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Bilateral Renal Agenesis: About A Rare Case

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ABSTRACT

Renal agenesis is the absence of renal tissue due to defects in early embryonic development. It is often associated with other congenital abnormalities. The incidence of bilateral renal agenesis ranges from 0.1 to 0.3 per 1,000 births, making it a rare condition. Its diagnosis is mainly based on obstetric ultrasound by direct and indirect signs. Magnetic Resonance Imaging (MRI) has a higher sensitivity and specificity, and has its place when in doubt or when other anomalies are suspected. Management of bilateral renal agenesis relies primarily on prevention of pulmonary hypoplasia during pregnancy by amnioinfusion and correction of postpartum complications related to renal absence. Although the prognosis for bilateral renal agenesis is extremely poor and the risk of death in the neonatal period is high. We report a pregnancy in which prenatal ultrasound revealed anhydramnios and the absence of both fetal kidneys, To overcome the difficulties in obtaining accurate information on fetal ultrasound in cases of oligohydramnios, we used fetal magnetic resonance imaging (MRI) to confirm renal status, which confirmed fetal renal agenesis and provided a more accurate diagnosis. The neonate was born with a complex dysmorphic syndrome and died 45 minutes after birth due to respiratory distress.

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INTRODUCTION

Bilateral renal agenesis is the complete absence of development of both kidneys in the fetus, and may be isolated or sometimes associated with complex malformative syndromes. It has a poor prognosis that ends in death due to pulmonary hypoplasia in the majority of cases (1). Fetal ultrasound is the traditional means of evaluating this condition, and the ultrasound diagnosis is based on the inability to visualize the fetal kidneys, the bladder, and the presence of severe oligohydramnios (2). However, in the absence of amniotic fluid, ultrasound evaluation of the fetus is complex and results are uncertain (3). In the past few years, fetal magnetic resonance imaging (MRI) has been increasingly used as an adjunct to ultrasound for more accurate prenatal diagnosis.

Through this case report, we highlight the importance of ultrasound and MRI in the prenatal diagnosis of bilateral renal agenesis and its orienting signs, and understand the pejorative prognosis in recent studies.

CASE PRESENTATION

Our patient was a 30 years old woman, gravida 4 para 3 live birth 3. Her husband was 37 years old. The couple was healthy and non-consanguineous and their family history was non-contributory. Their first pregnancies were normal, resulting in 2 girls aged 13 and 9 years old and a boy of 4 years old. The deliveries were by cesarean section, and the growth and psychomotor development of the 3 children were unremarkable. The current pregnancy was an unmonitored pregnancy; a fetal ultrasound was performed at 27 weeks of gestation for the decrease of active fetal movements, which revealed an evolving monofoetal pregnancy and positive cardiac activity, the vacuity of the renal chambers with an empty bladder associated with pulmonary hypoplasia and weight for gestational age less than 1 percentile, the Doppler ultrasound showed the absence of renal arteries, a harmonious placenta of normal thickness was well inserted. The fetus was diagnosed with bilateral renal agenesis. These findings of anhydramnios and invisibility of the kidneys, bladder, and renal vessels were confirmed on repeat ultrasound imaging 10 days later (Figure 1). The rest of the anatomy appeared to be normal, but the visibility was poor due to anhydramnios, hence the importance of a more reliable examination, an MRI (ultrafast MR imaging) (Figure 2) was performed at 31 weeks of gestation and again the kidneys, bladder, and renal vessels were not seen in the fetus. No other abnormalities were detected.

