Unilateral Renal Agenesis (URA) — Diagnosis and Complications

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Unilateral agenesis is defined as the complete absence of development of one kidney and ureter. The condition is usually silent and is usually detected on fetal ultrasound scan by the presence of empty renal fossa. The condition is inherited or occurs as a result of spontaneous gene mutations or associated in a syndrome. The incidence of URA is relatively common and is due to genetic as well as maternal causes. Human renal development is initiated by the 5th week of gestation and renal agenesis occurs when the ureteric bud fails to form the ureter, the renal pelvis, and renal mesenchyme. URA is often associated with anomalies of the contralateral kidney and ureter as well.

Sign and Symptoms of Unilateral Renal Agenesis

The disease course is usually silent and is detected on an ultrasound scan of the patients who present with **repeated urinary tract infections or hypertension**. Usually, the patients don't have any complaints due to fully functional other kidney but 20-30% of patients do develop proteinuria and hypertension which is associated with glomerular hyperfiltration.

Unilateral renal agenesis, in some cases also present with other birth defects and problems with the:

- Genitals
- Urinary tract
- Heart
- Lungs
- Stomach and intestines

The patients are at increased risk of Mullerian duct abnormalities leading to infertility and blocked menstrual flow. When unilateral renal agenesis is combined with a blind hemivagina and uterus didelphys, then the condition is known as Herlyn-Werner-Wunderlich syndrome.

Epidemiology of Unilateral Renal Agenesis

The incidence of unilateral renal agenesis is approximately 1 in 2000. The ratio of the condition is slightly more common in males as compared to females and 56% of URA occurs on the left side.

Etiology of Unilateral Renal Agenesis

Unilateral renal agenesis can be caused by genetic causes as well as maternal causes. The condition is associated with mutations in RET (10q11.2), BMP4 (14q22-q23), FRAS1 (4q21.21), FREM1 (9p22.3) or UPK3A (22q13.31), PAX2 (10q24.31), HNF1B (17q12), DSTYK (1q32) genes.

Also, causative factors associated with unilateral renal agenesis are:

- Maternal diabetes
- Maternal obesity
- Young maternal age
- Maternal smoking
- Consumption of large amounts of alcohol during pregnancy

There is a high risk of Hypertension, proteinuria and renal insufficiency is associated with unilateral renal agenesis.

Diagnosis of Unilateral Renal Agenesis

Diagnosis is made by ultrasonography showing an **empty renal fossa and solitary kidney which is larger in size**. Vesicoureteral reflux is usually present in patients with unilateral agenesis and it can be diagnosed by using voiding cystourethrogram. Urinalysis is performed to detect proteinuria and other signs of Urinary tract infections, by the measurement of GFR, urea, and electrolytes.

MRI can be performed for the diagnosis of other non-urinary defects. Genetic tests can be advised to detect abnormality related to an autosomal dominant gene.

Management and Treatment of Unilateral Renal Agenesis

Patients with unilateral renal agenesis require **long-term follow up** because they are at increased risk for development of hypertension and proteinuria. Proper functioning of the remaining solitary kidney should be ensured by kidney function tests such as urine

analysis and glomerular filtration rate.

Prognosis of Unilateral Renal Agenesis

Prognosis is usually good for patients with unilateral renal agenesis. Risk of renal failure is rare and minimal. However, the patients are prone to develop hypertension and proteinuria.

Prevention of Unilateral Renal Agenesis

As the **exact cause of unilateral renal agenesis is not known**, so genetic factors cannot be avoided but maternal factors leading to unilateral renal ageneses like maternal smoking and binge drinking can be avoided or modified accordingly to lower the risk for the development of unilateral renal agenesis in the fetus.

References

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