Indian Journal of Basic and Applied Medical Research; December 2016: Vol.-6, Issue- 1, P. 439-441

Case report

A case report of Autosomal Recessive Polycystic Kidney Disease: A rare entity

Dr. Meenal Shingade[1], Dr. Nalini Kadgi[1], Dr. Nicholas Dcunha[2], Dr. Shaila Puranik[3]

[1]Associate Proffesor, Dept. of Pathology, B.J.G.M.C, Pune.

[2] Resident doctor, Dept. of Pathology, B.J.G.M.C, Pune.

[3] Proffesor and Head of Department, Dept. of Pathology, B.J.G.M.C, Pune.

Corresponding author: Dr. Nicholas Dcunha

Abstract:

Autosomal recessive polycystic kidney disease (ARPKD) is a rare hereditary disorder with an estimated incidence of 1 per 20,000 live births (1). ARPKD is caused by mutations of a gene located on chromosome 6p21.1-p12 named polycystic kidney hepatic disease gene (PKHD1) (2). Herein we present a medico-legal postmortem case of ARPKD in a five month old female infant. The infant had multiple microcysts in the kidney and liver. On microscopy, the cysts were lined by cuboidal epithelium. Liver also showed bile duct proliferation and hepatic fibrosis. The case has been discussed here due to its rarity and close differential diagnosis.

Keywords: Autosomal recessive polycystic kidney disease, microcysts

Introduction:

ARPKD is one of the rare but important cystic renal diseases. It is a rare hereditary disorder with an estimated incidence of 1 per 20,000 live births (1). We present here a case of ARPKD in a 5 month old female infant.

Case History:

The viscera of a 5 month old female infant was brought to our autopsy department for histopathological examination. The clinical details are not available as it was a medicolegal autopsy. The viscera received were pieces of lungs, liver, spleen, kidney, cerebrum and whole heart.

Gross: Kidney

Grossly, both the kidneys were enlarged and measured 7x5x3cm each. External surface showed fetal lobulations and the capsule was easily stripped off. Cut section showed multiple small cysts measuring 1-2mm in size, completely replacing the cortex and medulla; giving it a sponge like appearance, but the reniform shape of the kidneys was maintained.

Microscopy: Kidney

On microscopy, there were multiple saccular and cylindrical cysts. The cysts were uniformly lined by cuboidal to flattened epithelium. Glomeruli were normal. There was no interstitial fibrosis or inflammatory infiltrate.

Gross: Liver

The liver did not show cysts grossly.

Microscopy: Liver.

The liver showed partially effaced architecture. Hepatocytes were unremarkable. Multiple microscopic cysts lined by flattened epithelium were seen. The liver also showed marked portal bile duct proliferation and fibrosis. Reticulin stain done on the liver confirmed the increased fibrosis around portal tracts.

The differential diagnosis considered in this case were ARPKD, Autosomal Dominant Polycystic

Kidney disease (ADPKD), nephronpthisis, medullary sponge kidney and multicystic renal dysplasia.

Discussion:

ARPKD is a rare form of renal cystic disease. It is distinct genetic and morphological profile from Autosomal Dominant Polycystic Kidney Disease (ADPKD) and other cystic diseases of the kidney. ARPKD is caused by mutations of a gene located on chromosome 6p21.1-p12 named polycystic kidney hepatic disease gene (PKHD1) (2). This gene is related to the protein called as polyductin or fibrocystin (03) which is highly expressed in the epithelial cells of collecting ducts and thick ascending loop of Henle and to a lesser extent in the biliary and pancreatic duct epithelia (04).

Morphologically, ARPKD consists of bilaterally enlarged kidneys but retaining their reniform configuration. The cysts tend to be linear and tend to radiate from the medulla to outer cortex. Our case showed a similar gross appearance. Microscopically, the cysts are lined by cuboidal or flattened epithelium. The liver also shows bile duct proliferation and fibrosis (05).

Ruling out the differential diagnoses: ADPKD is one of common cystic kidney diseases in adults but is extremely rare at such an age. Though there have been cases of ADPKD at this age, the absence of large cysts helps exclude this diagnosis (06). Nephrophthisis is another close differential to ARPKD, having similar cysts with hepatic fibrosis and also its infantile form falls in the same age group but the absence of a granular surface of kidney and no tubulo-interstitial fibrosis or lymphocytic infiltrate are points against this diagnosis. Multicystic renal dyplasia also occurs at the same age but microscopic picture shows cysts lined by cuboidal epithelium surrounded by immature stromal elements which is not present in our case. Medullary sponge kidney and simple renal cysts do not occur at such an early age (05).

Conclusion:

Considering the morphological findings, age and ruling out all the other differentials, a final diagnosis of ARPKD was given. The case has been discussed due to its rarity and close differential diagnosis.

