# A Fetus with Bilateral Polycystic Kidney Disease

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## ABSTRACT

### Background

Autosomal recessive polycystic kidney disease is a rare genetic disorder that can be detected through prenatal ultrasound.

### Case presentation

We presented a 23-year-old mother, gravida 2 at 23 weeks gestation, has been referred to the tertiary hospital for confirmation of the incidental abnormal ultrasonography findings on her growing fetus. Repeat ultrasonography by the maternal-fetal specialist is strongly suggestive of Autosomal Recessive Polycystic Kidney Disease. Given the poor prognosis, the pregnancy was terminated without any complications.

### Conclusion

Our case highlights the value of primary care physicians in early detection and appropriate referral as they are the first point of contact in the healthcare system. We also stress the importance of genetic counseling, which may have an impact on subsequent pregnancies in the family.



## Keywords

Autosomal recessive; genetic disorder; polycystic kidney; pulmonary hypoplasia


## INTRODUCTION

Autosomal Recessive Polycystic Kidney Disease (ARPKD) is part of a group of congenital hepatorenal fibrocystic syndromes that cause kidney and liver complications in children and are associated with high mortality rates [1]. ARPKD occurs in 1 in 20,000 live births, making it more uncommon than the autosomal dominant type. It often leads to fetal or neonatal death due to pulmonary hypoplasia and severely enlarged kidneys [2]. The affected fetuses may exhibit features of the "Potter" phenotype, such as widely separated eyes with epicanthal folds, broad nasal bridge, low-set ears, receding chin, and contracted limbs with club feet [2]. ARPKD is primarily caused by mutations in the polycystic kidney and hepatic disease 1 (PKHD1) gene. However, mutations in several other cystogenes can produce similar clinical features. Clinically, ARPKD typically presents much earlier, detected at birth or in utero, and is more severe than Autosomal Dominant Polycystic Kidney Disease (ADPKD) [1]. However, clinical presentation is frequently more diverse than generally assumed. This case report emphasizes the crucial role of primary care physicians in early detection and timely referral, as they are the first point of contact within the healthcare system. It also underscores the importance of genetic counseling, which can significantly influence decisions in future pregnancies in the family.


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## Case Presentation

Madam NS, 23 years old Malay woman, G2P0+1 (miscarriage) was referred to the tertiary center at 23 weeks period of gestation for suspected fetal hydronephrosis. She had regular menses but was unsure of her last menstrual date. She had an uneventful abortion for her first pregnancy. This is a consanguineous marriage but there was no family history of renal disease in either family. She did not experience any problems during her early pregnancy. At 23 weeks of gestation, she had a transabdominal ultrasound done which showed a singleton fetus with a large renal cystic lesion and anhydramnios. She was then referred to the tertiary center for further evaluation.

A detailed ultrasound done by an obstetrician showed bilateral lung hypoplasia, markedly enlarged bilateral kidneys with multiple cysts on the left kidney, hyperechoic right kidney with poor corticomedullary differentiation, absence of urinary bladder and anhydramnios (Figure 1). The findings were confirmed two weeks later by the maternal-fetal medicine specialist, revealing high suspicion of Autosomal Recessive Polycystic Kidney Disease.

The couple were informed regarding their fetal condition and counselled on the outcome of her pregnancy, which is usually poor. Expectant management or termination of pregnancy were also discussed. Initially they opted for conservative management with biweekly review. However, at 30 weeks of gestation, they agreed to proceed with the termination of pregnancy. The treating team decided on the vaginal delivery after assessing the fetal condition and the delivery process was successful. She delivered a physically normal-formed fresh stillbirth baby with a birth weight of 1.58kg. The couple was advised for postmortem however they declined.

## DISCUSSION

The incidence of ARPKD may be notably higher in consanguineous marriages, particularly within isolated or inbred populations [1]. Mutations in the PKHD1 gene are responsible for all classical forms of ARPKD. Although there is no documented familial hereditary renal disease in this couple's family, it is important to remember that de novo mutations can also cause ARPKD.

The prenatal manifestations of ARPKD are variable and some are related to the age at presentation. Approximately

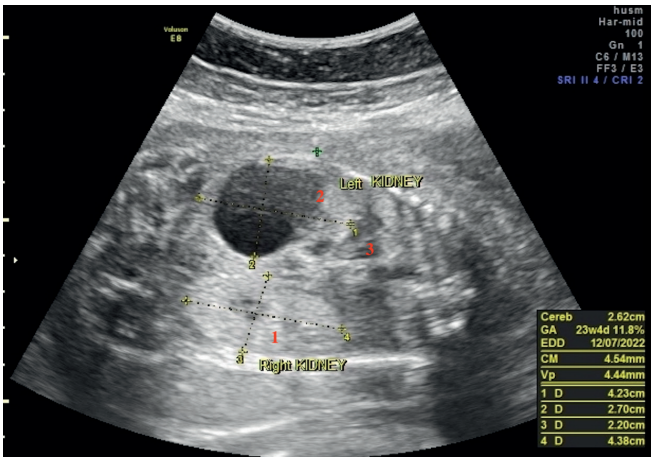


**Figure 1:** Image showing markedly enlarged bilateral kidneys, hyperechoic right kidney with poor corticomedullary differentiation (1), cyst on the left kidney (2), bilateral lung hypoplasia (3), absence of urinary bladder, and anhydramnios.