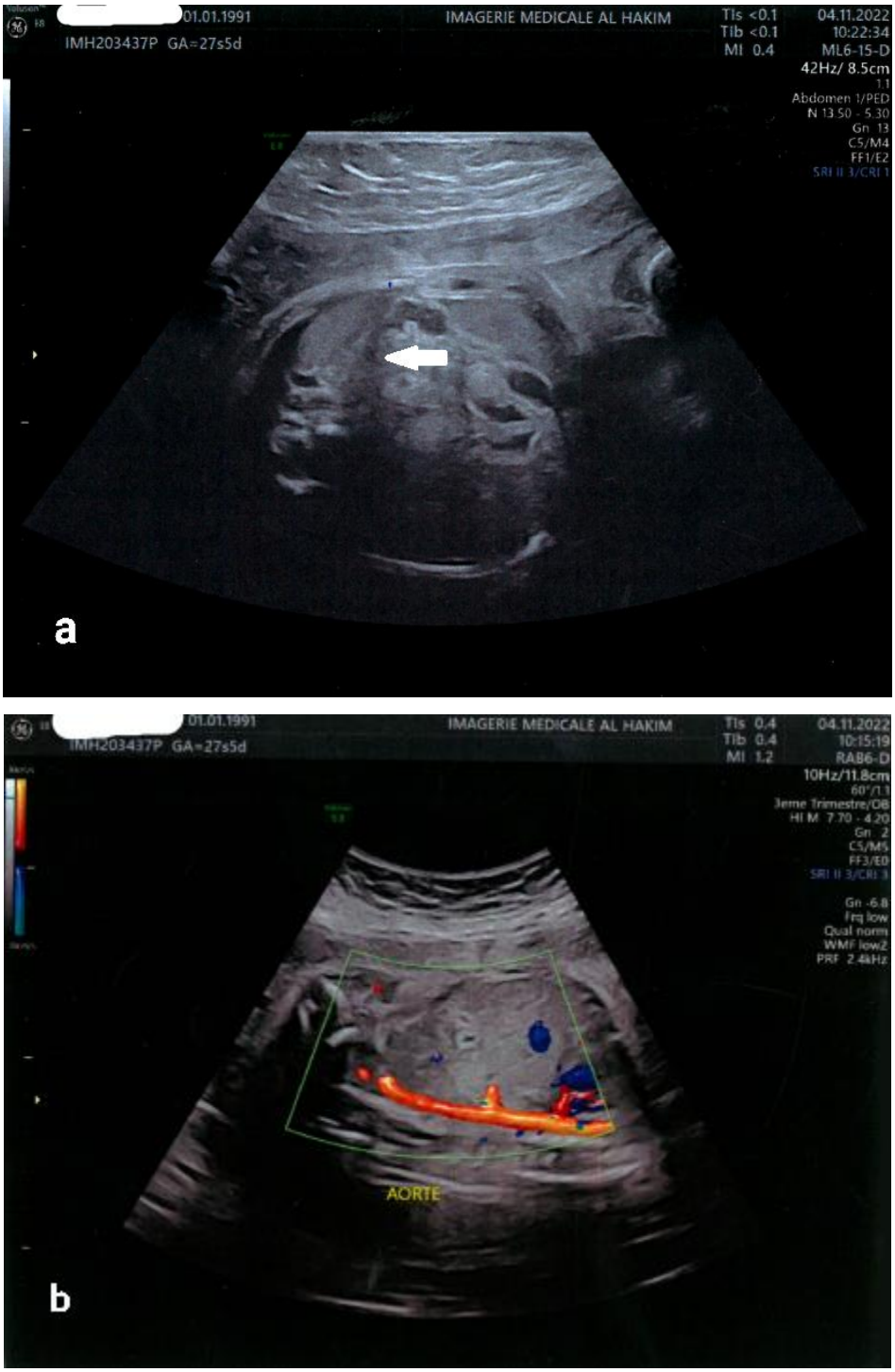


Figure 1: Ultrasound features of bilateral renal agenesis: (a) fetal ultrasound in transverse section through both renal lodges showing absence of both kidneys (b) Doppler ultrasound showing absence of both renal arteries.

