CASE REPORT

RECURRENT FETAL POLYCYSTIC KIDNEY DISEASE

Sadia Anwar, Sajjad Ahmad

Department of Gyaecology and Obstetrics, Women Medical College, Abbottabad
and Department of Pathology, Gomal Medical College, D.I.Khan, Pakistan

ABSTRACT

Fetal genitourinary abnormalities consist of a wide spectrum of heterogenous malformations. Serial ultrasound evaluation starting from 15 weeks of gestation can be used as a screening modality. We present a case of 30 year old lady who presented with recurrent autosomal recessive polycystic kidney disease (ARPKD) diagnosed by ultrasound in consecutive pregnancies.

KEY WORDS: Polycystic kidney disease, Fetus, Genitourinary abnormalities.

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INTRODUCTION

Fetal genitourinary abnormalities consist of a wide spectrum of heterogenous malformations. The hyperechogenicity of the kidneys can be diagnosed after 17 weeks of gestation with results from the presence of multiple micro cysts, dysplasia or tubular dilatation. If there are no other malformations in the fetus the main diagnosis is polycystic kidney disease recessive or dominant.

Urinary tract dilatations and kidney abnormalities are relatively common with prevalence ranging from 1:250 to 1:1000 deliveries.[1] Majority of genitourinary abnormalities are detected during routine screening sonography in the absence of any significant clinical or family history. The diagnosis of a lethal form becomes evident with severe oligohydramnios and patent dilatation of urinary tract.

It can also be the part of chromosomal abnormalities such as trisomy 13. Autosomal recessive polycystic kidney disease (ARPKD) is a common inheritable cystic renal disease that has a profound effect on the growing fetus and on subsequent pregnancies, being fatal in 30% to 50% cases in the neonatal period. Prenatal imaging studies are suggestive of the disorder only from the second trimester onwards. ARPKD can be accurately diagnosed on characteristic histopathological features if an autopsy is performed in cases of infant death where the prenatal imaging studies and clinical findings are suggestive of the disease.[2] The prognosis of these antenataly detected cases is poor with death occurring within the first two months due to uremia or respiratory failure.[3] We present the features of autosomal recessive polycystic kidney disease (ARPKD) diagnosed antenatally by ultrasound.

Corresponding author:
Sajjad Ahmad Baloch, Assistant Professor
Department of Pathology
Gomal Medical College, D.I.Khan, Pakistan
e-mail: sajjadabaloch@gmail.com

CASE HISTORY

A 30 year old gravida 2 with previous normal deliveries came to Punjab Social Security Hospital, Lahore on 20th October 2011 for routine antenatal checkup at 35 weeks of gestation. It was her first antenatal visit. After routine checkup her obstetrical ultrasound was performed which revealed single alive fetus with breach presentation having biparietal diameter (BPD) of 8.23 cm=gestational age 33 weeks and femur length of 6.73cm=gestational age 34.4 weeks. Fetal abdomen was filled by kidney shaped hyperechogenic masses pushing up the diaphragm and distorting the thoracic anatomy. Liquor was scanty, amniotic fluid index (AFI) less than 8cm, placenta fundal posterior and heavy (figure 1). The picture was suggestive of polycystic kidney disease. The patient was counselled for termination of pregnancy due to poor prognosis of baby. She was induced with vaginal misoprostol and delivered a female baby of 2.5kg. The baby abdomen was markedly distended and solid masses could be easily palpated through anterior abdominal wall. The baby expired after few hours.

A fter nine months the same patient again visited the antenatal clinic at 30 weeks of gestation. Her routine obstetrical ultrasound revealed a single alive cephalic fetus having BPD of 7.37 cm=gestational age 29.4 weeks and femur length of 5.7cm=gestational age 29.5 weeks. Liquor was markedly scanty with both kidneys markedly enlarged having increased echogenecity. Placenta was fundal posterior and very heavy. The picture was suggestive of polycystic kidney disease. The patient was induced and she delivered a female baby of 2.1kg. Who expired after few hours. The parents refused genetic examination and autopsy of the newborn.

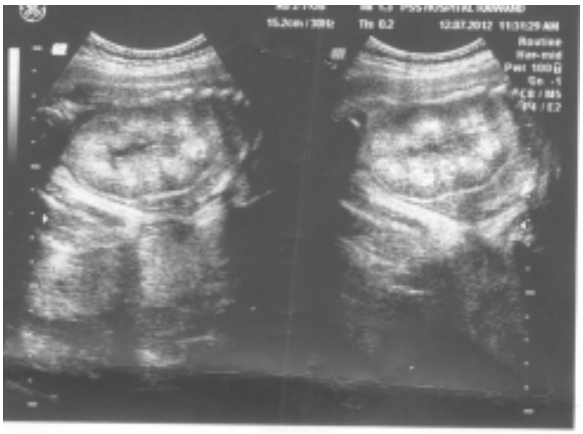

Figure 1: Sonographic view of fetal bilateral polycystic kidneys.

