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## An Autopsy Case of Infantile Polycystic Kidney

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A well-developed fourty-minutes-old male neonate with polycystic kidneys was held an autopsy. Both kidneys were remarkably enlarged and had uniform sponge-like appearances. The cortex and medulla was replaced by innumerable small cysts with lining cuboidal epithelium, reflecting their origin from tubules. The glomerulus was observed disseminatedly. The liver had several bile duct cysts varying in size.

There are various abnormalities of renal differentiation which results in cystic disease. One of them, infantile polycystic kidney is rare and specific type. Classification of polycystic kidney was reviewed here.

### CASE REPORT

Patinet is a male neonate who was born after 39 weeks' gestation to a 24-year-old prima gravida mother. His birth weight was 2,940 gm. The pregnant course and delivery were uncomplicated. The infant whose initial Apgar score was 1 had severe respiratory distress begining at birth, and vigorous resuscitative efforts were begun. But distension of the lungs were not developed, and cardiac arrest was elicited. The infant died fourty minutes later.

**Pathological Findings :** At autopsy, the abdomen was markedly distended by bilaterally enlarged kidneys, weighing 190 g and 200 g were normal in shape (Fig. 1). The external surface of the kidneys were smooth and light rouge in color (Fig. 2), showed innumerable miliarycysts varying from 1 to 2 mm or less in diameter beneath the thin capsule. On bisection they were entirely cystic and had "spongy" appearance. Cortico-medullary markings were observed. The cortical part was traversed by grossly visible thin-walled tubular or slender cysts having their long axis perpendicular to the cortical surface. The medullary part was composed of large numbers of thin-walled vesicular cysts varying from 1 to 3 mm or less in diamter (Fig. 3,4). Throughout the kidneys, no normal

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parenchyma could be recognized grossly. The pelves, ureters and bladder appeared normal.

Microscopically, the dilated cysts were lined by a single layer of cuboidal epithelium (Fig. 5), some of which formed stratification or short papillary projections (Fig. 6). Normal glomeruli were present probably in normal numbers and no dilated Bowmann's capsule were seen (Fig. 7). Renal stroma was somewhat edematous with mild fibrosis.

The liver weighed 140 g, was dark reddish brown in color with a smooth outer surface. The extrahepatic bile ducts and the gall bladder showed no abnormalities. The cut surface disclosed several cysts, varying from 2 to 5 mm in diameter, scattered irregularly throughout the right hepatic lobe. A few blood vessels were disclosed in the cystic cavities. The parenchyma of the liver was well preserved (Fig. 8).

Histological examination of the liver demonstrated a bizarre infolding, proliferation and ectasia of portal bile ducts, associated with perportal fibrosis. Some of the cysts were visible within the lobules, but the hepatic architecture was well preserved.

In the small intestine, at a distance of 80 cm from Treitz's ligamentum, there was a solitary polypoid lesion 5 mm in diameter with smooth contour (Fig.9). It was adenomyoma with proliferation of cystic ductules associated with increase of smooth muscle, which affected the submucosa and muscle layer, and no aberrant pancreas was demonstrated in the serial sections. No other cysts were found in other organs. The lungs were little aerated, the left weighing 12 g, and the right 15 g. The heart had no congenital anomaly.

## DISCUSSION

Several classification of cystic disease of the kidney in children have been presented by some authors<sup>1)2)3)</sup>. Gleason and coworkers proposed the classification with investigation of clinical, roentgenologic, and pathologic feature (table 1)<sup>2)</sup>. According to their classification, this case is "Polycystic renal disease, infantile type" (infantile polycystic kidney). Infantile polycystic kidney is always associated with specific hepatic lesions, and this description is applicable to this case. This has been observed in siblings<sup>4),5)</sup>, but not has been reported in more than one generation, because the early death of patients with infantile type are not concerned in reproduction<sup>6)</sup>. It has been generally considered to be autosomal recessive,<sup>4)5)7)</sup> but the genetics of infantile type is not always clear. The cases of adult type in a child can present in infancy or early childhood<sup>8)9)</sup>, but these cases are rarer, because they are asymptomatic in childhood.

