# Polycystic kidneys and liver in two siblings with other severe congenital abnormalities

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**Introduction**

THE MOST IMPORTANT developmental disturbance in the structure of the renal parenchyma, the polycystic kidney, is clinically found in two forms, one in adults and the other in infants and children (Dalgaard, 1963).

Only 26 cases have been reported in which polycystic kidneys occurred in two or more siblings, and only five of polycystic kidneys and liver occurring in siblings. This is probably the first report of a sibling pair who had not only polycystic kidneys and liver, but also malformations involving the brain (anencephaly in one and encephalocoel in the other) and spleen (splenic agenesis in one and multiple spleniculi in the other) in addition to other minor abnormalities.

**Case Reports**

These two infants resulted from consecutive pregnancies. There was no consanguinity between the parents nor was there any history of abnormality, as far as could be determined, in the relatives of either partner. The mother was 24 years old and the father 28 years old when the second baby was delivered. Both parents appeared normal. Serologic examination for syphilis was negative. Chromosome studies revealed no abnormality.

**Case 1.**

The mother, a primigravida, was perfectly healthy during this pregnancy. She was noticed to have hydramnios when first seen at 28 weeks' gestation. At 37 weeks' gestation, it was realised that the foetal head was not easily palpable. Radiologic examination demonstrated microcephaly and an enlarged abdomen. Hydrops foetalis was suspected. Labour was induced 23 days after term. The head and shoulders were easily delivered, but the foetus became impacted because of a grossly enlarged abdomen. The abdomen had to be incised; two large spongy kidneys were removed, after which the rest of the body was delivered with no difficulty.

The foetus, a female, weighed 3.63 kg. with a crown heel length of 45 cm. External malformations included polydactyly, lobed tongue and a large encephalocoel. Internally, bilateral grossly enlarged spongy kidneys (wt. 791 gm.) and multiple spleniculi were seen. On histologic study, the kidney had only a few normal glomeruli, the major part being made up of dilated tubules and loose mesenchymal tissue. The liver (wt. 128 gm.) had multiple hamartomatous areas consisting of proliferated bile ducts, some of which showed cystic dilation with fibrosis. The spleen was normal in structure. Much of the enlarged tongue was infiltrated with fat.

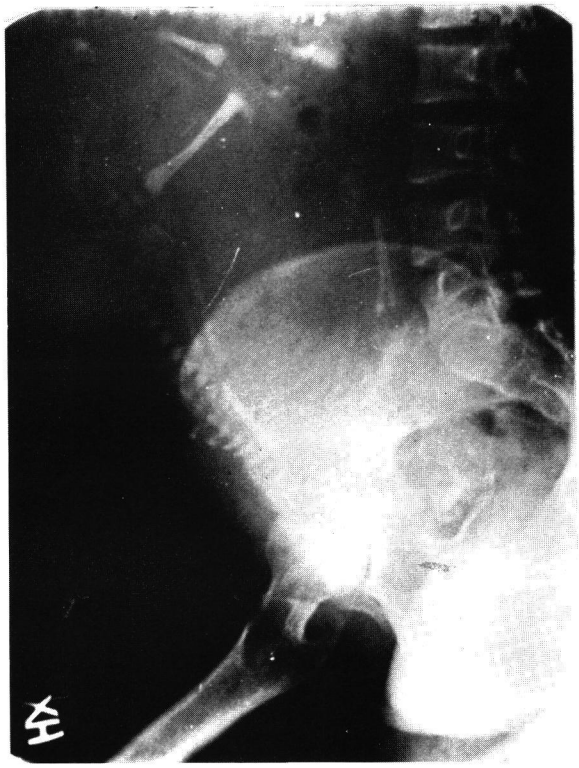

**Fig. 1: Case 2. X-ray of the abdomen at 36 weeks' gestation. The posture of the limbs of the foetus indicates a grossly enlarged abdomen, leading to a diagnosis of hydrops foetalis. Anencephaly was also present.**

**Fig. 2: Case 2. The two grossly enlarged spongy kidneys together with the enlarged congested liver are mainly responsible for the marked enlargement of the abdomen.**

**Case 2.**

Two years later, the mother became pregnant again; the pregnancy was uneventful. She presented with transverse lie which was corrected easily by external cephalic version. At 36 weeks' gestation, it was realised that the foetal head could not be felt. Radiographs revealed anencephaly and a "Buddha posture" indicating an enlarged abdomen. (Fig.1) Labour was induced, resulting in the delivery of a very feeble female baby who lived for only half an hour. The baby weighed 2.95 kg., had a crown heel length of 43 cm., and had anencephaly and a grossly enlarged abdomen, especially prominent in both flanks where two large craggy masses could be felt.

Autopsy revealed two grossly enlarged kidneys (wt. 659 gm.) on both flanks and occupying, with the liver, the major part of the abdomen. (Fig. 2). The kidneys appeared spongy with lobulated surfaces but having the general configuration of normal kidneys. The ureters and bladder were normal. The liver (wt.

