ISSN : e- ISSN 2395-3950, p-ISSN 2395-440X

Autosomal Recessive Polycystic Kidney Disease -A Case Report

[1]Vashistha Amitabh, [2]Rajpurohit Bajrang S, [3]Gehlot Kushal B, [4]Mittal Shilpi, [5]Sunil Saini, [6]Khan Alsaba,

[1]MD Radiodiagnosis, Jaipur, Rajasthan.
[2]MD Radiodiagnosis, Jhalawar Medical College, Rajasthan.
[3]Assistant Professor, Department of Radiodiagnosis, RNT Medical College, Udaipur, Rajasthan.
[4&5]III Year PG Resident, [6]I Year PG Resident, Department of Radiodiagnosis, RNT Medical College, Udaipur, Rajasthan.

ABSTRACT
Autosomal recessive polycystic kidney disease (ARPKD) is inherited in a recessive fashion with varying degrees of clinical presentations. There is medullary ductal ectasia and loss of renal function. Most children will die shortly after birth. Here we present a rare case of two year old female child presenting with complaints of abdominal distension and failure to thrive, diagnosed to have ARPCKD on USG and CT scan.

Key-words: Polycystic kidneys, Enlarged kidneys, Striated nephrogram and Caroli's disease.

Corresponding Contributor: Dr. Kushal B. Gehlot, Assistant Professor, Department of Radiodianosis, RNT Medical College, Udaipur, Rajkasthan. Email-**drkushalgehlot@gmail.com**

INTRODUCTION:

Autosomal recessive polycystic kidney disease (ARPCKD) is characterized by the circumferential epithelial proliferation of distal tubules and collecting ducts, resulting in tubular lengthening and ectasia with interstitial fibrosis. It is also associated with variable degree of periportal fibrosis and bile duct ectasia. ARPCKD is, the less common variant of polycystic kidney disease in which mutations in the PKHD1 (chromosomal locus 6p12.2) cause ARCPKD.[1]

CASE REPORT

A two year old female child had undergone sonography for abdominal distension, failure to thrive. USG revealed enlarged kidneys with smooth surface and diffusely increased echogenicity. She was referred for a CT scan for further evaluation. CECT scan (GE 16 slice spiral CT scanner) revealed enlarged kidneys with reduced attenuation on non contrast images (Fig.1). Delayed contrast enhancement with delayed persistent striated nephrogram noted in post contrast study (Fig.3). Hepatomegaly was noted with biliary dilatation and focal cystic dilatation in both lobes (Caroli's disease) (Fig.2).

DISCUSSION

ARPKD is a rare genetic disease with an incidence between 1:40,000 and 1:55,000 live births. The gene for ARPKD is located on chromosome 6. It has variable phenotypic presentation with greater the percentage of abnormal collecting tubules, the more severe renal compromise and earlier the clinical presentation. There is an inverse relationship between renal and hepatic involvement. Fetal abdominal cysts are frequently found on routine antenatal ultrasound.[2,3,4] In ARPCKD we find enlarged kidneys with pyramidal

ISSN : e- ISSN 2395-3950, p-ISSN 2395-440X

hyper-echogenicity similar to medullary nephrocalcinosis.[2] However, a definitive diagnosis is often not made until postnatal life, and detection of an intra-abdominal cyst antenatally rarely alters obstetric management.[3]

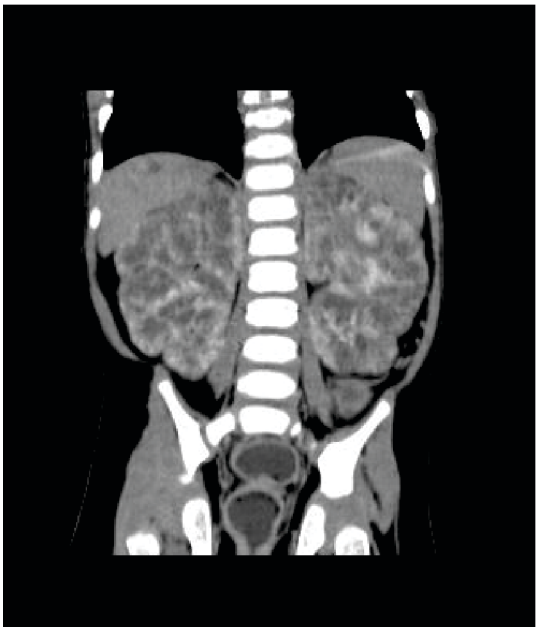

Fig.1- coronal NCCT image, showing enlarged bilateral kidneys with reduced attenuation.

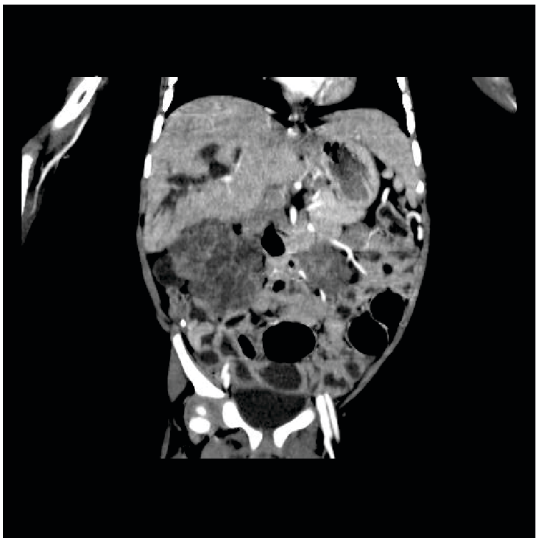

Fig.2- coronal NCCT scan showing hepatomegaly with biliary dilatation and focal cystic dilatation in both lobes (caroli's disease).