In the infantile type, there is confusion with true sponge kidney disease arises from the fact that the kidney resembles a sponge and has been described as spongy. Absolutely no connection exists between the infantile polycystic kidney and sponge kidney disease<sup>10)</sup>. Sponge kidney is historically named after a striking "spongy" appearance to the excretory urogram by Cacchi and Ricci (1949), and such kidney assigns to Osathanondh and Potter's type 3.

Blyth and Ockenden<sup>5)</sup> have presented the 25 cases of infantile polycystic kidney with actual family data, and separated into four groups. These are (1) Perinatal,

(2) Neonatal, (3) Infantile, and (4) Juvenile groups. The term "infantile polycystic kidney" may not be a suitable name for kidneys of this sort, because other varieties are found in infants more commonly than these. Blyth et al. have described this variety as "Childhood type of polycystic disease"<sup>5)</sup>. The four groups have been named after the age of clinical presentation, but the essential difference is the degree of renal involvement. The most severe form is the perinatal group in which about 90% or more of the renal tubules are involved. On the contrary, the extent of periportal fibrosis is minimal in this group, grading up to juvenile with gross of periportal fibrosis. Our presenting case is, from the clinical manifestations and the histological findings of the kidneys and liver, classifiable in the perinatal group. Within a family contained more than one affected child, they are always in the same group<sup>5)</sup>. In the present case, no family data has been given. This very specific combination of kidney and liver malformations characterizes the "Childhood type of polycystic disease of kidneys and liver"<sup>5)</sup>.

In relation to the congenital hepatic fibrosis<sup>1)5)8)</sup>, it may be a form of polycystic disease to be described as the juvenile group in which the renal involvement is minimal<sup>11)</sup>. The separate entity of congenital hepatic fibrosis is questionable. This parallelism of the renal and hepatic malformation in infantile polycystic diseases suggests that they are the result of causative factors common to the two sites. Although about the pathogenesis of cysts of liver it was thought that congenital fibrosis around the bile ducts was the primary change, Norris and Tyson<sup>12)</sup> have described abnormal enzyme patterns or chemical organizers. Benjamin et al.<sup>7)</sup> have suggested that the common factor may be deficiency of an enzyme activity.

Numerous theories to account for the histogenesis of polycystic kidney have been proposed, including the non-union hypothesis of Hildebrand, the failure of involution theory of Kampmeier, and hamartoma theory of Staemler. Osathanondh and Potter<sup>4)</sup> demonstrated that all polycystic kidneys arise from abnormalities in the formation of the fetal kidneys, and that the principle abnormality is in the collecting tubules; structure coming from the ureteral bud as a result of repeated dichotomous division. Therefore, it follows that the cysts are opened to the pelvis, although Lambert<sup>13)</sup>, from the study of serial reconstructions of the infantile type, concluded that the cysts are closed cavities consisting of distended nephrons which are isolated from the pelvis though the pelvis of the adult type communicates with nephrons. But it is, from only their conclusions, unclear whether the epithelial hyperplasia with stratification and papillary projections which are found in our case are primary, or secondary to cystic dilatation.

Osathanondh and Potter have divided polycystic kidneys into four types by micro-dissections. Type 1 is due to hyperplasia of interstitial portions of collecting tubules, and corresponds to infantile type, the differentiation into collecting tubules and nephrons is normal but there is hyperplasia of collecting tubules. It is interesting that the shape of the cysts is variable as collecting tubules branch: this takes the form of diverticular dilatation in the earliest generations of collecting tubules, saccular dilatation in the mid-generations, and diffuse dilatation of the last generation. In our presenting case the renal cysts are composed of two main types: one is vesicular or saccular, the other is

tubular or slender. The gromeuli remain normal. Type 1 is always bilateral and is fatal soon after birth. Intrahepatic bile ducts are invariably cystic. In presenting case, the histopathologic findings and clinical course are quite analogous to type 1.