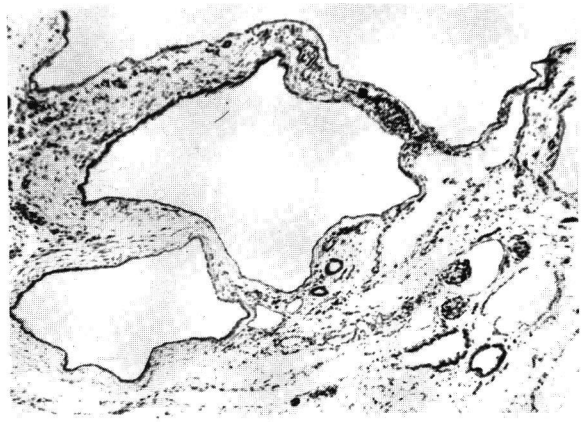

**Fig. 3: Case 2. Histology of the kidney (x 75). Cystic dilation of the tubules are seen. A few normal glomeruli are present scattered in different areas. (Haematoxylin and Eosin stain)**

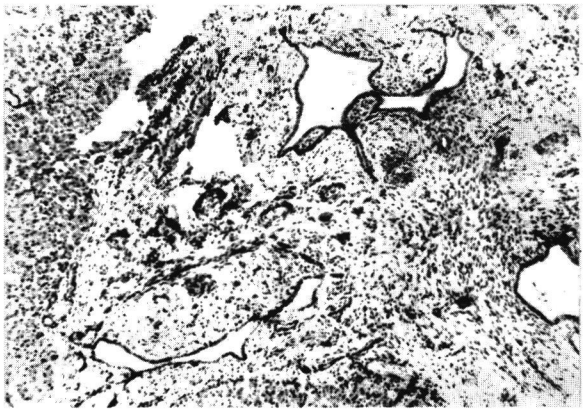

**Fig. 4: Case 2. Histology of the liver (x 150) showing the cysts and the connective tissue stroma in the portal areas. (Haematoxylin and Eosin stain)**

139 gm.) appeared enlarged and congested. No spleen could be detected. On microscopy, the kidney had multiple cysts consisting of dilation of the tubules. A few normal glomeruli were seen scattered in different areas. Mesenchymal tissue was abundant in the medullary area (Fig. 3). The liver had normal architecture with an increased amount of connective tissue and bile ducts in the portal areas. Some of the bile ducts showed cystic dilation (Fig. 4) very similar to that seen in the liver of the sibling.

## Discussion

Clinically, polycystic kidneys can be divided into two main groups, (though it is not as clear cut structurally), one occurring in adults and the other occurring in infants (Dalgaard, 1963, Bell, 1935, Fergusson, 1949) the former reaching a peak at the 5th – 6th decade of life and being usually inherited as a Mendelian dominant (Dalgaard, 1963). In contradistinction, polycystic kidney in infants is usually transmitted as an autosomal recessive (Dalgaard, 1963, Lundin et. al., 1959, Osathanondh, et. al., 1964, Greenberg et. al., 1967).

Polycystic kidneys occurring in infants is rare – the incidence varies from one in 219 autopsies (Roscher, 1933) to one in 448 autopsies (Dalgaard, 1957) and one in 6,000 births (Dalgaard, 1957) to one in 14,000 births (Book, 1951). Polycyctic livers accompanying polycystic kidneys is very well known since Bristowe (1856) first described it. It has been estimated that approximately ¼ to 1/5 of patients with polycystic kidneys have polycystic livers (Lathrop, 1959). Conversely, about half the patients with polycystic livers have polycystic kidneys (Comfort et. al., 1952). The liver cysts are due to distortion, segmentation, and dilatation of intrahepatic bild ducts, a process considered to be essentially degenerative, an abnormal extension of the process of resorption which occurs normally in the first generation of bile ducts (Comfort et. al., 1952, Norris et. al., 1947). Only four sibling pairs with polycystic kidneys and livers could be traced by Dalgaard (1963) though Lathrop (1959) reported a family with four, and probably six, or seven siblings being affected. The present sibling pair is therefore among the very few to present with these features.

That multiple deformities may accompany polycystic kidneys in infants has been well known since Gruber (1934) first described the association and managed to collect 16 examples of it. Smith et. al., (1965) reported a sibling pair with multiple developmental defects in addition to polycystic kidneys and liver. The present siblings presented with major abnormalities in addition to polycystic kidneys and liver – this is probably among the very few, if not the first report, of a sibling pair with such gross abnormalities.

According to the classification on pathogenesis of Osanathanondh and Potter (1964), the present two cases would fit into Type 1, which is due to hyperplasia of interstitial portions of collecting tubules – this type appears to be incompatible with prolonged survival, invariably bilateral, accompanied by cystic proliferation of bile ducts in the liver and seems to be the only variety seen in siblings. However, some cases have survived beyond the first decade of life (Lathrop, 1959). In Lundin and Olow's (1959) classification, the present cases would fit into Group 1 – they state that cysts in the pancreas and liver occur in 50 percent of such cases.

The condition reported here would appear to be of hereditary origin, probably an autosomal recessive.


## Summary

A sibling pair is presented who, in addition to polycystic kidneys and liver, also had major abnormalities affecting similar organs in other systems which were not compatible with life. This syndrome with polycystic kidneys is probably transmitted as an autosomal recessive trait.



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