

### Prenatal Diagnosis of Meckel's Syndrome

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The identification of invariably lethal anomalies in the prenatal period gives an option of early therapeutic abortion. The infantile polycystic disease of the kidneys in its severe form which presents as Meckel's syndrome is a potentially lethal condition. This can be reliably diagnosed in the antenatal period as illustrated here.

#### Case Report

A 24-year-old sixth gravida was referred for ultrasound scanning for identifying the presenting part. The details about the two intrauterine deaths which she had in the third trimester were not known. The present pregnancy was of 34 weeks duration and the uterine size was corresponding to the period of amenorrhea. There was no history of consanguinity, diabetes, significant maternal infection or exposure to teratogens.

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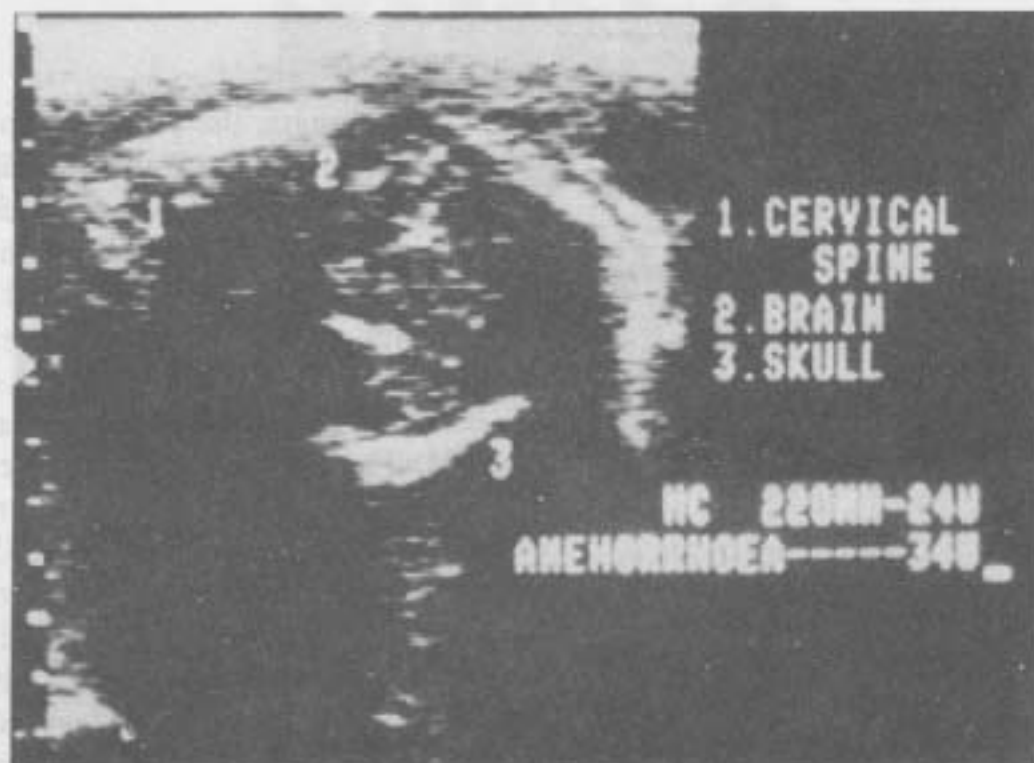
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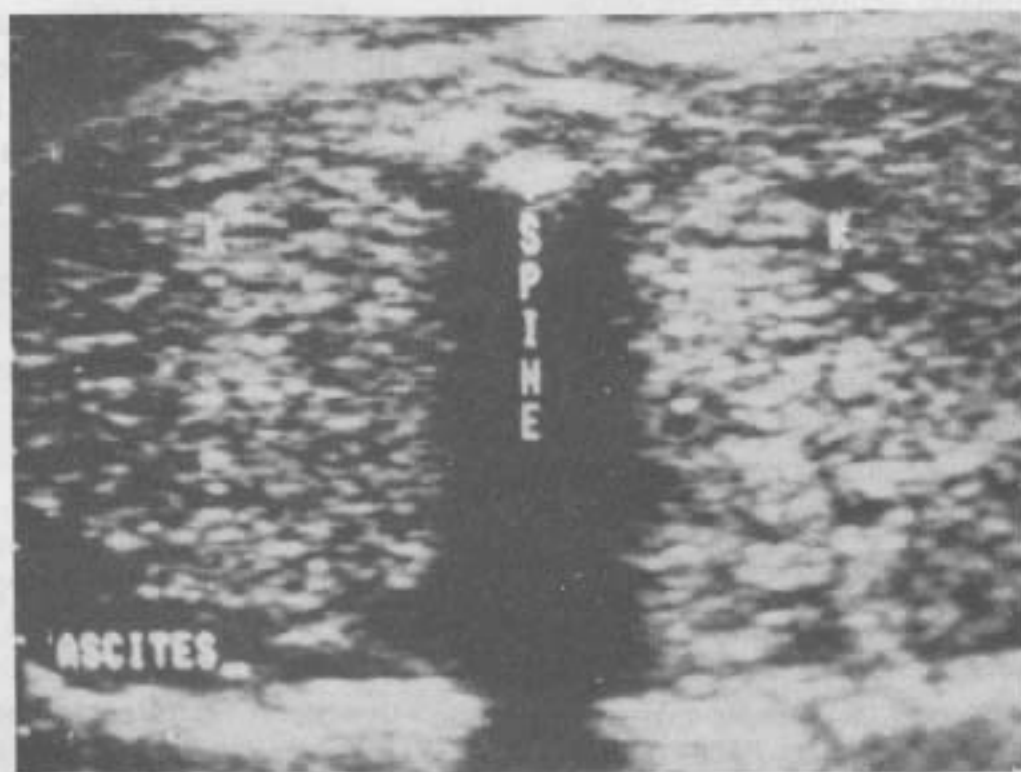
On scanning, the placental appearance was normal and the volume of liquor was sufficient. A female fetus was seen with its head in the uterine fundal region. The biparietal diameter was 56 mm and the head perimeter was 220 mm; all corresponding to an average size of 24 weeks (*Fig. 1*). There was no hydrocephalus or paracranial cysts. The spinal echo features were normal. The abdominal perimeter was unusually more to the extent of 355 mm by the presence of massively enlarged kidneys along with ascites (*Fig. 2*). The kidneys were occupying approximately 90% of the area in the cross sectional view of the abdomen. The renal parenchyma was abnormally more echoic (*Fig. 3*). Multiple small cystic spaces were seen in it. No pelvic dilatation was evident. Irrespective of repeated attempts the urinary bladder could not be seen. The liver was seen enlarged due to the presence of 3 cysts (*Fig. 4*) in it. The cardiac and the lung parenchymal features were normal. The limb bones were well formed and normal (femur 62 mm, humerus 54 mm). The estimated gestational age was around 32-34 weeks. Polydactyly could not be demonstrated.