40% of cases represent the most severe form, typically identified in utero during the first or second trimester. These early-detected cases often have a poorer prognosis compared to those diagnosed after one month of age, which are associated with longer renal survival [3,4]. In fetuses with ARPKD, the kidneys show reduced or absent corticomedullary differentiation (CMD), while reverse CMD – characterized by a hyperechogenic medulla and hypoechogenic cortex – is less common. Renal cysts are present in approximately 60.6% of cases. Oligohydramnios or anhydramnios which is a common clinical presentation, occurs in about 84.6% of the affected fetuses. Although amniotic fluid levels can reduce or remain unchanged as pregnancy progresses, they are not definitive diagnostic indicators of ARPKD [5].

Pulmonary complications such as pulmonary hypoplasia can be lethal in some cases. In infants with ARPKD, respiratory failure after birth is often linked to renal oligohydramnios, which reduces amniotic fluid filling in the lung and leads to pulmonary hypoplasia. However, the severity of respiratory outcomes may depend on the timing of oligohydramnios onset during gestation. Oligohydramnios occurring after 28 weeks of gestation may be associated with a better prognosis. This should be carefully considered before predicting a lethal postnatal outcome [6]. In contrast, severe oligohydramnios is generally considered as a poor prognostic sign due to high risk of accompanying pulmonary hypoplasia [7]. Another contributing factor to respiratory compromise

is significant kidney enlargement, which can impair the diaphragm's mobility and lead to breathing difficulties [3]. Madam NS had an uneventful early pregnancy journey until a transabdominal ultrasound performed at 23 weeks of gestation revealed features suggestive of ARPKD. Progress and advances in technology have decisively contributed to the development of prenatal diagnostics of ARPKD based on the time of presentation and ultrasound features [5].

When the diagnosis of ARPKD was confirmed by the maternal-foetal medicine specialist at 26 weeks of gestation, the couple opted for expectant management. It is important to involve the patient in decision-making process, as she has the right to decide, and her decision must be respected. News of this pregnancy was a source of joy to the couple as first-time parents but knowing that their baby is not physically normal can be distressing and upsetting. It is normal for the couple to feel shocked and emotional distress. Proper counselling sessions provided to parents have been shown to enhance their acceptance and motivation, enabling them to address challenges more effectively [8]. The couple must also receive emotional support and counselling during this challenging time. They felt bound between religious beliefs and medical perspectives in the decision-making process. Some religions forbid abortion if the pregnancy has reached or passed the 120 days, unless in situations that endanger the mother's life or in the presence of foetal abnormalities which is incompatible with life [9]. Although the foetal diagnosis was established in the second trimester, the termination of pregnancy was commenced in the third trimester. Despite the potential risks of termination of pregnancy, which include bleeding, uterine rupture, and uterine hypotonia, the risks of continuation of pregnancy, such as obstructed labour, were also discussed with the couple. Evidence has shown that there are no significant differences in maternal morbidity between the second and third trimesters' termination of pregnancy [10]. The mode of delivery is determined by the fetal abdominal circumference and the gestational age. In cases where there is fast growing of the kidneys, there is a risk of abdominal dystocia, either through vaginal delivery or even caesarean section [11]. Thus, the correct timing of delivery should be discussed and decided wisely.

Primary care plays a crucial role in providing support and coordinating care for expectant parents and their future pregnancies. Since ARPKD is genetically inherited, giving them genetic counselling is the best option before they plan for future pregnancies. If both parents carry the ARPKD gene, prenatal testing such as chorionic villous sampling can be offered to detect ARPKD in the foetus during early pregnancy. Early prenatal diagnostic exclusion by ultrasound during the next pregnancy can also be done; however, it cannot be guaranteed in view of the possibility of intrafamilial variability of severity in ARPKD, although the onset in the current pregnancy is quite early. Coping with the genetic risk and making decisions about future pregnancies can be emotionally challenging for Madam NS and her partner. However, by increasing their awareness of the high recurrence risk, they will be more prepared and accepting of the next pregnancy.

## CONCLUSION

ARPKD is an inherited disease that is associated with a high morbidity and mortality rate. The clinical spectrum of this disease varies and depends on the age at presentation, and it can be diagnosed prenatally or during the neonatal period. The availability of good antenatal care with early ultrasound detection may improve case detection with early and timely referral. The prognosis of this condition is unpredictable due to its range of complications, and the prognosis was poorer in cases diagnosed at less than 28 weeks of gestation.

### Authors undertaking

Data gathering: Ermadina Mohd Kalam, Engku Husna Engku Ismail

Writing manuscript: Ermadina Mohamad Kalam, Azlina Ishak

Editing and approval of final draft: Ermadina Mohamad Kalam, Azlina Ishak, Siti Suhaila Mohd Yusoff, Engku Husna Engku Ismail

Approval of final draft: Azlina Ishak

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### Ethical approval about publication

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### Conflicts of interest

No conflict of interest is declared by the authors.

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