PRENATAL DIAGNOSIS OF INFANTILE POLYCYSTIC KIDNEYS: A CASE REPORT

S. RAMAN
S. P. RACHAGAN
P. BOOPALAN
S. JEYARANI

SUMMARY

A case of infantile polycystic kidneys diagnosed prenatally by ultrasound is presented here. This condition was confirmed at post-mortem following delivery of the child. The clinical and pathological features of this inheritable disease is discussed.

INTRODUCTION

Infantile polycystic kidneys is an uncommon condition which is invariably lethal. Being an autosomal recessive condition, the recurrence rate is 25%. This condition may be diagnosed by ultrasonography after the twentieth week of gestation. Early diagnosis can help the clinician to adequately counsel the parents regarding the prognosis of the ongoing pregnancy and also inform them of the risk of recurrence in subsequent pregnancies.

S. Raman, AM, MBBS, MRCOG, FICS
S. P. Rachagan, AM, MBBS, MRCOG
Department of Obstetrics & Gynaecology
Faculty of Medicine
University of Malaya
59100 Kuala Lumpur

P. Boopalan, AM, MBBS, FRCOG, FICS
Consultant Obstetrician

S. Jeyarani, MBBS
Department of Pathology
Faculty of Medicine
University of Malaya
59100 Kuala Lumpur

CASE HISTORY

A 26-year-old Gravid 2 Para 1 was referred to the University Hospital, Kuala Lumpur at 36 weeks of gestation because of oligohydramnios and 'large kidneys' visualised during antenatal ultrasonography. She had been followed up by the referring doctor from early pregnancy and serial ultrasound scans revealed that the kidneys were progressively getting larger in size.

In her past obstetric history, she delivered vaginally a grossly hydropic baby at 33 weeks of gestation. This baby died shortly after birth. No investigations were done on this baby. Both parents were investigated but no abnormality was detected.

There was no known family history of any foetal abnormality. The marriage was non-consanguinous. On examination, there was no pallor. The blood pressure was 120/80 and the pulse was 84 per minute. There was no abnormality detected in the cardiovascular and respiratory systems.

Abdominal examination revealed a 34-weeks-sized pregnancy. A single foetus in longitudinal lie and cephalic presentation was noted. There

was clinical oligohydramnios. The foetal heart rate was 140 per minute and regular.

Ultrasound examination confirmed a single foetus in cephalic presentation. There was marked decrease in liquor volume. The obvious abnormality noted was in the foetal kidneys which was echogenic and bilaterally enlarged being 8.5 x 4.5 cm each (Fig. 1). Both kidneys together occupied half the abdominal circumference. The foetal bladder was not visualised. The foetal liver appeared normal. The placenta was in the upper segment. A diagnosis of infantile polycystic kidneys was made.

Both parents were jointly counselled by the Obstetrician and Paediatrician regarding the poor prognosis and subsequent recurrence rate.

Labour was induced with prostaglandin vaginal pessaries and she subsequently delivered vaginally a male baby weighing 3,010g with an Apgar score of 9/6. There was no abnormal facies apart from the low set ears. The air entry in both lungs was poor. On abdominal examination, the liver was smooth and just palpable. The kidneys were bilaterally enlarged and ballotable. No other abnormalities were noted. The condition of the baby deteriorated rapidly and death occurred three hours after delivery.

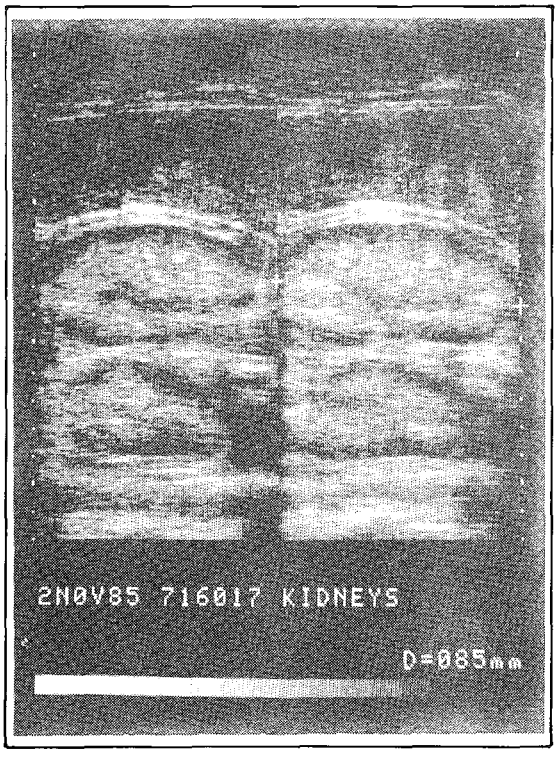

Fig. 1 Ultrasound photo of the foetal abdomen showing echogenic and enlarged kidneys (between crosses) (2 exposures taken together).

