

Two Cases of Polycystic Kidneys in Two Siblings

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The kidneys that have several cysts are called polycystic, but the real polycystic disease of the kidneys is a special entity, and should not be mistaken with cystic hypoplasia of the kidneys; solitary cysts of the kidney and multi-cystic kidney, because they are different macroscopically and microscopically.

This disease is a general lesion in which the whole kidney tissue is cystic and affected. The cysts are similar, from 1 to 5cm. in diameter and the kidney has a spongy appearance. This disease is nearly always bilateral and the unilateral form is very rare. The size of the kidneys differ. Sometimes they are smaller than a normal kidney and sometimes they are so big that the vaginal delivery is difficult or even impossible.

The infant may die before or after birth.

In the adults, the signs of the disease appear about the age of 30. In the mild congenital forms, the signs of renal insufficiency appear in the first few months of the baby's life. The smallest fetus reported with this disease was 35 grams and had the age of 14 weeks. (Bell)

Sometimes cysts are found in the liver, lungs, pancreas and spleen. The liver may be larger than normal with no cysts in it (our second case). Polycactilism, hydrocephalus and congenital heart malformations are also observed.

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This disease is found in siblings and sometimes it is seen in a few generations of the family. It is a hereditary disease that in the infantile form is transmitted recessively and in the adult form dominantly.

As mentioned before, the disease has two forms:

1. The adult form which is not discussed here.
2. The infantile form which is divided into two groups. (Potter)
 - (A) In this group the kidney is larger than normal having preserved its original shape macroscopically. In the sections of the kidney cysts are observed and the kidney has a spongy appearance (our first case). Microscopically the amount of connective tissue is not increased. The shape and the size of the cysts are similar. The cysts are formed from the lengthening and widening of the tubules or from the dilatation of the internal space of the Bowman's capsule. The vascular structure of the glomerules is normal and the nephrons are not reduced.
 - (B) In this group the kidneys may be smaller than normal. They are hard and have the shape of a normal kidney (second case). In sections the cysts are spherical and different in size. Their origin is from the tubules. Microscopically the glomerules are abnormal and the nephrons are reduced. The amount of connective tissue is more than normal.

In both forms the ureters are normal and the changes in the pelvis are due to the pressure of the cystic kidney.

Case Reports:

Mrs. R. G., 34 years old from Teheran, referred to the hospital with abdominal pain on March 14, 1966. She insisted that she was pregnant and in her seventh month but the uterus was larger than a normal term pregnancy. The dilatation was 2cm. and the effacement was 80%. The bag of water was intact. The presentation was breech. On 3/16/66 the abdominal x-ray was normal. On 3/17/66 because a vaginal delivery was impossible, a cesarean section was performed. She had oligoamnios and the infant was 4200 grams with a big abdomen,

did not breathe and died after birth.

Autopsy revealed a large abdomen with internal pressure, when opened two big bean shaped tumors were observed at the site of the kidneys. The kidney tissue was spongy and their sections were full of similar round or elliptic cysts. (Figure 1)

In microscopic view a cystic dysplasia of the kidney from the form "Potter A" was observed (Figure 2). The ureters and the bladder were normal. The lungs were also normal.

We started investigation about the couple. They were cousins. The laboratory analysis performed for the couple were all normal.

Case 2:

This patient was referred to the hospital on 9/2/69 infertility and with treatment became pregnant on 10/20/69. The pregnancy was normal until the seventh month when the uterus started to enlarge abnormally, and the amniotic fluid diminished. On 7/1/70 an abdominal x-ray was taken which showed a vertex presentation and the skull bones were overlapped. The vertebral column was angulated in the lumber area which was suspected of a fetal death, but the fetal heart was normal. The patient underwent a Cesarean section on 7/8/70. She had oligoamniosis. The enfant was a female weighing 3500 grams. The skull bones were soft with mild hydrocephaly. There were malformations in the vertebral column and the legs. The child died.

Autopsy revealed the kidneys to be larger than normal but they did not have the shape of a kidney. Macroscopically the consistency and the appearance of the kidney sections was like connective tissue. In microscopic view the connective interstitial tissue had increased and in some parts it looked like normal connective tissue and in other parts it was like hyalinized connective tissue. The tubules were reduced and some of them were cystic and seldom glomerular structures were seen (Fig. 3 & 4). This is like group "Potter B".

