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An Antenatal Diagnosis of Potter Syndrome

A Case Report

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Abstract: Potter syndrome, also known as Potter sequence, is a rare and lethal congenital condition caused by bilateral renal agenesis, which leads to severe oligohydramnios, pulmonary hypoplasia, and characteristic facial and limb deformities. We present the case of a 40-year-old gravida 4 para 3 woman referred for evaluation of anhydramnios at 27 weeks gestation. Ultrasound revealed absent fetal kidneys, an empty bladder, and severe oligohydramnios, signs consistent with a diagnosis of Potter syndrome. The fetus didn't show features suggestive of Potter facies or limb deformities. The pregnancy was continued after counseling. The neonate died shortly after birth due to respiratory failure. Potter syndrome carries a uniformly poor prognosis. Prenatal diagnosis is essential for guiding clinical decisions, psychological support, and ethical counseling. A multidisciplinary approach is crucial in managing affected pregnancies and supporting families.

Keywords: *Potter Syndrome, Potter Sequence, Bilateral Renal Agenesis, Pulmonary Hypoplasia, Oligohydramnios, Prenatal Diagnosis, Fetal Anomalies.*

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I. INTRODUCTION

Potter syndrome, or Potter sequence, is a rare and characteristically fatal congenital condition resulting from severe oligohydramnios, most often secondary to bilateral renal agenesis or other significant urinary tract abnormalities (1). First described in detail by Dr. Edith Potter in 1946, the syndrome is considered a sequence because a single initiating anomaly—usually renal agenesis—leads to a cascade of secondary deformities (2).

The syndrome is characterized by an established phenotype that includes pulmonary hypoplasia, limb deformities, and distinct facial features such as low-set ears, flattened nose, and micrognathia—collectively referred to as "Potter facies" (3). The pulmonary hypoplasia is due to prolonged oligohydramnios, and is typically the cause of neonatal demise in affected newborns (4).

The occurrence of Potter syndrome is estimated at approximately 1 in 2,000 to 1 in 5,000 pregnancies, though its true prevalence is difficult to conclude due to the high rate of intrauterine demise or early neonatal death (5). Diagnosis is most commonly made prenatally via ultrasonographic findings, which may include absent, or non-visualized kidneys, empty bladder, severe oligohydramnios, and associated skeletal or craniofacial abnormalities (6). Despite improvements in prenatal imaging and diagnosis, the prognosis remains poor, with most infants dying shortly after birth due to respiratory failure (7).

This case report discusses a prenatally diagnosed case of Potter syndrome, highlighting the sonographic features and fetal MRI findings, diagnostic challenges, and ethical considerations related to counseling and perinatal management.

II. CASE PRESENTATION

We report the case of a 40-year-old gravida 4 para 3 woman referred to our fetal medicine unit at 27 weeks of gestation, for evaluation of severe oligohydramnios noted on routine obstetric ultrasound. The patient had a medical history of type 2 diabetes, with no known consanguinity, and no significant family history of congenital disorders. Her first 2 pregnancies had been uncomplicated; the third pregnancy was marked by in utero fetal death due to suspected genetic

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fetal anomalies no genetic screening was done post partum to the patient's request.

Ultrasound examination revealed anhydramnios, non-visualization of both fetal kidneys, an empty fetal bladder, and severe intrauterine growth restriction. Detailed anatomical examination was limited due to the lack of amniotic fluid but showed abnormal positioning of the lower limbs and a flattened facial profile suggestive of Potter facies.

An MRI was done to confirm the bilateral renal agenesis, it showed empty renal chambers (figure 1, 3) and an empty bladder compartment (Figure2).

Fetal echocardiography was normal. Given the poor prognosis, the patient was counseled extensively regarding the diagnosis of bilateral renal agenesis consistent with Potter syndrome. After multidisciplinary consultation and psychological support, the couple elected to continue the pregnancy.

At 34 weeks, spontaneous preterm labor occurred, and the infant was delivered via vaginal delivery. The male neonate weighed 2,000 grams and exhibited features consistent with Potter syndrome, including flattened nose, low-set ears (figure 4), limb deformity and contractures (figure 5), and severe respiratory distress.

