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SCIENTIFIC ARTICLE - CASE REPORT

DIAGNOSTIC AND PROGNOSTIC IMPLICATIONS OF AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE IN PRENATAL CARE: CASE STUDY AND CLINICAL CONSIDERATIONS

VINICIUS CAPELLARI GABANA[1], LUÍSA RIGON BORBA[1], YASMIN RICARTE HASS LOPES[1], ANDRÉ CAMPOS DA CUNHA[1], RAFAEL ROSA[1], JORGE ALBERTO BIANCHI TELLES[2]

1. Universidade Federal de Ciências da Saúde de Porto Alegre (UFCSPA), Porto Alegre, RS
2. Hospital Materno Infantil Presidente Vargas (HMIPV), Porto Alegre, RS

ABSTRACT

INTRODUCTION: Autosomal recessive polycystic kidney disease (ARPKD) is the most frequently observed cystic kidney disease in the prenatal period. Our objective was to describe the prenatal findings of a fetus diagnosed with ARPKD, highlighting their importance for the diagnosis, management, and prognosis of patients.

CASE REPORT: The patient was a 16-year-old primigravida whose husband was consanguineous. She was referred for evaluation due to multicystic dysplastic kidneys in the fetus. The examination at 22 weeks revealed enlarged, hyperechoic dysplastic kidneys with cysts, associated with oligohydramnios. The ultrasound at 31 weeks showed reduced thoracic circumference and apparent pulmonary hypoplasia. Fetal magnetic resonance imaging (MRI) revealed similar findings, consistent with ARPKD. The child was born with Potter's facies and a severely distended abdomen, and passed away after birth due to respiratory dysfunction.

DISCUSSION: Prenatal identification through ultrasound of the renal characteristics associated with oligohydramnios was crucial for the diagnosis of ARPKD, especially in light of various differential diagnoses. Data from the clinical history and the results of the MRI evaluation were also important for confirming the diagnosis. Additionally, other findings, such as reduced thoracic circumference, assisted in the planning of the birth and determining the severity of the prognosis.

CONCLUSION: Our report highlights the importance of prenatal evaluation through ultrasound for the detection of findings that play a crucial role in both the diagnosis of ARPKD and its management and prognosis.

KEYWORDS: AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE; PRENATAL DIAGNOSIS; OLIGOHYDRAMNIOS; CLINICAL MANAGEMENT; PROGNOSIS.

INTRODUCTION

Polycystic kidney disease is a genetic condition that can follow either an autosomal dominant or autosomal recessive inheritance pattern. However, both forms are characterized by the presence of multiple renal cysts and are classified as ciliopathies due to the abnormal structure and function of cilia (organelles present on the apical surface of almost all epithelial cells and many endothelial cells), which contribute to cystic cell proliferation, fluid secretion, and changes in the extracellular matrix [1,2].

These conditions can be detected in the prenatal period through the identification of characteristic ultrasound findings. Additionally, these findings allow for the differentiation between adult disease, which follows a dominant pattern, and childhood disease, which has an autosomal recessive pattern. The latter is more commonly known as autosomal recessive polycystic kidney disease (ARPKD) and is the most frequently observed cystic kidney disease in the intrauterine period [3].

Thus, the aim of this report was to describe the prenatal findings of a fetus diagnosed with ARPKD, emphasizing their importance for determining not only the diagnosis but also the management and prognosis of patients.

CASE REPORT

The patient was a 16-year-old woman in her first pregnancy. She was referred to the fetal medicine service after an ultrasound showed a fetus with evidence of multicystic dysplastic kidneys. The husband was consanguineous, a first-degree cousin. In the second-trimester ultrasound at 22 weeks of gestation, dysplastic kidneys were observed, enlarged in

size (the right kidney measured 6.1 x 3.2 x 3.2 cm, with a volume of 33.1 cm³, and the left kidney measured 5.7 x 3.5 x 3.3 cm, with a volume of 34.2 cm³), hyperechoic with multiple cysts inside, showing poor corticomedullary differentiation and associated with the presence of oligohydramnios (Fig. 1).

