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Case Study

Genetic Evaluation of a 30-Year-Old Female with Unilateral Renal Agenesis: A Case Study

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ABSTRACT Unilateral renal agenesis (URA) is a congenital anomaly where one kidney fails to develop, often diagnosed in childhood or prenatally. While many cases are asymptomatic, URA is associated with long-term renal risks and potential genetic implications. This study presents the case of a 30-year-old female with incidentally detected URA, evaluating her renal function, genetic background, and clinical implications. Ultrasound confirmed a solitary kidney with no surgical history. Renal function tests were normal. Genetic analysis revealed no pathogenic variants, suggesting a sporadic occurrence. This case highlights the importance of URA diagnosis in asymptomatic adults, emphasizing long-term monitoring and genetic evaluation to guide clinical management and patient counseling.

Keywords: Unilateral Renal Agenesis; Kidney; Genetics.

INTRODUCTION

Renal agenesis is the lack of renal tissue arising from an early embryonic developmental abnormality [1]. Additional congenital defects have occasionally been associated with both unilateral and bilateral renal agenesis. The disorder results from the ureteric bud failing to fuse with the metanephric blastema, therefore absenting the nephron and often the ureter. Rare occurring in fewer than one in 3,000 to 4,000 births, bilateral renal agenesis is connected to severe neonatal morbidity and death resulting from lethal pulmonary hypoplasia brought on by the absence of amniotic fluid. By contrast, 1 per 1,000 live infants have URA, usually linked with a good prognosis due to compensatory enlargement of the solitary kidney [2].

Although the origin of congenital kidney and urinary tract anomalies is unknown, considerable evidence points to a genetic link; family clustering has been noted in certain cases [3]. Diverse mutational mechanisms and molecular pathways suggest that about 20% of people with congenital kidney abnormalities may have an undetected inherited disease. Different congenital kidney and urinary tract abnormalities seem to have a shared genetic basis impacted by polygenic inheritance, cytogenetic abnormalities (e.g., copy number variations), and both autosomal dominant and recessive mutations [5].

In this case, we present a patient with URA, highlighting the role of genetic testing in assessing potential hereditary factors. Whether URA arises in isolation or is linked to mutations in important renal developmental genes including PAX2, RET, EYA1, SIX1, and HNF1B, which are linked with renal abnormalities, is mostly dependent on genetic study [6]. Risk assessment, long-term treatment, and family counseling all depend on an awareness of the genetic foundation of URA. This paper emphasizes the need of including genetic testing into clinical practice to improve diagnosis accuracy and maximize patient treatment.

CASE PRESENTATION

A thirty-year-old lady had a full medical checkup, laboratory testing, and ultrasonic imaging. The patient was observed on an ultrasonic scan to have a solitary kidney without accompanying kidney in situ and no signs of surgical excision or scarring. The patient claims no positive urine problems; she has no renal pathology and no recognized congenital defects. Normal serum electrolytes coupled with normal renal function including serum urea at 3.5mmol/L, creatinine at 74 micromole/L, and an eGFR within normal range were shown by laboratory analysis. as hereditary testing was done to screen for PAX2, RET, EYA1, SIX1, HNF1B, and other genes linked with renal development and congenital abnormalities as renal agenesis is typically linked with genetic problems.

To investigate other extra-renal congenital abnormalities like hearing loss, ear pits, cleft lip or palate, and anosmia that commonly have syndromic associations with renal agenesis, a detailed examination was conducted. The study sought to ascertain if the condition was an independent aberration or a component of some syndromic multi-phenotypic disease,

thereby guiding the design of therapy, identification of aetiologic variables, and accurate genetic counseling.

METHODS CASE PRESENTATION

Genetic Testing:

A genetic assessment for the possible origins of URA was performed on the patient. The next step was to conduct next generation sequencing (NGS) on the exons of genes associated with kidney development and their abnormalities. This was done through a targeted gene panel including PAX2, RET, EYA1, SIX1, and HNF1B which are well known for their links with renal agenesis and related disorders of the kidney. The NGS technology employed provided unparalleled sensitivity and specificity in detecting pathogenic variants, which made sustentative diagnosis and clinical intervention feasible.
[6].

Clinical Assessment

Tests for renal funciton, such as serum creatnine and other glomerokus filtration estimations were done to see how well the remaning kidney works. Ultrasound was done to assess the kidney's structure and morphology.

RESULTS

Genetic Testing:

Genetic analysis revealed no pathogenic variants or mutations in genes associated with renal development or congenital anomalies. The patient genetic makeup was within the normal range, suggesting that her URA may not have a significant genetic basis.

Medical Evaluation:

The results for serum creatinine levels and the eGFR were within the anticipated values for a single kidney patient, indicating normal kidney function. The ultrasound confirms that a right solitary kidney exists. In the top image, the kidney is identifiable by its bean shape with identifiable echogenic renal cortex and hypoechoic medulla. In the bottom image, the right side labeled "R" is the indicating the right kidney with normal morphology while the left side "EMPTY" indicates no kidney is present at the position of left renal fossa. This ultrasound report correlates with the description of empty left renal fossa which substantiates the presence of single right kidney.