Figure Legends:

Fig 1: Kidney: Gross appearance : Kidney shows multiple small cysts in cortex and medulla – sponge kidney.

Fig 2: Kidney: Microscopy; Kidney shows multiple cysts lined by flattened epithelium: H&E stain– 100x.

Fig 3: Liver: microscopy; Liver shows cysts and bile duct proliferation: H&E – 100x .

Fig 4: Liver: microscopy; Liver show increased fibrosis; Reticulin stain - 100x.

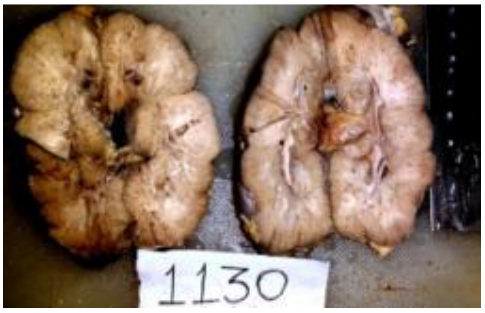

Fig 1: Kidney: Gross appearance : Kidney shows multiple small cysts in cortex and medulla, perpendicular to the surface.

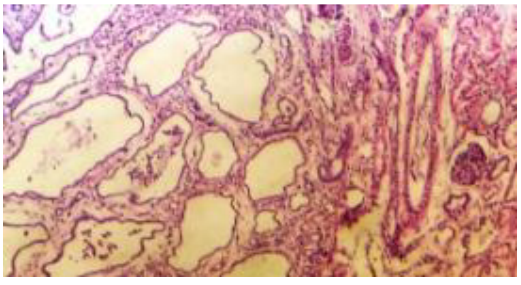

Fig 2: Kidney: Microscopy; H&E – 100x: Kidney shows multiple flat lined cysts

www.ijbamr.com P ISSN: 2250-284X , E ISSN : 2250-2858

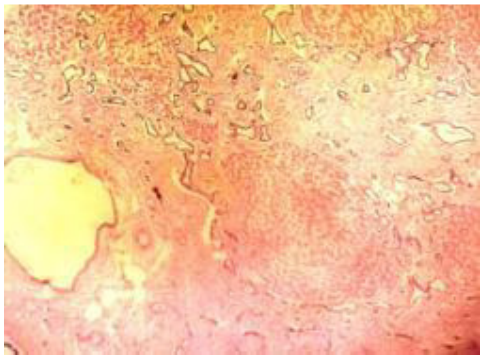

Fig 3: Liver: microscopy; H&E – 100x: Liver shows cysts with architectural loss.

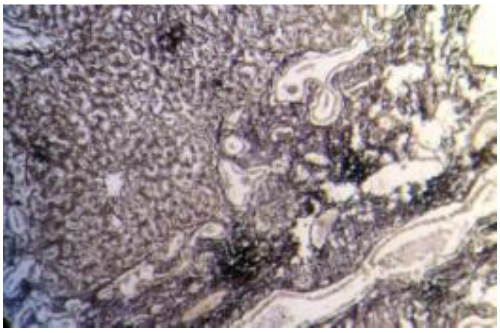

Fig 4: Liver: microscopy; Reticulin stain – 100x : Liver show increased fibrosis.

References:

(01) Guay-Woodford LM, Desmond RA. Autosomal recessive polycystic kidney disease: the clinical experience in North America. Pediatrics. 2003 May 1;111(5):1072-80.

(02) Zerres K, Mücher G, Bachner L, Deschennes G, Eggermann T, Kääriäinen H, Knapp M, Lennert T, Misselwitz J, Von Mühlendahl KE, Neumann HP. Mapping of the gene for autosomal recessive polycystic kidney disease (ARPKD) to chromosome 6p21–cen. Nature genetics. 1994 Jul 1;7(3):429-32.

(03) Mücher G, Becker J, Knapp M, Büttner R, Moser M, Rudnik-Schöneborn S, Somlo S, Germino G, Onuchic L, Avner E, Guay-Woodford L. Fine Mapping of the Autosomal Recessive Polycystic Kidney Disease Locus (PKHD1) and the Genes MUT, RDS, CSNK2β, and GSTA1 at 6p21. 1–p12. Genomics. 1998 Feb 15;48(1):40-5.

(04) Menezes LF, Onuchic LF. Molecular and cellular pathogenesis of autosomal recessive polycystic kidney disease. Brazilian journal of medical and biological research. 2006 Dec;39(12):1537-48.

(05) Ordonez N, Rosai J, Urinary tract In: Rosai J, Surgical Pathology. 10th edition vol 1. USA: Elsevier; 2011. Pg 1171.

(06) Nahm AM, Henriquez DE, Ritz E. Renal cystic disease (ADPKD and ARPKD). Nephrology Dialysis Transplantation. 2002 Feb 1;17(2):311-4.