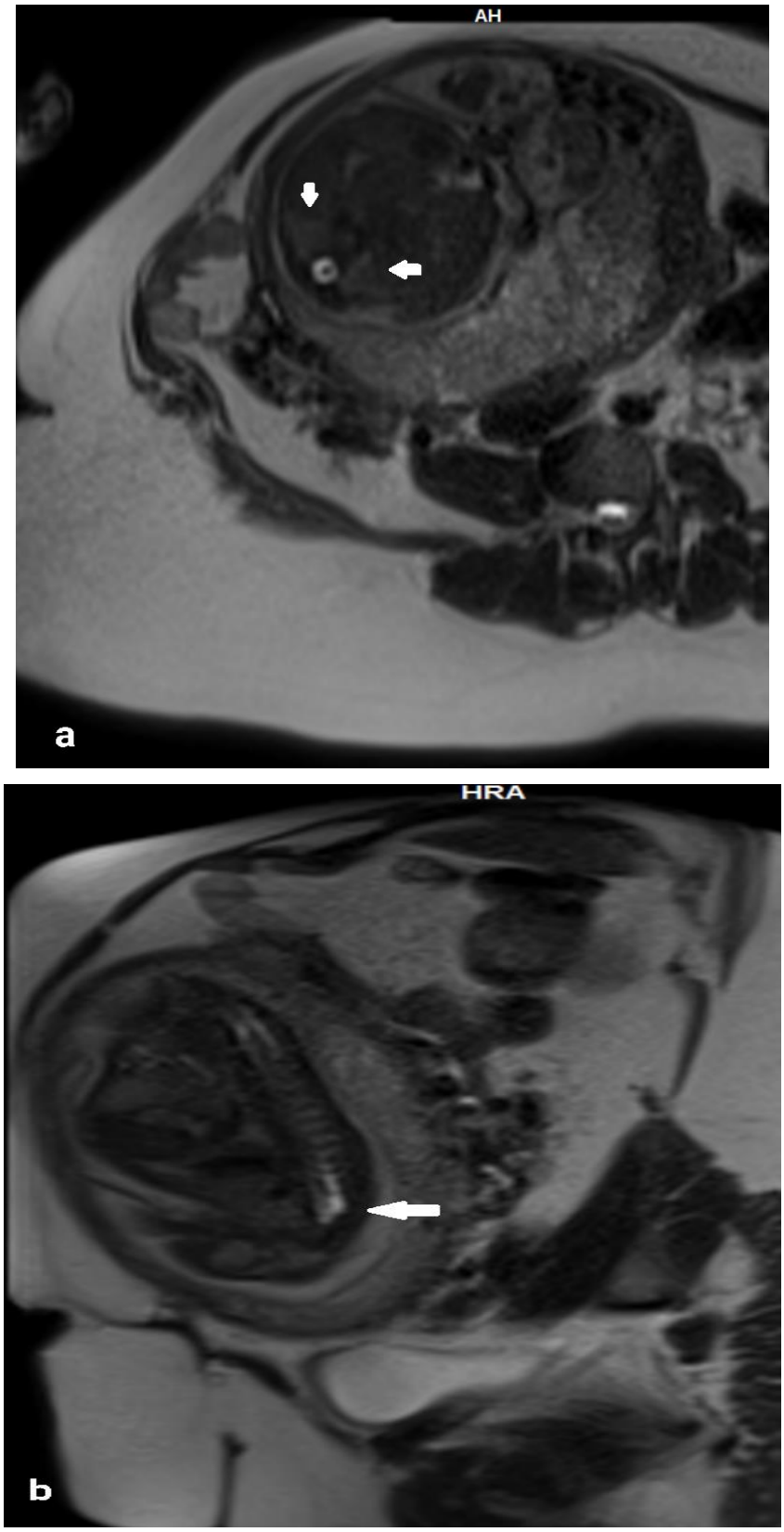