Despite supportive measures, the newborn died within 30 minutes of life. No resuscitation was pursued as per prenatal decisions.

No postmortem ultrasound was conducted to confirm the absence of both kidneys. The parents declined a full autopsy but agreed to genetic counseling and storage of fetal DNA for future analysis.

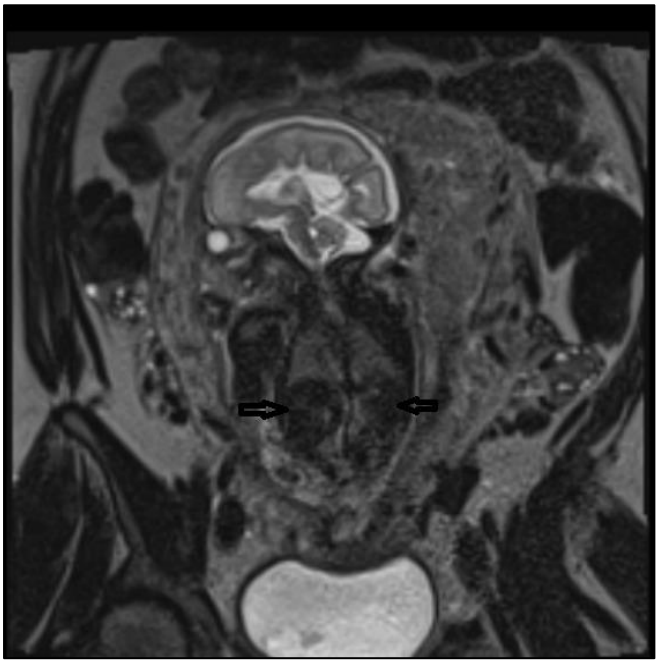

Fig 1 Coronal Section Through the Empty Renal Chambers. (MRI T2 Sequence)

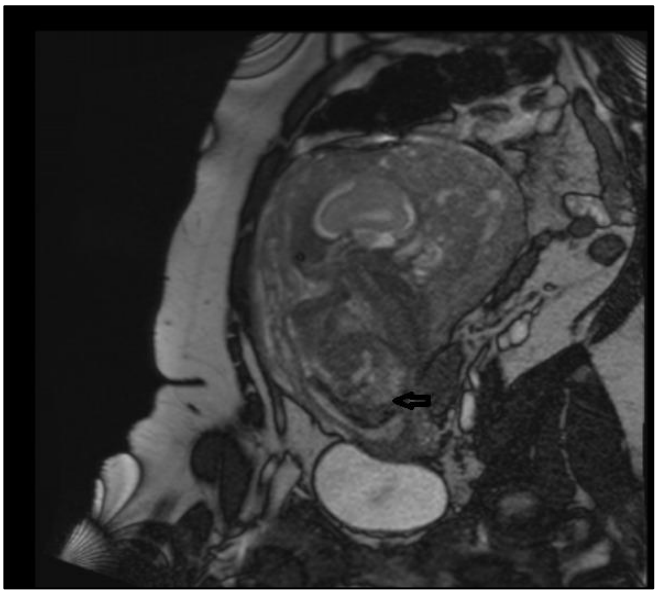

Fig 2 Oblique Sagittal Section of Fetus in Breech Position, Empty Bladder Compartment. (MRI T2 Sequence)