Various congenital anomalies have been reported to associated with infantile polycystic kidney, but none of them have been reported consistently and repetitively. Lieberman et al.<sup>8)</sup> have summerized from the survey of the literatures, the frequency of various congenital malformations in 214 patients with infantile polycystic disease, and 91 patients of them have had various congenital anomalies which are genitourinary, pulmonary, cardiovascular, musculoskeletal, central nervous, and gastrointestinal. In their investigation, liver involvement has not been described as a separated anomaly because the hepatic changes of ductal dilatation and periportal fibrosis are a component of infantile polycystic disease. Pancreatic cysts are extremely rare. This case is associated with adenomyoma in the jejunum, it is quitey unclear whether this adenomyoma is fortuitiously associated or possibly feature of specific lesion, which is associated with dilated tubules. Infantile polycystic kidney may be irrelevant to the incidence of neoplasm.

This disease is fatal, a high percentage of the affected infant are stillborn, and the majority of those severely affected at birth die within 3 months, survaival may last for several years at the longest. The cause of early death is not always clear, although it is generally agreed that contributory cause of death is renal failure. But those patients die earlier than would be expected if death were from uremia<sup>4)</sup>. In the present case, laboratory studies was not performed. Respiratory embarassment by upward pressure due to the bilaterally enlarged kidneys might considerably contribute to the cause of death.

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Table 1

Cystic Disease of the Kidney in Children<sup>2)</sup>

- A. Renal dysplasia: (1) total, (2) segmental, (3) focal, and (4) associated with congenital obstruction.
- B. Polycystic renal disease: (1) adult type and (2) infantile type.
- C. Medullary cystic disease: (1) the sponge kidney and (2) uremic medullary cystic disease.
- D. Simple renal cysts.
- E. Multilocular renal cysts.
- F. Calyceal diverticula.
- G. Miscellaneous cyst of renal origin: (1) retroperitoneal cysts of nephric origin; and (2) dysontogenic cysts in the renal fossa; a. renal teratoderms. b. endometrial cysts of the kidney.
- H. Cysts of other than nephric origin (not necessarily developmental): (1) pericalyceal lymphangiectasis; (2) perinephric pseudocysts.

