as no genetic probes are available at the moment for this disease although it has been shown that the mutations leading to autosomal recessive polycystic kidney disease are not allelic with autosomal dominant polycystic kidney disease(17). All efforts should be made to identify autosomal dominant polycystic kidney disease in family members who may be unaware of their illness. Early identification of cases with autosomal dominant polycystic kidney disease may increase the life span of such individuals(6). Such information can also be of help in genetic counselling to such parents.

It is also important to differentiate polycystic kidney disease (both autosomal recessive and autosomal dominant) from cases of multicystic dysplasia as the later is a sporadic condition, chances of recurrence of which are minimal.

TABLE II- Autopsy findings in infants with Cystic Kidney Disease

S. No	Kidney	Urinary Tract	Liver	Others	Diagnosis
1	Bilateral multiple cysts 2-5 mm size, renal dysplasia present	Duplication of right ureter, multiple strictures in ureter, small rudimentary urinary bladder	Normal	Lungs hypoplastic with wide areas of atelectasis and mild interstitial emphysema	Renal dysplasia
2	Bilateral fetal lobulation poorly developed glomeruli and tubules	Pelvicalcyceal system could not be made out on left side, normal on right side	Normal	Alveolar emphysema with collapse of intervening alveoli, pleural effusion right side, absent left adrenal, hemorrhage in right adrenal	Renal dysplasia
3	Bilateral infantile polycystic kidney	Normal	Dilated proliferating biliary duct and increased periportal and septal fibrous tissue	Premature fetal lungs with diffuse interstitial emphysema, wide PDA, ostium primum defect	Autosomal recessige PKD
4	Bilaterally radially arranged cysts 1-5mm size, cortex and medulla could not be differentiated	Normal	Normal	Cystic lesions in lungs, most marked right upper lobe and lingual of left lobe	Indeterminate PKD
5	Bilateral congenital dysplastic kidneys with occasional cysts	Underdevelope d pelvicalcyceal system, ureters normal size with blind distal end	Normal	Premature fetal lungs, wide PDA	Renal dysplasia
6	Bilateral cysts in radial distribution 10-20 mm in size, some cysts fluid filled	Normal	Normal	Premature lungs with poor alveoli, wide PDA, Meckel's diverticulum, multiple intestinal stenosis	Indeterminate PKD
7	Bilateral cysts arranged in radial distribution, 2-8mm size, cortex and medulla can not be made out	Norma	Peri portal and septal fibrous tissue increased with some proliferating ductules	Bilateral primary atelectasis with focal areas of partial expansion	Autosomal recessive PKD

PKD= Polycystic Kidney Disease, PDA= Patent ductus arteriosus

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