Pathogenesis:

The pathogenesis of this disease is not quite clear. There are different theories but none are proved. Some authors believe that during the foetal life the ureteral eminence had not joined the nephrotic eminence and there is no connection between the nephron and the excretory system and thus the proximal part becomes cystic. There is no explanation for this hypothesis when there are cysts in other organs too.

Discussion:

As we mentioned before, the polycystic disease of the kidneys is a hereditary disease which is transmitted recessively in the infantile form and dominantly in the adult form.

The interesting point about our patient is that the two forms of group A and B (Potter) are seen in the siblings of a family. A question arises whether these two forms of this disease are resulted from one genetic factor or are two different diseases caused by two different genetic factors?

The next question is whether the newborns of this mother in her future pregnancies will have the same disease or not?

Summary

Polycystic disease of the kidneys is a generalized lesion of characteristic clinico-pathological entity. It has two clinical forms: adult and infantile. The adult type is transmitted as a dominant disease where the latter is a recessive trait. The infantile form had two pathological patterns. Form "A" in which the kidney is large and spongy, of normal shape and the cysts are numerous. Microscopically the connective tissue is not increased. The cysts are of equal size and shape. The nephrons are not reduced. In the form "B" the kidney may not have a normal shape and is not as large as the form "A". Microscopically the connective tissue is increased intensively. The cysts are of spherical shape and different sizes. The nephrons are reduced in number.