Gross Pathology

The kidneys were enlarged bilaterally, and were smooth and spongy to the touch (Fig. 2). The right and left kidneys weighed 80g and 86g respectively (normal weight in relation to body weight is 25 ± 5g). Cut surfaces of both kidneys showed fusiform dilatations running radially through the cortex and medulla. The pelvicalyseal system was normal in shape. The ureters and bladder were normal.

The right and left lungs appeared hypoplastic and weighed 12.0g and 11.5g respectively. (Normal weight of both lungs in a foetus of 36 weeks gestation is 55g.) Both were unremarkable except for prominent bronchial markings. No cysts were seen.

The liver weighed 118g and was grossly normal in appearance.

Histology

Sections of the kidneys showed enormously dilated collecting tubules lined by cuboidal and flattened epithelial cells (Fig. 3). The tubules were arranged parallel to each other and perpendicular to the capsular surface. Normal nephron elements were present between the tubules. The findings were consistent with those seen in infantile polycystic kidneys.

A section of the liver showed an increase in fibrous tissue in the portal tracts with a striking increase in the number of bile ducts. Some of these ducts exhibited curious angulated branching. They were lined by columnar epithelial cells. Occasional cystically-dilated ducts were noted. The hepatocytes were essentially normal. This

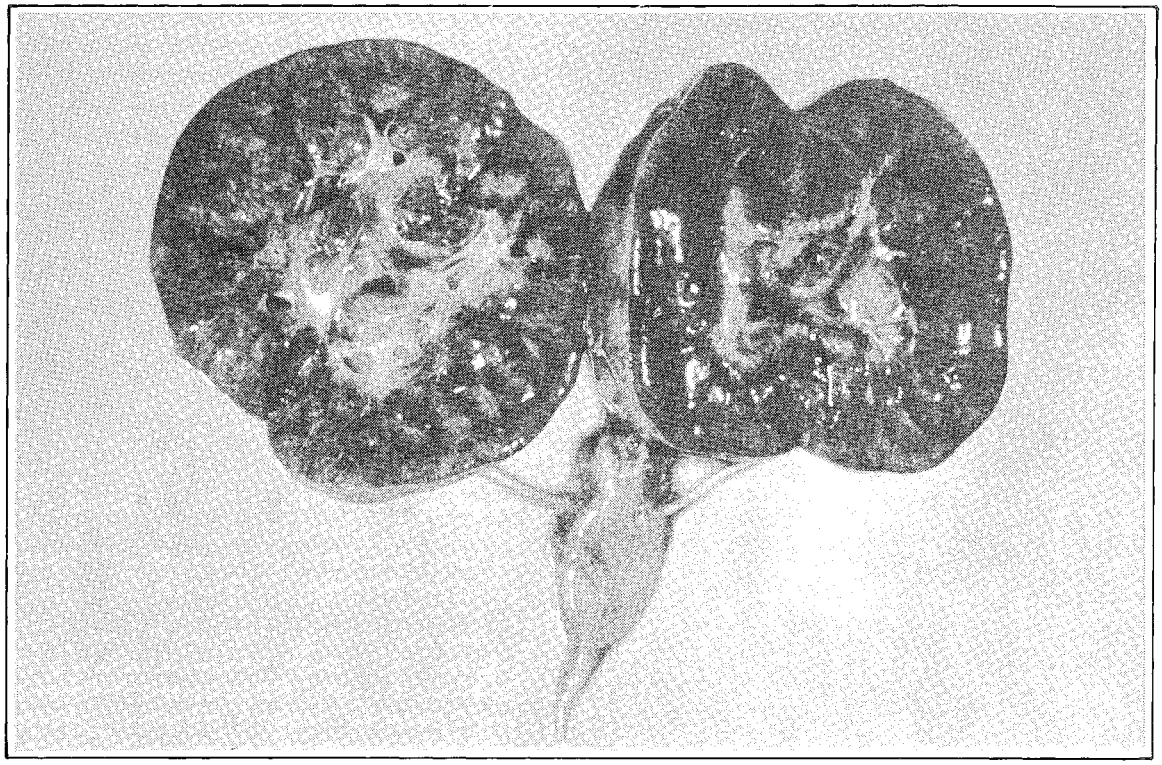

Fig. 2 Post-mortem photo of the enlarged kidneys with their ureters and small bladder.

finding was consistent with congenital hepatic fibrosis.