Table 1: Renal Function Tests. The table indicates the renal function tests with all parameters which show the normal condition for all parameters

Test Name	Result	Reference Range
COLOR	YELLOW	YELLOW
Reaction (pH)	6.0	6.0
Specific Gravity	1.015	1.000 - 1.030
Protein	NIL	NIL
Glucose	NIL	NIL
Acetone	negative	NEGATIVE
Nitrite	Negative	NEGATIVE
Bilirubin	Nil	NIL
Urobilinogen	Normal	NIL
RBC	3-5	< 1
Pus cell/HPF	20-25	1 - 5
Epithelial Cells	+++	NIL
Crystals	NIL	NIL
Casts / H.P.F	NIL	NIL
Parasites & Ova	NIL	NIL
Mucus Threads	NIL	NIL
Others	BACTERIA ++ YEAST CELLS +	NIL

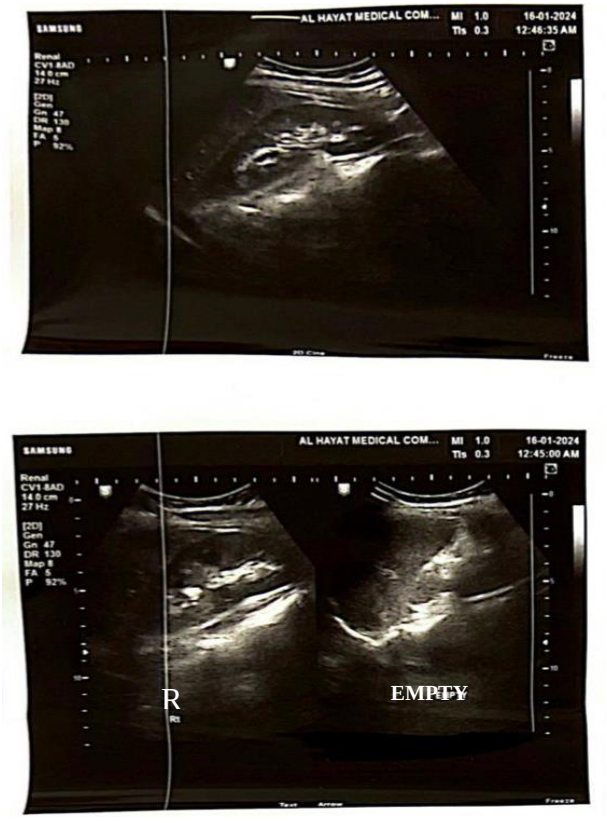

Figure 2: Ultrasound image depicting a solitary kidney with normal morphology and structural integrity. The nephrologist has confirmed that the case is entirely normal, with no abnormalities detected. The abdominal and pelvic ultrasound reveals a normal-sized liver with homogeneous texture, no focal lesions, and normal portal vein (PV), common bile duct (CBD), and biliary tree. The gallbladder has a normal wall thickness with no stones. The right kidney is of normal size, with a preserved corticomedullary differentiation (CMD), normal echopattern, and no evidence of renal gravels or backpressure changes. The left renal fossa is empty.

DISCUSSION

Large pedigrees suitable for linkage analysis of renal agenesis, hypoplasia, and dysplasia are difficult to identify due to the partial penetrance of these conditions, which are influenced by both genetic and environmental modifiers. Furthermore some anomalies, such URA, could be asymptomatic and undetectable without thorough family

screening. Locus heterogeneity poses a difficulty in candidate gene investigations, thereby maybe compromising the effectiveness of linkage studies [7].

The lack of harmful mutations in genes linked to renal development in this patient points to URA perhaps being sporadic or idiopathic rather than clearly genetically derived. Although environmental impacts and developmental abnormalities may also be important in those with a normal genetic background, genetic elements contribute to some cases [8]. This emphasizes how urgently more study on non-genetic causes of renal agenesis is needed. Early detection and therapy of probable consequences depend on long-term renal function monitoring even if later life brings possible hazards related to hypertension, proteinuria, and chronic kidney disease (CKD) [9].

Unilateral renal agenesis is often linked to genetic factors, with mutations in genes like PAX2, RET, EYA1, SIX1, and HNF1B playing a crucial role in kidney development. It is associated with syndromes such as Renal Coloboma Syndrome, Branchio-Oto-Renal Syndrome, Mayer-Rokitansky-Küster-Hauser Syndrome, and VACTERL association, which may present with additional congenital abnormalities [10]. The prevalence of URA-related syndromes varies by region, with consanguinity increasing the risk of inherited renal anomalies. Environmental factors, including maternal diabetes and teratogenic exposures, may also contribute. Given these associations, genetic counseling and targeted genetic testing are essential for diagnosis, prognosis, and reproductive planning [6].

Individuals diagnosed with URA require continuous oversight to maintain renal health and prevent complications such as chronic kidney disease (CKD), proteinuria, and hypertension. Early detection of dysfunction is dependent on regular urinalysis, blood pressure monitoring, and evaluation of renal function. Protective strategies for renal tissues include adequate hydration, a balanced diet, and the avoidance of nephrotoxic medications. Routine ultrasound examinations of the kidneys are essential for identifying structural abnormalities and compensatory hypertrophy. In aging patients, growth monitoring is advisable, and women may require gynecological assessments due to potential links with Müllerian anomalies. In areas with a higher incidence of congenital renal deformities, nephrology monitoring and genetic counseling may be advantageous. Effective intervention and risk reduction depend on the consistent monitoring of renal health, regardless of symptom presentation [2-5].

CONCLUSIONS

This case study highlights the importance of genetic evaluation and medical monitoring in individuals with URA. While some cases may have a genetic basis, others may occur sporadically or have multifactorial etiologies. Understanding the genetic and health implications of URA in individuals with normal genetic makeup is crucial for providing personalized care and genetic counseling.

AUTHOR CONTRIBUTIONS:

The author solely conceived, designed, and executed the study, including data collection, analysis, and interpretation. The author also drafted, revised, and approved the final manuscript for submission.

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ETHICAL CONSIDERATIONS:

The consent of the case was obtained for this case study and genetic testing.

CONFLICTS OF INTEREST

The author declares no conflict of interest.

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