International Journal of Recent Trends in Science And Technology, ISSN 2277-2812 E-ISSN 2249-8109, Volume 6, Issue 1, 2013 pp 41-42

Congenital Unilateral Renal Agenesis in Association with Genital Anomalies: A Case Report

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Case Report

Abstract: Congenital unilateral renal agenesis occurs in a incidence of 1 in 450 to 1000 birth, and diagnosed incidentally during imaging examination. Genital anomalies occur in 37–60% of females and 12% of males and 25% of them have associated cardiovascular, gastrointestinal, skeletal abnormalitise. Here we present one case of congenital unilateral renal agenesis in association with genital anomalies.

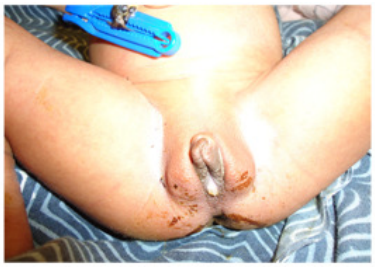
Key Words: Renal agenesis, Polycystic kidney disease.

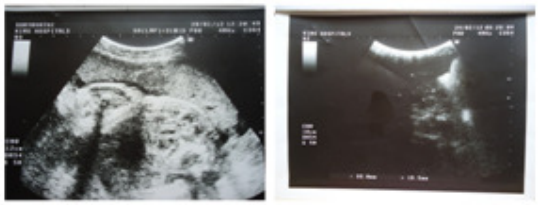
Introduction

Congenital unilateral renal agenesis occurs in a incidence of 1 in 450 to 1000 birth [1].It may be ectopic or malrotated in 5 to 10% of cases. The left kidney is common than right, with males are predominantly more. It is associated with other abnormalities as Genital anomalies (agenesis, , unicornuate or bicornuate uterus, double or absent vagina, absent ovary, absent fallopian tube, abnormal external genitalia) in 37–60% of females and 12% of males(cryptorchidism, seminal vesicle cyst, and hypospadias) and 25% of them with cardiovascular, gastrointestinal, skeletal system[2]. Congenital solitary kidney is compatible with longevity but may be prone to diseases such as pyelonephritis, obstruction and calculus formation [3]. So early detection of a congenital solitary kidney by routine prenatal ultrasound or by incidental imaging studies, alert the physician to look for associated genital anomalies. Here we present one case of prenatal ectopic kidney, became abscent postnataly in association with genital abnormalties, as enlarged clitoris like glans penis, abnormal labia majora with abscence of uterus. This is particularly important in young females, since one of every three with renal agenesis will also have a significant anomaly of the uterus, ovary or vagina.

Case Report

A 2 days old term female child, who presented with abnormal external genitalia as enlarged clitoris, enlarged abnormal labia majora. She had no other organ abnormlity. Family history was essentially negative. Physical examination revealed a healthy looking female child with weight of 2.5 kg, length 52 cm with stable vitals. All Systemic examinations being normal.Genitalia showed detectable abnormalities as enlarged clitoris like glans penis, abnormal labia majora(Figure-1) with absence of uterus by ultrasougraphy. Urinalysis showed a specific gravity of 1.015, pH 7, and negative protein and cells. Serum urea nitrogen was 15 mg%, creatinine 0.9 mg% and normal range electrolytes .Abdominal and pelvic ultrasound revealed a ectopic left kidney with right kidney on right side at the 33 wk gestation in utero(Figure -2) and confirmed absence of the left kidney and uterus with Rt kidney 33×19.5mm at the age of 24 hrs of birth(Figure - 3).





Discussion

Congenital unilateral renal agenesis occurs due to interaction between environmental factors as well as genetic factors during embyogenesis. It may be isolated or may be a part of a multisystem syndrome .It is usually diagnosed prenataly by routine ultrasound or postnataly on an incidental imaging examination. Unilateral renal agenesis may be an expression of a single dominant gene and the association of mullerian agenesis and renal agenesis could be an autosomal dominant disorder [4]. McGillivray et al [5] suggested, searching segment 5q11.2-q13.3 for the gene responsible for hereditary renal dysplasia. The

development of the urinary tract is a sequential and integrated process of the primitive renal elements. Interaction between environmental factors such as maternal illness and exposure to toxic agents, as well as genetic factors, around 15 and 94 days of fetal life result in malformations of this system[6]. Abnormalities of the mullarian system, ovaries and kidney have the same embryologic defect since the wolffian and mullarian ducts develop in close anatomical relationship. A defect in the entire region of the urogenital ridge formed from the gonad and mesonephros could account for this failure in multiple organ developments. Winter et al[7] described the association of renal aplasia or hypoplasia, vaginal atresia and anomalies of the ossicles of the middle ear. When skeletal defects are present, the anomaly is referred to a MURCS association (MUllerian aplasia, Renal aplasia, Cervico-thoracic Somatic dysplasia). Renal agenesis and/or ectopy occur in 88% of MURCS patients. The combination of absence of the vagina, abnormal uterus, renal and skeletal anomalies is known as the Mayer-Rokitanski-Kuster-Hauser syndrome [8]. Unilateral renal agenesis was also reported in patients with Familial Kallmann syndrome, an X-linked syndrome of anosomic, hypogonadotropic hypogonadism[9]. Seventy to 89% of patients with unilateral renal agenesis may have associated genital anomalies [10]. Prenatal diagnosis of the solitary kidney and other renal abnormalities by ultrasound is possible as early as 12 to 16 weeks of gestation. Recognition of a congenital solitary kidney is important in order to monitor the affected individual for urinary infection, obstruction or calculi and warn the individual to avoid contact sports and similar activities that might endanger the solitary kidney.

Conclusion

As during development, first appearance of urinary system induces development of genital system and in young females, since one of every three with renal agenesis will also have a significant genital anomaly. So during early detection of congenital solitary kidney, physician should alert to look for associated genital anomaly .In our case the genital anomalies were like ambiguous genitalia which was not enlisted in literature before and required cytogenetic study in follow up.

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