Histologically, the lungs were generally congested and many of the alveoli were collapsed. No obvious pathology was noted.

DISCUSSION

Infantile polycystic kidneys occur in about 5% of all abdominal masses in the foetus. This condition is an autosomal recessive one and is associated with hypoplastic lungs,[2] congestive heart failure, Potter's facies[1] and encephalocele.[2] The recurrence rate of infantile polycystic kidneys alone is 25%.

This conditon is caused by dilatation and hyperplasia of collecting tubules causing microcysts leading to large kidneys. Being almost always bilateral, it is generally incompatible with life beyond the neonatal period.[1]

This condition may be diagnosed by ultrasonographic examination even before the 20th week of gestation.[2] The microcysts give a dense echogenic appearance during ultrasonographic examination. The kidneys are also enlarged for their gestation and the kidney circumference/abdominal circumference ratio is increased. Oligohydramnios is a known finding in these patients and was also noted in our patient. The bladder and ureters are normal as in our case.

Foetuses with infantile polycystic kidneys have a condition called congenital hepatic fibrosis in the liver. This is a constant feature and was also noted in our patient's baby. The causation and significance of this associated condition is not known.

In this patient with a previous perinatal death from hydrops fetalis of unknown cause and infantile polycystic kidneys in this pregnancy, the

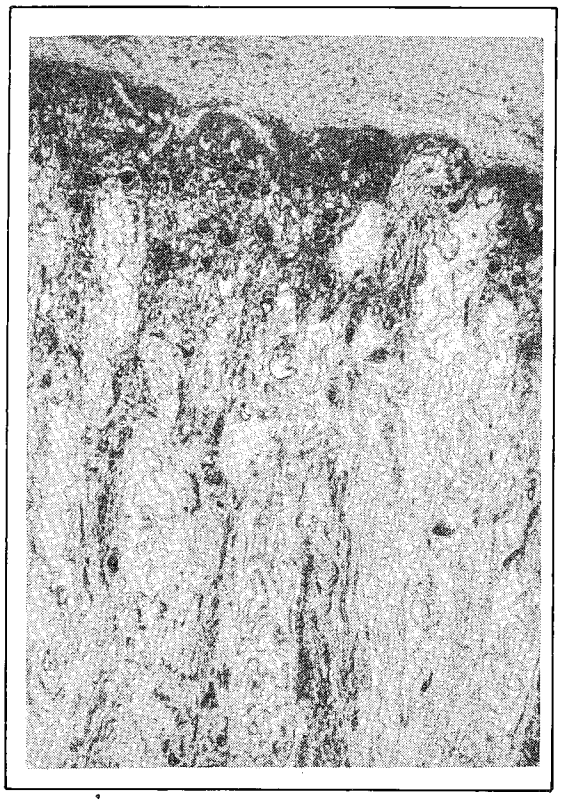

Fig. 3 Section of the kidney showing enormously dilated collecting tubules lined by cuboidal and flattened epithelial cells (x 250).

chances of delivering a normal baby is low. Both the parents were counselled regarding this and told to come for early antenatal care if they decide to embark on any future pregnancies. Serial ultrasonographic examinations could detect any recurrence of either condition and would be useful in managing the pregnancy.

REFERENCES

[1] Kurjak A, Latin V, Mandruzzato G, D'Addario V, Rajhvajin B. Ultrasound diagnosis and perinatal management of foetal genito-urinary abnormalities. *J Perinatal Med* 1984; 12 : 291–312.

[2] Hobbies J C, Winsberg F, Berkowitz R L. Normal and abnormal foetal anatomy. In Hobbies J C, Winsberg F, Berkowitz R L (eds). *Ultrasonography in Obstetrics and Gynaecology.* Baltimore: Williams and Williams, 1983 : 152.

[3] Pearce J M, Campbell S. The prenatal diagnosis of foetal urinary tract anomalies. In Rodeck C H, Nicolaides K H (eds). *Prenatal Diagnosis.* Chichester: John Wiley and Sons, 1983 : 317–318.