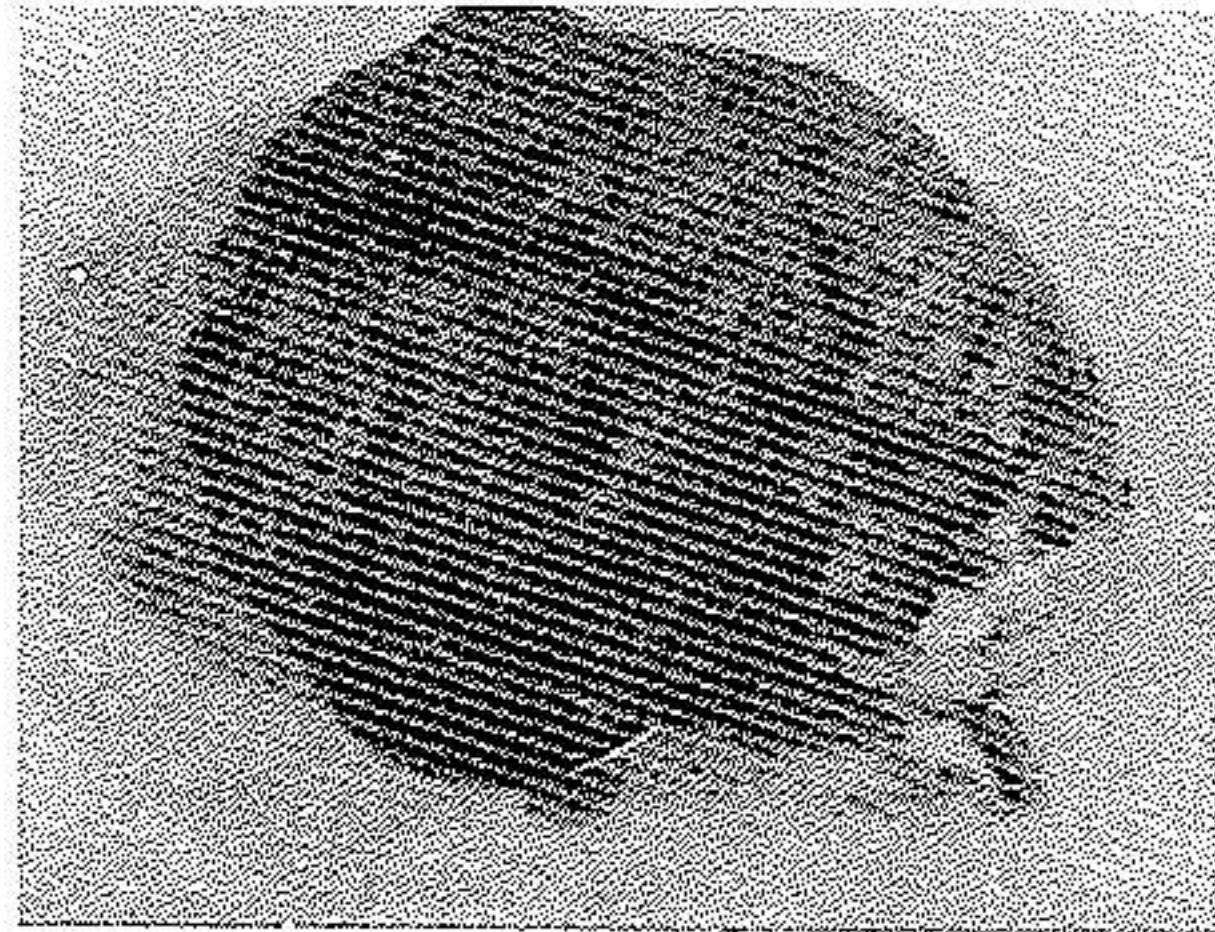


Fig. 1- Large kidney with spongy appearance and numerous cysts

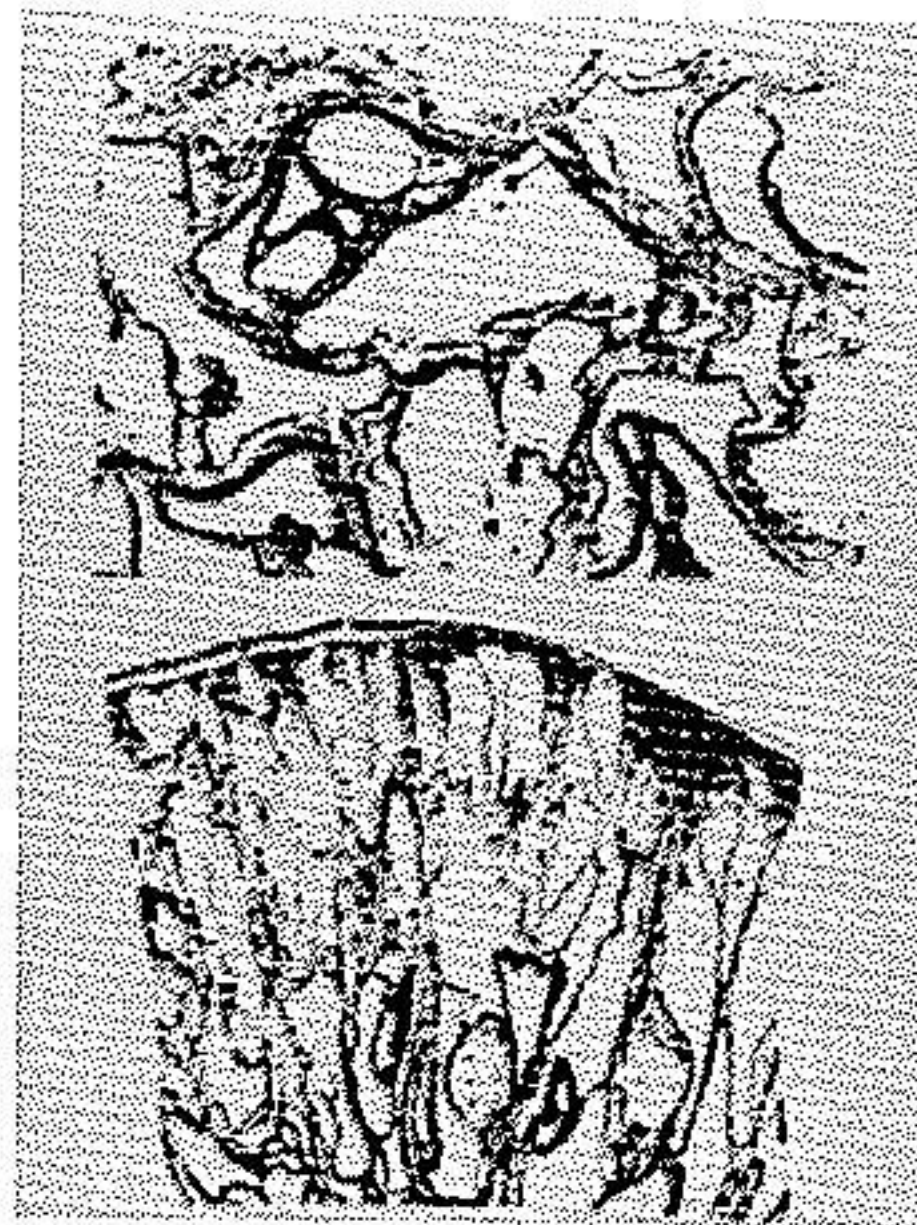


Fig. 1- The glomerules are normal. The tubules are dilated and cystic and are lined by a layer