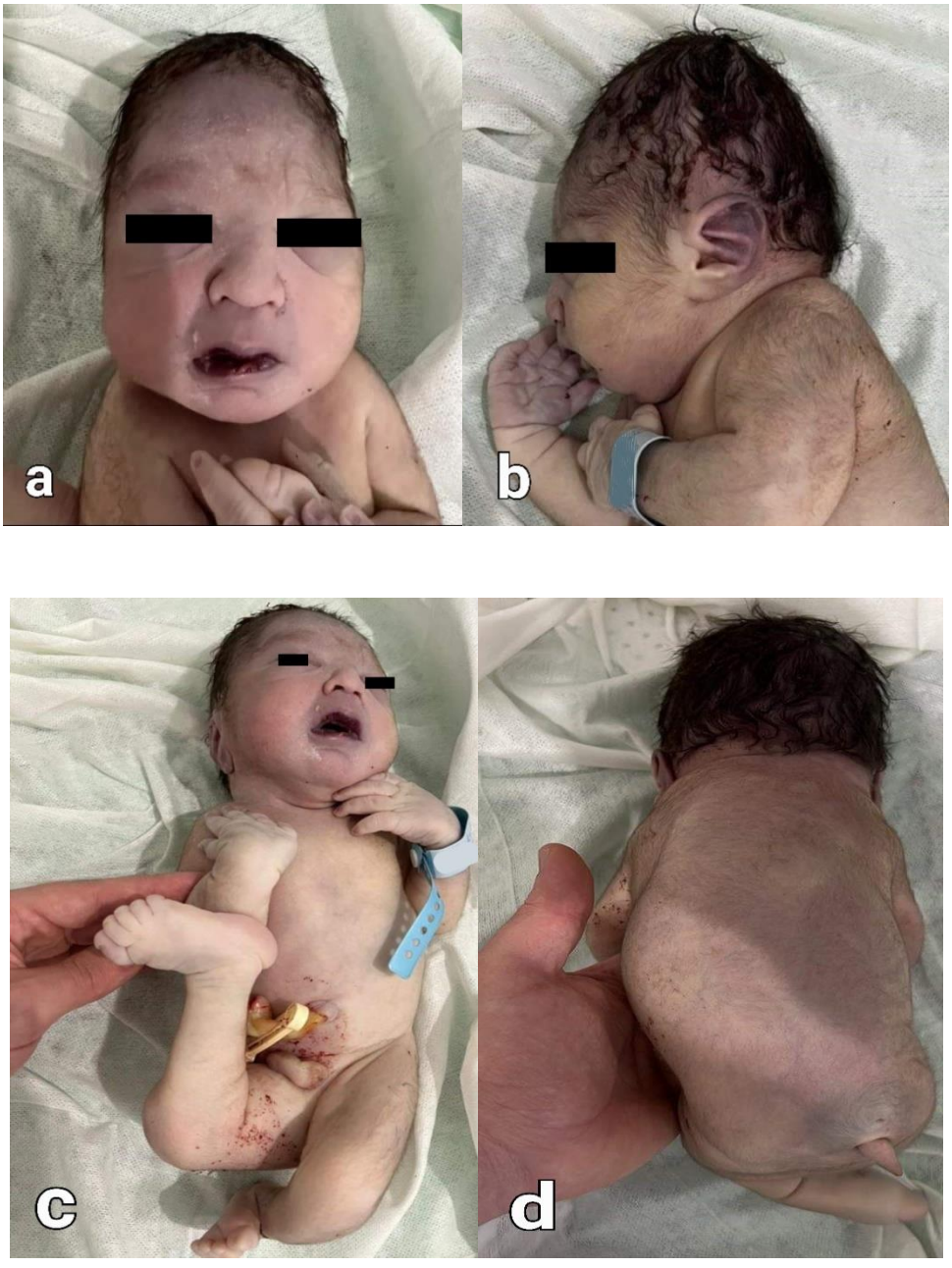