DISCUSSION

The hyperechogenicity of the kidneys can be diagnosed after 17 weeks gestation which results from the presence of multiple micro cysts, dysplasia or tubular dilatation.The differential diagnosis should take into account family history and the presence of associated anomalies. If there are no other malformations in the fetus the main diagnosis is polycystic kidney disease with recessive or dominant genetic disorder.[3] In dominant polycystic kidney disease the fetus shows macro cysts and the amniotic fluid is normal.

Autosomal recessive polycystic kidney disease is the most common heritable cystic renal disease with mutations of a single localized gene in an area in Chromosome 6 (PHKD1).[4] PHKD1 gene is expressed at high levels in fetal and adult kidneys and at lower levels in the liver and this corresponds to the principal sites of the disease. The characteristic pathologic changes occur in the kidneys and the liver with a reciprocal relationship between the degree of renal and hepatic involvement. The hallmark manifestation in the liver is congenital hepatic fibrosis with varying degrees of biliary ectasia and periportal fibrosis. Kidneys show subcapsular cysts, representing ectasia of the collecting tubules. In the cross-section, these dilated tubules can be seen in a radial arrangement extending from the calyx to the capsule. There is epithelial hyperplasia along the collecting ducts and these hyperplastic cells undergo a functional change from resorption to secretion. The combination of epithelial hyperplasia and fluid secretion results in ductal ectasia. Depending on the extent of ductal involvement there is a wide variability of renal dysfunction. Molecular prenatal diagnostic techniques can be used to detect ARPKD in early pregnancy.[5]

Serial ultrasound evaluation starting at 15 weeks can be used as a screening modality. There is a spectrum of findings in ARPKD which are better depicted using high-resolution ultrasound techniques.[6] The characteristic findings in ultrasound are enlarged, homogenously hyperechogenic kidneys with the absence of corticomedullary differentiation and difficulty in identifying fetal bladder.[7] The liver is usually normal in echogenecity. These cases have significant nephromegaly with about 90% of the ducts involved, which may impede delivery. The impairment in renal function leads to oligohydramnios leading to pulmonary hypoplasia, club foot and Potter's facies. Repeat sonographic measurement of the length of the kidneys appears to be a useful parameter to diagnose ARPKD.[8]

CONCLUSION

The presence of isolated large and hyperechogenic fetal kidneys suggests polycystic kidney disease. The antenatal diagnosis should be made as it has serious implications in the continuation of pregnancy. Termination of pregnancy or preterm induction of labor should be considered in order to avoid caesarean section.

REFERENCES

1. Thomas J, Manjunath AP, Rai L, Kudva R. Autosomal recessive polycystic kidney disease diagnosed in fetus: Indian J Urol 2007; 23:328-9.
2. Jezová M, Prásilová S. Autosomal dominant polycystic kidney disease in the fetus with seemingly negative family history. A case report. Cesk Patol 2010; 46:68-71.
3. Dukic L, Schaffelder R, Schaible T, Sütterlin M, Siemer J. Massive increase of foetal abdominal circumference due to hereditary polycystic kidney disease. Z Geburtshilfe Neonatol 2010; 214:119-22.
4. Chaumoitre K, Brun M, Cassart M, Maugey-Laulom, Eurin D, Didier F, et al. Differential diagnosis of fetal hyperechogenic cystic kidneys unrelated to renal tract anomalies: A multicentre study. Ultrasound Obstet Gynecol. 2006; 28:911–7.
5. Zerres K, Mucher G, Becker J, Steinkamm C, Rudnik-Schoneborn S, Heikkila P, et al. Prenatal diagnosis of autosomal recessive polycystic kidney disease (ARPKD): Molecular genetics, clinical experience and fetal morphology. Am J Med Genet. 1998; 76:137–44.
6. Bergmann C, Senderek J, Schneider F, Dornia C, Kupper F, Eggermann T, et al. PKHD1 mutations in families requesting prenatal diagnosis for autosomal recessive polycystic kidney disease (ARPKD). Hum Mutat. 2004; 23:487–95.
7. Traubici J, Daneman A. High-resolution renal sonography in children with autosomal recessive polycystic kidney disease. AJR Am J Roentgenol. 2005; 184:1630–3.
8. Roume J, Ville Y. Prenatal diagnosis of genetic renal diseases: Breaking the code. Ultrasound Obstet Gynecol. 2004; 24:10–8.

CONFLICT OF INTEREST
Authors declare no conflict of interest.
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