of flat cells. The connective tissue is not increased.

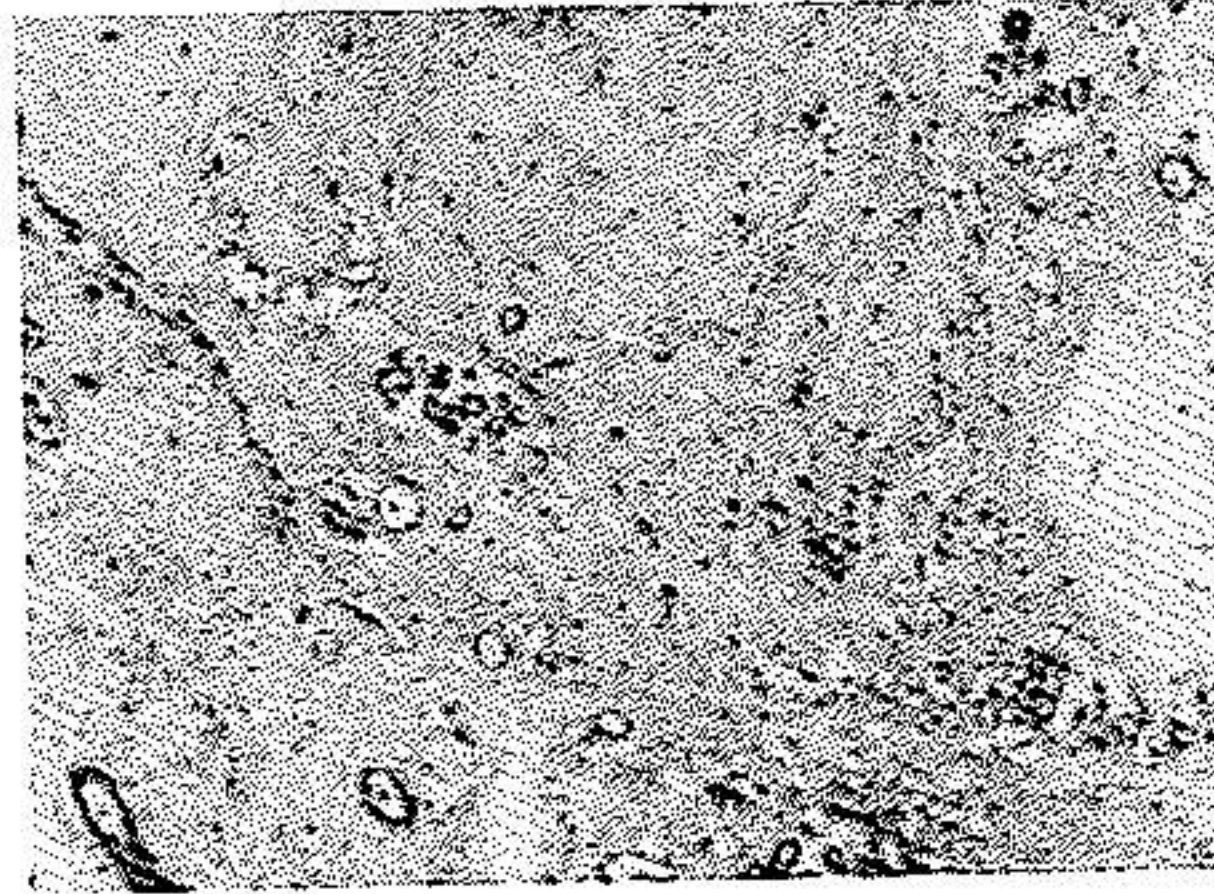


Fig. 3- The connective tissue is highly increased. Small number of tubules some of which are cystic are seen. Scarcely a glomerule can be found.