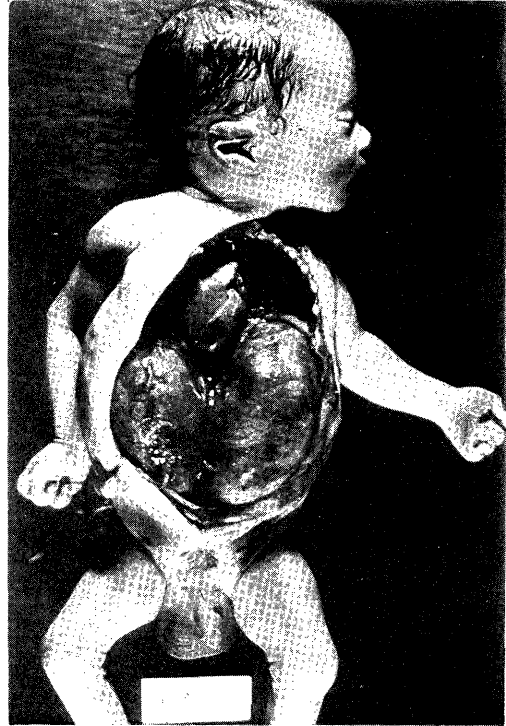


Fig. 1

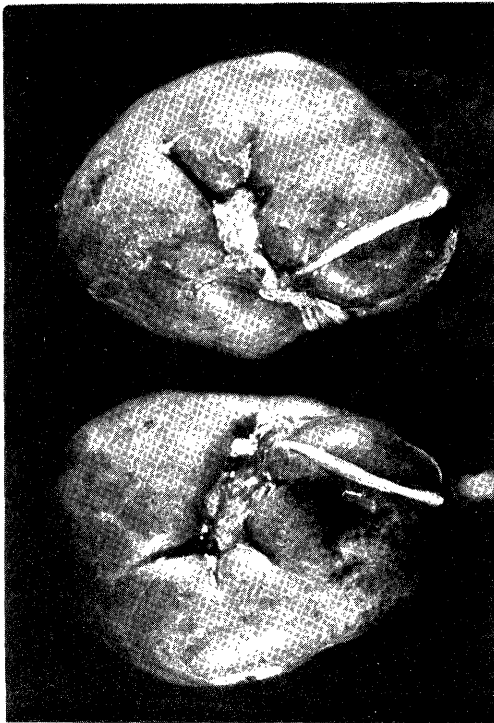


Fig. 2



Fig. 3

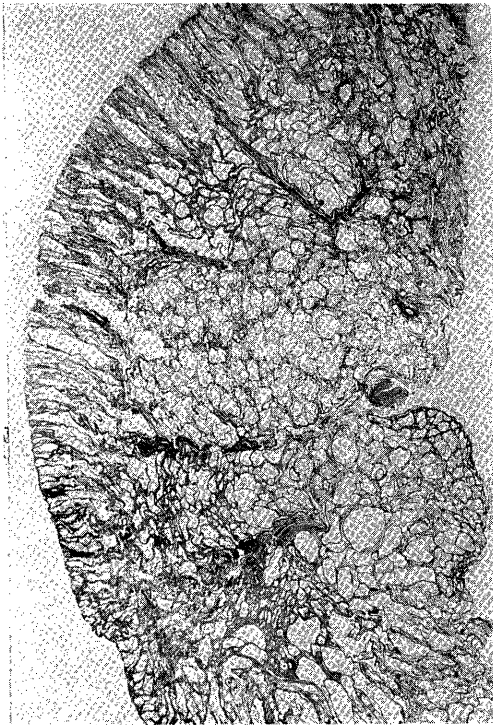


Fig. 4

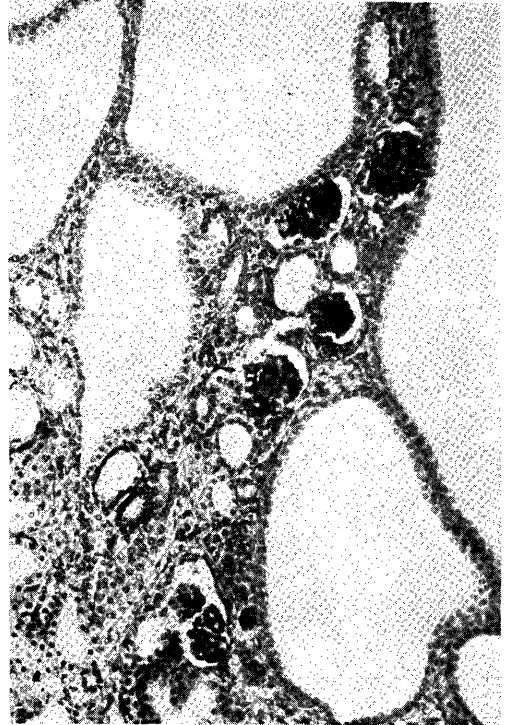


Fig. 5



Fig. 6

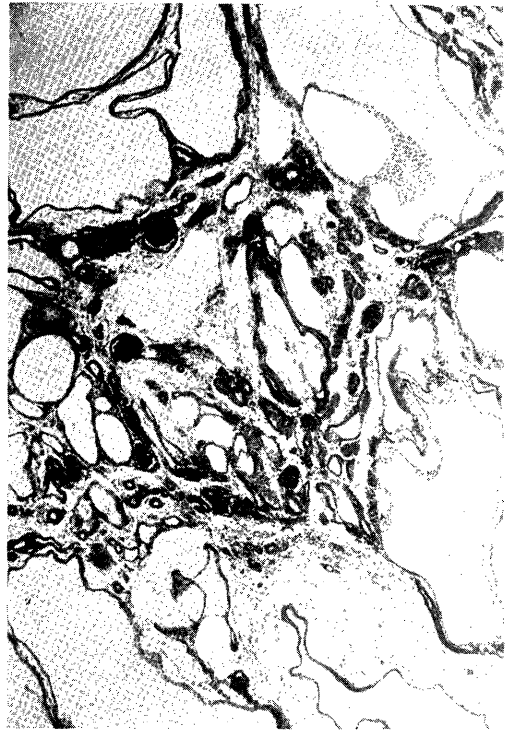


Fig. 7



Fig. 8



Fig. 9