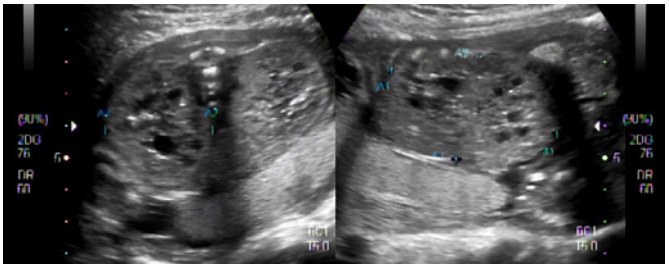

Figure 1. Two-dimensional ultrasound images taken at 22 weeks of gestation, showing the appearance of the kidneys, which are dysplastic and enlarged, as well as hyperechoic, with multiple cysts inside and poor corticomedullary differentiation.

Fetal echocardiography was normal. The fetal karyotype, obtained through cordocentesis, revealed a normal male chromosomal constitution (46,XY). The ultrasound performed at 31 weeks of gestation also showed reduced thoracic circumference associated with apparent pulmonary hypoplasia.

Fetal magnetic resonance imaging (MRI) revealed findings similar to those observed in the ultrasound evaluation, which were consistent with the diagnosis of ARPKD (Fig. 2).

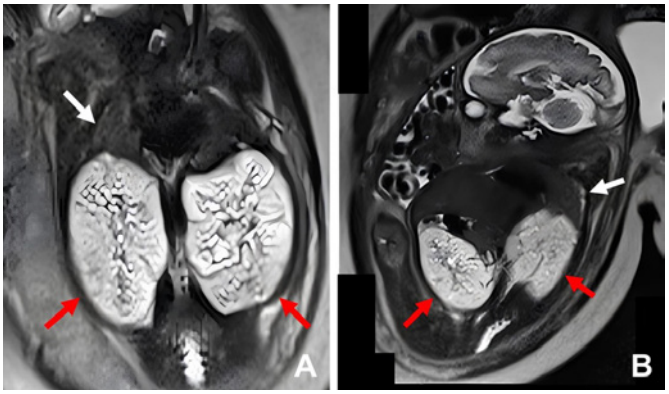

Figure 2. Appearance of the kidneys as seen through fetal magnetic resonance imaging. Note the similarity to the findings observed in the ultrasound examination. The red arrows indicate the kidneys, and the white arrows indicate the lungs, which appear hypoplastic based on their size (A and B).

The child was born via cesarean section, at term, weighing 3,130 g, with an Apgar score of 2 at both one and five minutes. Upon evaluation of the newborn, Potter's facies were observed (flattened face with prominent infraorbital grooves, micrognathia, and low-set ears), as well as a severely distended abdomen. The infant developed severe respiratory dysfunction and passed away a few hours after birth.

DISCUSSION

The assessment of fetal anatomy in the second trimester of pregnancy through ultrasound is an essential tool for diagnosing various congenital defects, with approximately 20% of these related to renal anomalies. One of the most suggestive findings of ARPKD is the presence of renal cysts, usually small (1-2 mm in diameter), detected between 21 and 24 weeks of gestation. The quantity and size of the cysts are important factors for diagnosis. Although hepatic involvement is characteristic of ARPKD, it is generally not detected before birth. Other relevant ultrasound findings include enlarged kidneys and reduced amniotic fluid volume, indicating renal dysfunction. The dilation of renal tubules also causes renal hyperechogenicity, and the fetal bladder may be small or not visualized due to low urine production [3-5].

In this case, fetal MRI complemented the ultrasound findings, confirming the diagnosis of ARPKD. MRI is particularly useful in situations of oligohydramnios, anhydramnios, maternal obesity, or poor fetal positioning, which can hinder the visualization of the kidneys and urinary tract [6].

The differential diagnosis of ARPKD includes various conditions involving renal cysts, such as Autosomal Dominant Polycystic Kidney Disease (ADPKD) and multicystic dysplastic kidneys. ARPKD may resemble Meckel-Gruber syndrome, but the latter presents additional characteristics, such as occipital encephalocele and postaxial polydactyly[4-6].