#### Discussion

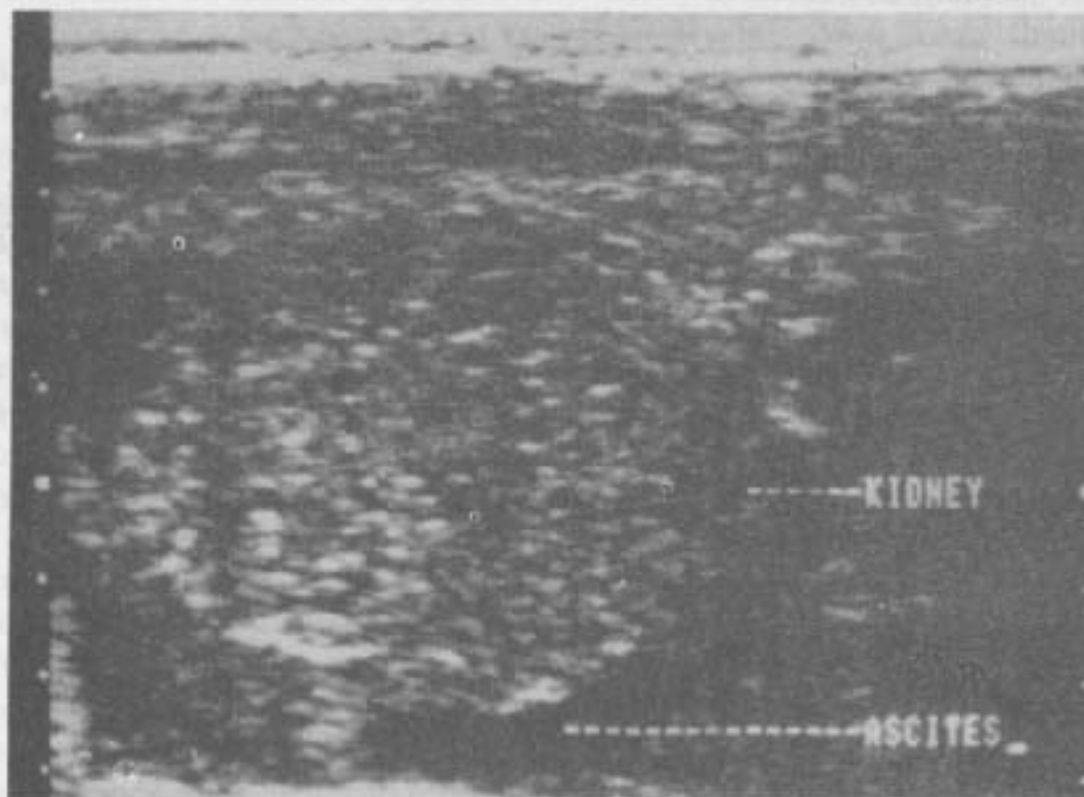
The occurrence of infantile polycystic kidney is about 1 in 50000, as reported by Potter(1). This has an autosomal recessive inheritance and manifests with different grades of severity with a recurrence rate of 25%. The severe variety can be diagnosed in the antenatal period and mild degree of involvement may manifest in due course. A defective collecting duct system is respon-



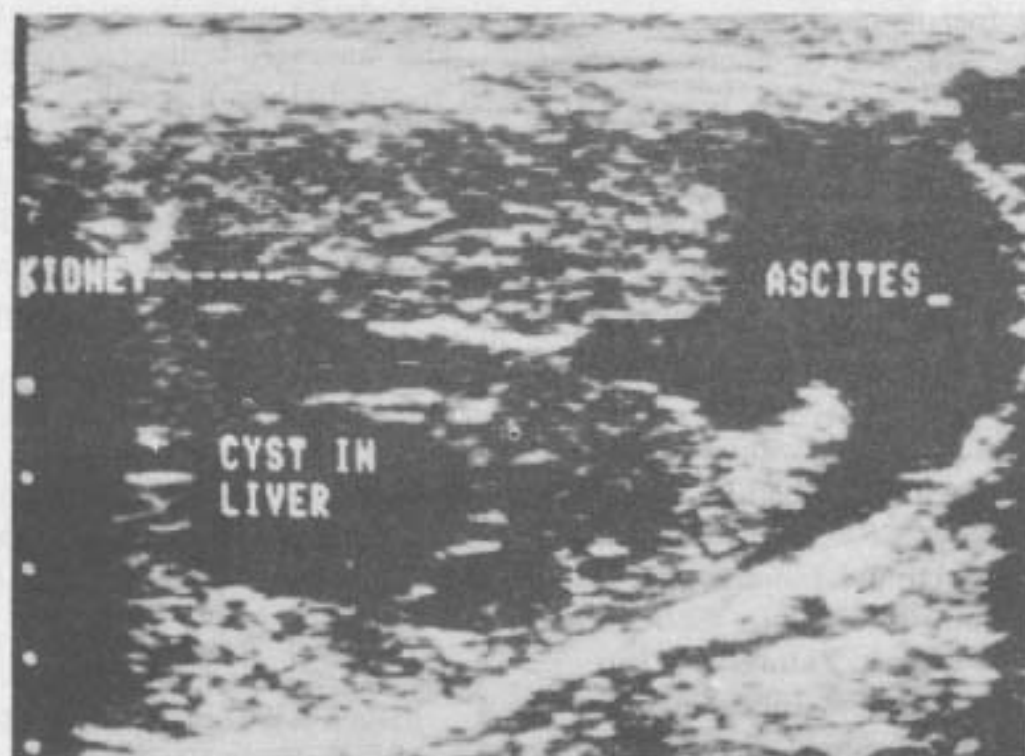
*Fig. 1. The axial section of the head at the plane of biparietal diameter showing microcephaly.*