Figure 2: Fetal MRI on T2-weighted images (a) transverse section through the 2 renal loges showing an absence of both kidneys with the " lying-down" adrenal sign (b) sagittal section through the fetal spine showing a low attached spinal cord.

Despite of these findings were incompatible with life, the couple decided to continue the pregnancy. Four weeks later the patient was admitted to the maternity department with an emergency premature delivery, which was performed by cesarean section for triply scarred

uterus. The newborn was delivered after the onset of spontaneous labor at 35 weeks of gestation. He weighed 1930 grams, and had a head circumference of 31, 5 cm, and a length of 43.0 cm, with Apgar scores of 2 at 5 min of life, His oxygen saturation level in room air was 70%. There was no spontaneous breathing and his heart rate was 50-60 bpm. Visually he had a dysmorphic syndrome with hypertelorism, epicanthus, nasal enlargement and flattening, retrognathism, long low set ears, clubfoot suggestive of Potter's syndrome. He also had a caudal appendage and anal imperforation (figure 3). The newborn was reanimated for 25 minutes, and was declared dead 45 minutes after delivery.



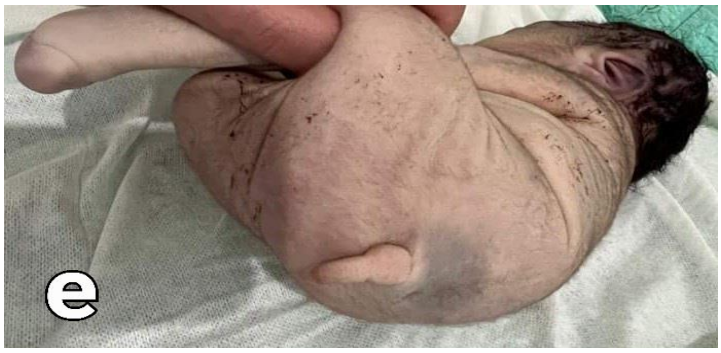


Figure 3: images of the newborn diagnosed with bilateral renal agenesis prenatally: a, b: typical Potter's syndrome face with hypertelorism, epicanthus and long low set ears. c: male newborn with a malformation of the lower limbs such as clubfoot. d, e: presence of a caudal appendage and anal imperforation.

DISCUSSION

Ultrasound (US) has been the main imaging modality for the prenatal diagnosis of fetal abnormalities for decades. In the early 1980s, fetal MRI was introduced with MRI sequences lasting several minutes. Longer imaging times require fetal paralysis using percutaneous umbilical cordocentesis or immobilization of the fetus by maternal sedation, which has its attendant risks. The introduction of ultra-fast T2-weighted sequences enables the acquisition of MRI sequences in approximately 20 seconds without fetal immobilization(4), which has facilitated the diagnosis of fetal malformations and particularly organ malformations, including the kidneys. As it is known its development is a very complex process that includes three stages: pronephros, mesonephros and metanephros. Disorders in these early stages of development can result in renal and/or urinary tract malformations. The overall incidence of these anomalies ranges from 0.1 to 1 per 100 births, whereas the specific incidence of bilateral renal agenesis ranges from 0.1 to 0.3 per 1,000 births, making it rare (5).

Prenatal Ultrasonography

Prenatal ultrasound is the primary imaging modality for the discovery of fetal malformations. It is the first-line examination for pregnancy monitoring and diagnosis of fetal and placental anomalies and urogenital evaluation (6). The fetal kidneys appear lobular and can be seen on a prenatal ultrasound as early as 9 to 12 gestational weeks in the paraspinal region. In addition, corticomedullary differentiation occurs between 15 and 20 weeks. Glomerular filtration begins at approximately 9 weeks but does not significantly affect the amniotic fluid until 19-20 weeks when the skin develops. The bladder can be seen on ultrasound from 10 to 14 weeks, and voiding can be seen 15 weeks later. Urine becomes an important contributor to amniotic fluid and increases from 20 weeks to birth (7).Ultrasound indicators have variable specificity and sensitivity. The absence of fetal kidneys in the renal fossa, an empty bladder and the presence of anhydramnios after 16 weeks of gestation are strong indicators of