Fetal karyotyping was performed not to confirm ARPKD, as this is a genetic condition, but to exclude chromosomal abnormalities, such as those seen in trisomies of chromosomes 13 and 18, which can also be associated with cystic kidneys. Cordocentesis was chosen due to the presence of anhydramnios. Fetal echocardiography was conducted to rule out congenital heart defects, which are common in chromosomal syndromes [6].

ARPKD is a severe cystic kidney disease that can also affect the liver and biliary tract, with an estimated incidence of 1 in 20,000 births. It is caused by mutations in the PKHD1 gene, located on chromosome 6p12.3-p12.2. The mutations are generally unique to each family, complicating genotype-phenotype correlations and the implementation of direct diagnostic testing [3,7].

As an autosomal recessive condition, ARPKD can be associated with parental consanguinity and a history of affected siblings, although the absence of these factors does not exclude the diagnosis. Accurate diagnosis is essential for genetic counseling and determining the risk of recurrence, which is 25% for future children of the couple[4,8].

Furthermore, genetic counseling should address the prognosis and management of the pregnancy. In cases of Potter's sequence, such as in our patient, the prognosis is severe, with a high rate of extrauterine mortality. In less severe situations,

family counseling through birth planning and preparation for neonatal interventions, such as dialysis and mechanical ventilation, is essential [6,9].

In cases of ARPKD, the disease itself is not always present in the prenatal period; however, its early expression during this time is considered a factor for poor prognosis. The presence of oligohydramnios or anhydramnios is the most impactful finding in determining survival [4]. Additionally, reduced thoracic circumference is associated with pulmonary hypoplasia, which is common in these cases, usually secondary to the reduction or absence of amniotic fluid (oligohydramnios or anhydramnios). This is due to the absence of fetal urine production resulting from renal impairment, leading to fetal constriction and Potter's sequence, with findings such as facial flattening, prominent infraorbital grooves, and low-set ears (Potter's facies), as well as deformities with contractures or arthrogryposis of the limbs and pulmonary hypoplasia [1,2].

Pulmonary hypoplasia, secondary to oligohydramnios, is the leading cause of postnatal death, accounting for approximately 30% of deaths shortly after birth. However, neonatal survival has improved in cases with less severe renal impairment and absence of oligohydramnios [1,4,7].

Thus, the prenatal identification of these findings in suspected cases of ARPKD can be used as a tool for predicting perinatal risk and long-term prognosis [4].

CONCLUSION

This report highlights the importance of ultrasound in the prenatal diagnosis of ARPKD, allowing for the identification of renal and gestational findings that suggest the condition, even among various cystic kidney diseases. Clinical data and supplementary tests, such as MRI, also contribute to confirming the diagnosis. Additional examinations, such as karyotyping and fetal echocardiography, are useful in the differential diagnosis. Findings like oligohydramnios and reduced thoracic circumference are relevant for birth planning and prognosis determination.

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VINICIUS CAPELLARI GABANA
http://lattes.cnpq.br/0689989993576383
https://orcid.org/0009-0005-0310-7617

LUÍSA RIGON BORBA
http://lattes.cnpq.br/1748098286012527
https://orcid.org/0009-0003-6444-2318

YASMIN RICARTE HASS LOPES
http://lattes.cnpq.br/5658666448394680
https://orcid.org/0009-0008-8082-4996

ANDRÉ CAMPOS DA CUNHA
http://lattes.cnpq.br/8565636243001078
https://orcid.org/0000-0002-9893-3348

JORGE ALBERTO BIANCHI TELLES
http://lattes.cnpq.br/8253976877321806
https://orcid.org/0000-0002-8108-9638

RAFAEL FABIANO MACHADO ROSA
http://lattes.cnpq.br/7255679916272398
https://orcid.org/0000-0003-1317-642X

MAILING ADDRESS:
JORGE ALBERTO BIANCHI TELLES
Avenida Independência, 661 Bairro Independência
Porto Alegre, RS, Brasil. CEP: 90035-074
E-mail: telles383@gmail.com

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