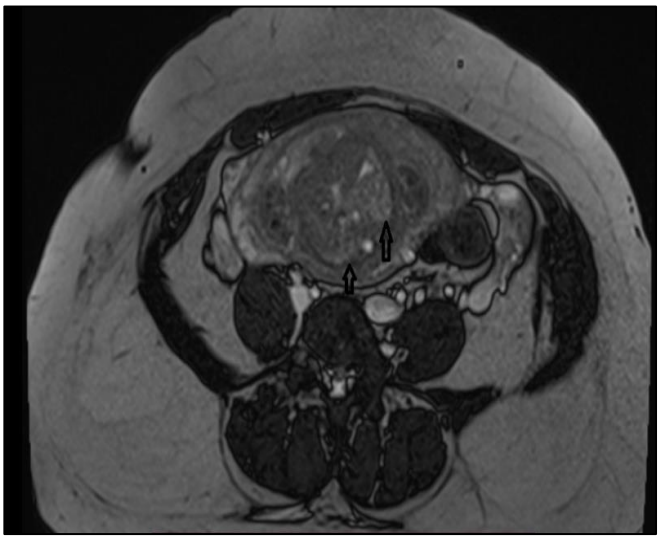

Fig 3 Axial Section Passing Through the Empty Renal Chambers. (MRI T2 Sequence)

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Fig 4 Aspect of Flattened Nose and Low Set Ears.

Fig 5 Aspect of Clubfeet.

III. DISCUSSION

Potter syndrome, or Potter sequence, is a fatal congenital anomaly that results primarily from severe oligohydramnios due to bilateral renal agenesis or other urinary tract malformations (2). In the presented case, prenatal ultrasound findings—including anhydramnios, absent fetal kidneys, an empty bladder, and intrauterine growth restriction—were pathognomonic of this condition. The patient's advanced maternal age and pre-existing type 2 diabetes are notable, as both have been associated with an increased risk of congenital anomalies (8).

The term "sequence" is used because the phenotype results from a cascade of effects initiated by a single primary anomaly—in this case, renal agenesis—which leads to oligohydramnios and subsequent compression-related deformities. These include limb contractures, flattened facies (Potter facies), and pulmonary hypoplasia, the latter being the principal cause of neonatal death (3,4).

The ultrasound diagnosis is usually suspected in the second trimester when oligohydramnios becomes more apparent and renal structures are difficult or impossible to visualize (6). In this case, diagnosis was made at 27 weeks, relatively late, possibly due to the challenges posed by anhydramnios, which limits the ability to perform a complete fetal assessment (9) and established with a fetal MRI exam helping confirm the previously mentioned ultrasound findings.

Although rare, Potter syndrome can be associated with inherited syndromes such as autosomal recessive polycystic kidney disease, Fraser syndrome, or branchio-oto-renal syndrome, especially in cases with a positive family history or consanguinity (7). In this case, the absence of known consanguinity and the decision to pursue genetic counseling and DNA storage were appropriate steps in the evaluation of potential recurrence risk.

Management of Potter syndrome is very limited and largely supportive. Experimental approaches such as serial amnioinfusions have shown limited promise but remain controversial, particularly in the situation of complete bilateral renal agenesis where survival is practically nil (10,11). For this reason, timely diagnosis allows for essential multidisciplinary care planning and psychosocial support. In this case, the parents were counseled thoroughly, and a palliative approach was chosen. This aligns with current ethical recommendations prioritizing informed parental choice and compassionate care in lethal fetal conditions (12).

This case underscores the importance of detailed prenatal assessment in high-risk pregnancies, especially in women with comorbidities such as diabetes. It also highlights the significance of psychological and ethical support in managing pregnancies with a lethal fetal diagnosis.

IV. CONCLUSION

Potter syndrome, primarily resulting from bilateral renal agenesis, carries a uniformly fatal prognosis due to associated pulmonary hypoplasia. Timely prenatal diagnosis allows for appropriate counseling, psychological support, and individualized management planning. Multidisciplinary collaboration is essential in guiding both the medical and emotional journey of affected families. This case underscores the importance of non-directive counseling and empathetic care in the context of lethal fetal anomalies.

- *Ethical Approval*

Ethics approval has been obtained to proceed with the current study.

- *Funding*

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- *Author Contribution*

Hassnaa SARHANE, Kaoutar BAHIDA, Maha LHALOUI, Nouhaila YARTAOUI, RIACHE Hafsa: *study concept and design, data collection, data analysis and interpretation, writing the paper.*

Najia ZERAIDI, Amina ETBER, Aziz BAYDADA: *literature review, supervision*

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