Polycystic Kidney in a Foetus
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Abstract:
Polycystic kidney disease is one of the life-threatening inherited disorder summated by the development of bilateral or unilateral renal cysts that might lead to renal failure in due course of time. This disorder affects 1 in 1000 live births. A stillborn male human foetus of 31 weeks gestation was brought from one of the private nursing homes by postgraduate students for study of foetal anomalies as part of their project work. During dissection of the foetus, the kidneys were found to be of 8 cms in length and 5.5 cms in width which were almost of the adult size.

The kidneys were subjected to histological examination. Microscopic examination revealed scanty cortical areas with glomeruli and proximal convoluted tubules with large cystic cavities at the juxta cortical regions.

Key Words: Cystic cavities, glomeruli, renal cysts.

Introduction:
Polycystic kidney disease (PKD), a common genetic cause of chronic renal failure in children and adults, is characterized by the accumulation of fluid-filled cysts in the kidney and other organs. Polycystic kidney disease can be inherited as an autosomal dominant trait (ADPKD) or an autosomal recessive trait (ARPKD). Autosomal dominant trait PKD is a common disease that occurs both in children and adults, whereas autosomal recessive trait PKD is uncommon and occurs primarily in neonates and children. The gene responsible for autosomal recessive polycystic kidney disease (PKHD1) has recently been identified on short arm of chromosome no. 6 (Verghese, 2006). Autosomal recessive trait PKD occurs in 1 in 6000 to 1 in 40,000 live births (Zerres et al, 1998). Disease was first recognized in 1902; however, the histology was not reported until 1947 (Verghese, 2006). The primary defect in autosomal recessive PKD is linked to ciliary dysfunction. It is also associated with growth retardation, urinary tract infection and hyponatremia (Verghese, 2006).

Case Report:
A 30 year old female gravida II para-I delivered a still born male fetus of 31 weeks gestational period in a private nursing home and was brought to the Department of Anatomy, Maharajah’s Institute of Medical Sciences, Nellimarla, by postgraduate students as a part of their project work for congenital anomalies. There was no history of consanguinity amongst the parents. First born was a male child who had no congenital abnormality. Besides the detailed family history for congenital anomalies, a detailed antenatal & post natal history was recorded. After fixation, the abdomen of the still born foetus was opened for study. Kidneys were examined macroscopically and then were subjected to histological examination.

The antenatal period was uneventful. The cord blood was collected by the nursing home authorities at the time of delivery. Chromosomal analysis was done but Gene mapping was not done.

On gross examination the kidneys on both sides showed lobulations and enlargement. The length of both the kidneys was 8 cms while breadth was 5 and 5.5 cms on right and left sides respectively at the hilum. The right and left kidneys weighed 80 and 85 gms respectively (Fig. I).
Coronal section of kidneys showed bilateral symmetrical cystic dilatations. The cysts were subcortical. The cortico-medullary junction was not distinct due to cystic transformation of medulla causing distortion of renal columns (Fig. II).

Fig. II: Arrows showing cystic cavities, cortex, medulla & corticomedullary junction.

On histological examination, cortex revealed areas of glomeruli with dilated proximal and distal convoluted tubules. Cortico-medullary junction revealed large cavities and medulla showed very large dilated sections of collecting tubules (Fig. III).

![Fig. III: Arrows showing cystic dilatations, sections of glomeruli & dilated collecting tubules (H & E, 10X).](image)

There was distortion of glomerular membrane and an increase in the urinary spaces (Fig. IV). Proximal convoluted tubules were dilated and their normal architecture was lost. No difference was appreciated in the cytology of proximal and distal convoluted tubules (Fig. V).

In longitudinal sections, medulla showed dilated descending and ascending limbs of loop of Henle and its lining epithelium was completely distorted. Collecting tubules were not clearly demarcated (Fig. VI). Dilated collecting tubules showed distorted lining epithelium which was cuboidal and multilayered. Closely packed mesenchymal cells along with sections of blood vessels were observed in the interductular area (Fig. VII). Other organs did not show any abnormality.

**Discussion:**

The permanent kidney develops from two sources, one in the mesoderm of sacral part of nephrogenic cord and other from the ureteric bud, a diverticulum arising from the mesonephric duct. Further, development involves epithelial mesenchymal interaction. Epithelium of ureteric bud interacts with mesenchyme of metanephric blastema. The mesenchyme expresses a transcription factor that makes this tissue competent to respond to the induction of ureteric bud. In the autosomal recessive polycystic kidney disease, cysts occur from collecting ducts, due to deficient or non-responsive cell adhesion molecules, syndecan and E-catherin which are essential for condensation of mesenchyme to epithelium (Sadler, 2006).

All typical cases of autosomal recessive PKD are due to mutations of PKHD1 gene on chromosome no. 6p21.1 (short arm p of chromosome no. 6, band 21, region 1). The autosomal recessive polycystic kidney presents as bilateral enlarged kidneys and severe renal
failure (Deget et al, 1995). In the present case also the kidneys were bilaterally enlarged and were of approximately the adult size.

According to the history taken, the pedigree chart was drawn as below. It shows that the polycystic kidney disease was seen only in this generation.

Since, the polycystic kidney disease has not been observed in the parents and preveious generations, the mode of inheritance may be autosomal recessive or may be a new mutation. Chromosomal analysis was found to be 46 XY.

The renal cystic disease typically begins in utero and manifests as fusiform dilatation of collecting ducts that radiate from medulla to cortex (Blyth & Ockenden, 1971). The present study is in agreement with the study cited above.

Qian et al (2005) observed that histologically kidney showed simple cysts lined by a single layer of epithelial cells, multilayered cysts lined by a hyperplastic epithelium. The present histological study also confirms the cysts with single layer as well as multilayered epithelium.

**Conclusions:**

It is a case of autosomal recessive PKD as it has affected the foetus in utero. The kidneys were lobulated and were of the adult size.

Cystic dilatations were seen through out the cortex and medulla macroscopically. Reniform shape was maintained most probably, due to the fact that the cysts were present in collecting ducts and were of <5 mm in size.

**Bibliography:**