*Fig. 2. The transverse section of the abdomen at the level of the kidneys in which the renal tissue is almost filling the abdomen.*



*Fig. 3. Enlarged view of a kidney with the hyperechoic parenchyma filled with multiple small cysts.*



*Fig. 4. Longitudinal section of the abdomen showing liver cysts and ascites.*

sible for this anomaly, as a result of which multiple small cysts occupy the renal parenchyma making it hyperechoic. The condition is to be strongly suspected when there is either a family history of similar disease or when the maternal serum or amniotic fluid level of alfa-fetoprotein is raised(2).

Out of the 4 groups of presentation of this disease, the commonest is the perinatal type; the other groups are neonatal, infantile and juvenile(3). Here, the renal failure starts in the intrauterine life and often ends in neonatal death. The presence of cystic changes in the liver without the involvement of the hepatocytes is usually an associated anomaly(4).

The antenatal ultrasound features of infantile polycystic kidneys are the presence of bilateral massively enlarged kidneys, absence of the fetal bladder and associated oligohydramnios(4,5). These features were typically present in this condition except for oligohydramnios. Apart from this, the presence of microcephaly and cysts in the liver were typical of Meckel's syndrome(6). There was no encephalocele in this case, which is usually noted in this syndrome. The presence of ascites could not be explained. Similar variability of the presentation was often noted by others(7). The postabortal fetus showed polydactyly.

Apart from Meckel's syndrome many other conditions are described in association with renal cysts; such as tuberous sclerosis, Von Hippel-Lindau disease (autosomal dominant), Jeune's asphyxiating thoracic dystrophy, Zellweger's cerebrohepatorenal syndrome (autosomal recessive), Orofacial digital syndrome Type 1, (X linked) and some other chromosomal disorders (Trisomy D, Down's syndrome and Turner's syndrome(8)). The ultra-

sonographic features of these entities are yet to be established.

In the differential diagnoses, the autosomal dominant adult polycystic kidney disease has to be considered first(6). The echo features are almost the same in these cases but massive enlargement of the kidneys is unusual. Moreover, parental screening may show evidence of similar illness. Multicystic kidney disease is characterized by the presence of multiple peripheral cysts of varying size sometimes causing confusion with the diagnosis of pelviureteric junction obstruction.

The head size was 5 SD below the mean which was typical of microcephaly. There was no hydrocephalus in this case which is otherwise a usual association. Though ascites was evident there was no evidence of hydrops. The anomalies noted in this fetus is a contradiction to the opinion expressed by some authors(10,11) that polycystic kidney disease is rarely associated with multiple anomalies.

This anomaly is invariably lethal and soft tissue dystocia due to the enormously enlarged kidneys is to be anticipated at the time of delivery if pregnancy proceeds. Hence, therapeutic abortion was done. The post abortal appearance and the histopathological findings of the kidneys verified the antenatal diagnosis.

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## **Pulmonary Atresia with Intact Ventricular Septum and Large Right ventricle as a Cause of Neonatal Heart Failure**

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Pulmonary atresia with intact ventricular septum is an uncommon but serious cardiac malformation and constitutes upto 2.5% of all structural cardiac defects(1). It is usually associated with a hypoplastic right ventricular cavity, however cases with large right ventricular cavity have been reported in literature and these cases have been classified into Type I and Type II depending upon the right ventricular cavity size(2-4). Type II cases with a normal or dilated right ventricular cavity constitute about 15-20% cases of pulmonary atresia with intact ventricular septum(5-9) and may be associated with Ebstein like anomaly of tricuspid valve(6) or even absent tricuspid valve leaflets(7).

We report a rare case of pulmonary atresia with large right ventricular cavity causing heart failure in the neonatal period.

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