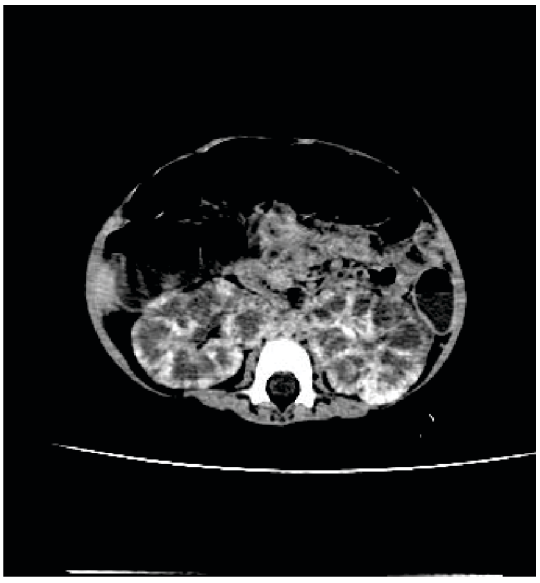

Fig.3- Axial CECT image showing Delayed contrast enhancement with delayed persistent striated nephrogram noted on post contrast study.

Blythe and Oekenden classified ARPCKD in four forms –

1. Perineatal form (more common).
   - 90% of tubules are affected, showing cystic changes.
   - Onset of renal failure in utero.
   - Enlarged kidneys with oligohydramnios and pulmonary hypoplasia.
   - It is uniformly fatal.
2. Neonatal form:-
   60% of tubules are affected + minimal hepatic fibrosis and bile duct proliferation.
   - Onset of renal failure within one moth.
3. Infantile form:
   - 25% of tubules are affected with mild to moderate periportal fibrosis and duct ectasia.
   - It manifest as chronic renal failure, hypertension and portal hypertension with onset at 3-6months of age.
4. Juvenile form:-

ISSN : e- ISSN 2395-3950, p-ISSN 2395-440X

- 10% of tubules are affected with gross hepatic fibrosis and bile duct ectasia (caroli's disease).
- Onset at 6 months to 5 yrs.
- Death from portal hypertension.

On USG kidneys are enlarged with generalized increased echogenicity because of increased interfaces produced by dilated tubules. Corticomedullary differentiation is poor. Occasionally small (0.5-1cm) cysts are visible. On CT–Kidneys are enlarged with reduced density and show delayed opacification with persistent striated nephrogram reflecting stasis of contrast in the dilated collecting tubules which can be prolonged for days after the CT Scan. The pelvicalyceal systems are normal. Hypodense linear bands corresponding to hepatic fibrosis and dilated bile ducts seen with a spectrum of portal hypertension. Life-table survival rates calculated from birth revealed that 86% were alive at 3 months, 79% at 1 year, 51% at 10 years, and 46% at 15 years. Calculations based on patients who survived to 1 year of age showed that 82% were alive at 10 years and 79% at 15 years. These results reveal an improved prognosis for a condition once assumed to be fatal.[5]

TREATMENT

Although a cure for PKD is not available, treatment can ease the symptoms and prolong the life. Over-the-counter pain medications, such as paracetamol can relive pain, urinary tract infections which can be treated with antibiotics. Cyst infections are difficult to treat because many antibiotics do not penetrate into the cysts. Keeping blood pressure under control can slow the effects of PKD. Finally, we will have to replace kidney functions by dialysis or transplantation. Healthy (non-PKD) kidneys transplanted into PKD patients do not develop cysts.

CONCLUSION: Because both the renal and hepatic involvement in ARPKD tend to progress, most patients, if they survive to adulthood, will develop renal insufficiency and portal hypertension, although progression is not inevitable. Early diagnosis and intervention may help slow or arrest some features of the disease, extending the life expectancy and quality of life for affected individuals.

Support: NO

Conflicts of interest: NO

References:

1. Adeva M. El-Youssef M, Rossetti S, Kamath PS, Kubly V, Consugar MB et al. Clinical and molecular characterization defines a broadened spectrum of autosomal recessive polycystic kidney disease (ARPKD). Medicine. 2006;85:1-21. PubMed ID:**16523049**.
2. Okumura M,' Bunduki V, Zugaib M. **Prenatal Diagnosis** 2006; 26(4):330-32.
3. Blews D. Sonography of the neonatal genitourinary tract. Radiologic Clinics of North America, 37(6):1199-1208.
4. McEwing, Rachae, Hayward, Christina F, Margaret. Australasian Radiology. 2003;47(2):101-10.
5. Kaplan BS, Fay J, Shah V, Dillon MJ. Barratt TM Hospital for Sick Children, London, UK. **Pediatric Nephrol. 1989;3(1):43-49**.
6. Dalgaard OZ. Bilateral polycystic disease of the kidneys; a follow-up of two hundred and eighty-four patients and their families". Acta Med Scand. 1957;328:1-255.
7. Zerres K, Mücher G, Becker J, et al. Prenatal diagnosis of autosomal recessive polycystic kidney disease (ARPKD): molecular genetics, clinical experience, and fetal morphology". Am. J. Med. Genet. 1998;76(2):137-44.