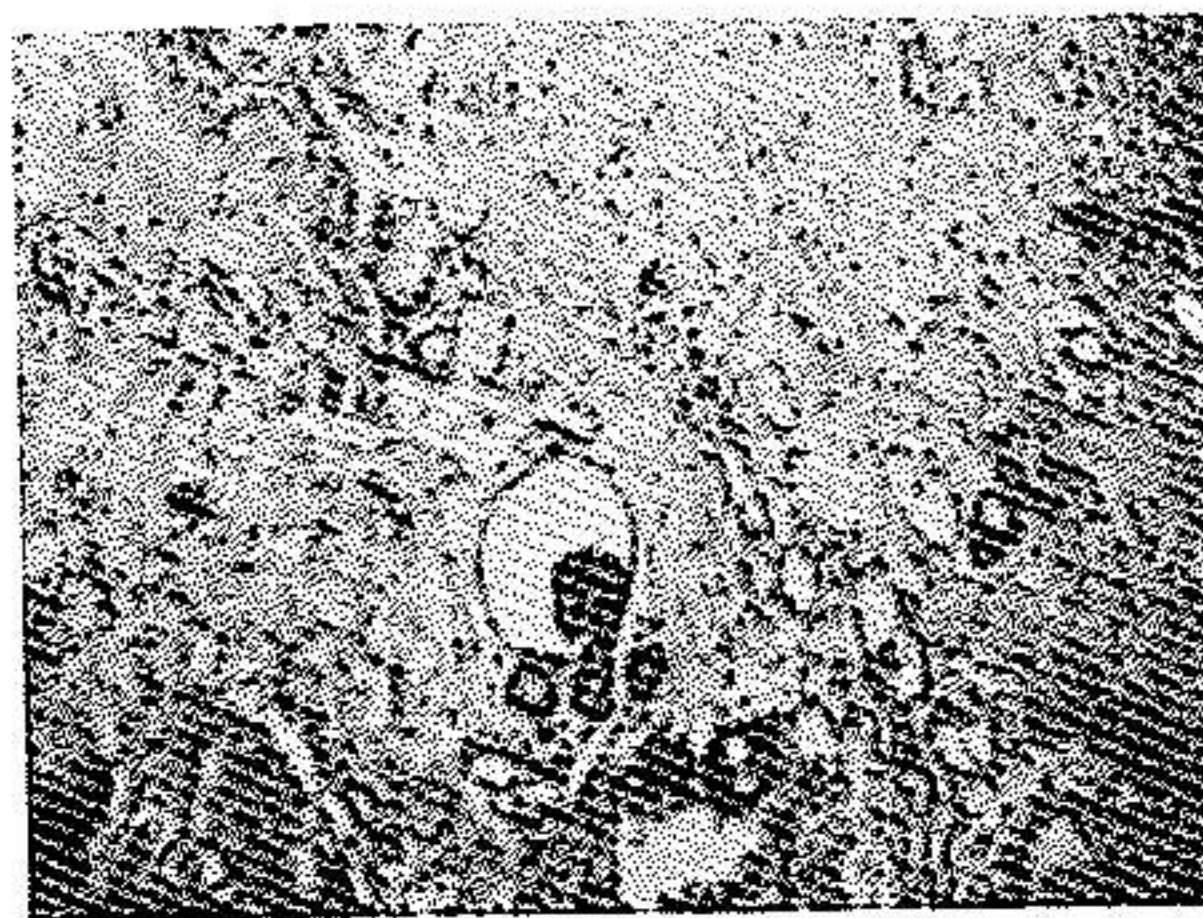


Fig. 4- A part of the last slide with higher magnification. In the middle a glomerule with distended capsule is seen. Stromal hyalinized connective tissue with collagenous fibers is also observed.

We are reporting two cases of infantile polycystic kidneys in two siblings, one of whom represented form "A" and the other was similar to form "B". These two cases are interesting from the genetical point of view. We want to know whether the two forms are different manifestations of one disease with one genetic factor or they are two different diseases with two genetic factors?

One can assume that the infantile polycystic kidney is a genetical disease with two different pathological manifestations or two different genetic disorders due to two different genetic factors.

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