bilateral renal agenesis (2). Other sonographic findings of renal agenesis include the " lying-down" adrenal sign, in which the adrenals appear large and flattened rather than maintaining their normal "Y" configuration. The echogenic adrenal medulla between the cortex creates a layered appearance in the adrenal glands; they lack corticomedullary pyramidal differentiation and should not be confused with renal tissue (8). Insufficient amniotic fluid blurs ultrasound; therefore, diagnostic amnioinfusion describes a technique to confirm the diagnosis and clearly visualizing the lesions (1). In addition to ultrasound criteria mentioned above, we have found in the literature that the last criterion for diagnosis is the anterior deviation of the aorta at the level of the upper part of the adrenal glands. This can be explained by the fact that the adrenal glands come together at the top of the gland and push the fetal aorta forward (6). Color Doppler ultrasound may also be used if the ultrasound diagnosis is equivocal. The normal renal artery should be seen in the posterior coronal view as a direct branch of the abdominal aorta below the origin of the superior mesenteric artery. The absence of renal arteries strongly suggests bilateral renal agenesis (9).

Our case displays the same ultrasound signs of bilateral renal agenesis as described in the literature, including a bladder void, empty renal compartments, and the absence of renal arteries on Doppler ultrasound.

Most urinary tract abnormalities can be detected by obstetric ultrasonography. These abnormalities range from mild to severe and can be potentially fatal. However, oligohydramnios and maternal obesity may limit the diagnostic accuracy of ultrasound, which may make additional imaging studies useful (3).

Prenatal MRI

Fetal MRI is a safe and effective technique in cases of ambiguous signs. Congenital kidney anomalies are often associated with oligohydramnios, which can make ultrasound evaluation difficult, whereas fetal MRI is not significantly affected by reduced amniotic fluid. MRI is currently a validated technique for fetal imaging, although it also has its limitations, such as morbid obesity, motion artifacts, cost, availability and claustrophobia. Currently, no adverse effects have been demonstrated. Magnetic Resonance Imaging also has good reliability and validity to quantify fetal pulmonary hypoplasia with high accuracy in assessing lung volumes (10). Only a few studies have been published on the role of MRI in detecting renal agenesis and its consequences. In our case, the MRI helped in visualizing fetal anomalies well, especially the pulmonary hypoplasia and its severity, and exploring the renal agenesia found by the ultrasound exploration

To evaluate the role of MRI in the diagnosis of bilateral renal agenesis, the study of Pico et al (10) carried out on 108 fetuses with echographic suspicion of renal and urinary malformations, they benefited from an MRI to confirm or exclude or complete the diagnosis,

The results showed concordant results between MRI and obstetrical ultrasound in 67% of the cases, including 13 fetuses with renal agenesis. On the other hand, MRI provided contradictory or complementary data in 33% of the cases, including 7 fetuses with renal agenesis. To study the sensitivity and specificity of MRI in the diagnosis of renal anomalies, we chose the Ibrahim et al. study (11), which was conducted on 52 fetuses suspected of having renal malformations including bilateral renal agenesis, the MRI results were compared with the post-natal assessment, which provided the same diagnosis in 46 cases (88%), while it gave a different diagnosis (false positive) in 6 cases (11%), MRI was 100% sensitive, 99.9% specific with a positive predictive value (PPV) of 89.5% and accuracy in the diagnosis of suspected renal anomalies. Similar to the study by Bazeed et al. (12), MRI and US were concordant in diagnosing bilateral renal agenesis in 73.5% of the cases. MRI modified the diagnosis in 17.6% of cases from bilateral renal agenisis to unilateral renal agenisis or the opposit. MRI also changed the diagnosis in 8.8% of cases from inconclusive to bilateral renal agenesis. The evaluation of the urinary bladder was nearly equal in both techniques, with the exception of one fetus, in which the urinary bladder was seen only by the US, mainly because the bladder was empty at the time of the MRI. According to the study of Poutamo et al. (13), the urinary tract anomalies were correctly diagnosed by ultrasound in 15 of 22 fetuses and by MRI in 20 of 22 fetuses. The study also found that diagnostic concordance of MRI was better than that of the US (97% of concordance with MRI versus 56% with ultrasound). Furthermore, Cassart et al. (3) showed that MRI provides more accurate information than ultrasound and can influence obstetric decision-making. They recommended medical abortion for three fetuses and helped reassure parents and obstetricians by correcting the misdiagnosis, allowing one case to continue with the pregnancy. From this, it can be concluded that MRI is recommended before any medical intervention if the renal malformation is suspected. Additionally MRI should be recommended when an ultrasound examination suggests bilateral renal agenesis but does not provide a definite diagnosis.

It should also be noted that bilateral renal agenesis may be associated with other malformations in a complex polymalformative syndrome. While ultrasound can often provide a basic diagnosis, some abnormalities cannot be seen visually. The position of the fetus and the appearance of the mother's body can prevent detection and visualization of abnormalities, and a thorough and accurate assessment of normal and abnormal anatomy can be time-consuming and require special skills. In contrast, fetal MRI can quickly and accurately visualize the entire fetus and uterine contents, and can detect a variety of abnormalities, allowing for a more specific and accurate diagnosis of any malformation (4). Fetal MRI will likely will soon be routinely performed for some fetal anomalies due to its sensitivity and specificity, and therefore obstetricians should be familiar with this imaging method (5).

In our case, the fetal MRI did not identify the accompanying abnormalities that are present in the newborn at birth, such as facial dysmorphism, anal imperforation, and caudal appendage.

Prognosis and outcome

Management of bilateral renal agenesis during pregnancy relies primarily on prevention of pulmonary hypoplasia which is a serious and very lethal condition for the fetus secondary to Anhydramnios, the amniotic fluid is a primordial factor for lung growth and the prevention of alveolar collapse. Amnioinfusion is a prenatal procedure in which fluid is introduced into the amniotic cavity either transabdominally or transvaginally. It is an attempt to restore the physiological conditions of the intrauterine environment for the fetus. Amniotic fluid perfusion can reduce uterine pressure caused by oligohydramnios, maintain alveolar expansion, and promote fetal lung growth. Regular perfusion should be used to improve the prognosis of fetal live birth (1).

To understand the prognosis of bilateral renal hypoplasia, Riddle et al. (14) evaluated 47 patients with bilateral renal agenesis, 8 of whom underwent amnioinfusion to prevent complications of oligohydramnios. All these cases were born alive, the family chose comfort care in one case, for the other 7 cases, their families chose to pursue aggressive neonatal interventions, 4 of them died of cardiopulmonary arrest within the first 48 hours, 3 survived 7 days or more and 2 survived 30 days or more and died of sepsis after the start of peritoneal dialysis, no patient could survive for long. To explain the poor prognosis even after amnioinfusion and neonatal dialysis, and its association with cardiac failure, hypotension and non-withdrawal of vasoactive drugs in the newborn, Riddle et al (15) have discovered new physiology in bilateral renal agenesis based on the rennin-angiotensin-aldosterone system (RAS), As is known, renin is an enzyme formed by the kidneys that stimulate the adrenal glands to secrete aldosterone, which allows the maintenance of correct blood pressure and regular cardiovascular activity. Renin deficiency in bilateral renal agenesis can be responsible for relative adrenal insufficiency and consequently for vascular collapse and cardiogenic shock. This finding may improve the neonatal prognosis for these patients by correcting this associated adrenal insufficiency.

CONCLUSION

Bilateral renal agenesis can be diagnosed before birth. Diagnostic criteria include absence of fetal kidneys and their arteries, oligohydramnios, and unvisibility of fetal bladder. Diagnosis should be made early to facilitate patient counseling and safe termination of pregnancy or participation in serial amnioinfusion clinical trials. While prenatal ultrasonography and Doppler ultrasonography are the primary tools for diagnosing fetal abnormalities, MRI may be useful when visualization is suboptimal. Although the prognosis of bilateral renal agenesis

is extremely poor, it is mainly attributed to pulmonary hypoplasia in the neonatal period. Therefore, serial amnioinfusion can be used as an intervention to promote fetal lung growth. However, there are no rigorous prospective studies for this therapy, and clinical trials are needed to evaluate the feasibility of this intervention and long-term infant dialysis and eventually renal transplantation.

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COMPETING INTERESTS:

Authors have declared that no competing interests exist.

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