Original Article

Antenatal diagnosis, prevalence and outcome of congenital anomalies of the kidney and urinary tract in Saudi Arabia

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Abstract

Objective: To study the prevalence, pattern of distribution, and the outcome of different types of kidney and urinary tract anomalies (CAKUT) diagnosed during the antenatal period. The second objective is to test the accuracy of antenatal diagnosis of CAKUT.

Materials and Methods: In a cross-sectional hospital-based study, all cases diagnosed antenatally with urinary tract anomalies at King Abdulaziz University Hospital (KAUH), Jeddah, Kingdom of Saudi Arabia, were studied. The prevalence, pattern of distribution, and immediate postnatal outcomes, in addition to the accuracy of antenatal diagnosis, of those cases are reported.

Results: One hundred and forty-one cases of urinary tract anomalies were antenatally diagnosed; postnatal diagnosis was confirmed in 128 cases (90.1%). The prevalence of CAKUT in our population is 3.26 per 1000 births. The most common abnormalities detected were hydronephrosis, polycystic kidney disease, multicystic dysplastic kidney, and renal agenesis, in descending order of frequency. The perinatal mortality rate among fetuses with CAKUT is 310 per 1000, the majority of these cases (90%) occurred in cases with renal parenchyma involvement.

Conclusions: The prevalence of different types of CAKUT is higher than that reported in developed countries. Urinary tract anomalies can be accurately diagnosed and classified in the antenatal period using ultrasonography imaging. Antenatal diagnosis is a helpful tool in planning immediate postnatal care and deciding the place for delivery. This might prevent or slow renal function deterioration and help in early identification of patients who need early surgical intervention.

Key Words: Hydronephrosis multicystic dysplastic kidney, perinatal diagnosis, polycystic kidney, urinary tract anomalies

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INTRODUCTION

Congenital anomalies of the kidney and urinary tract (CAKUT) represent 20% to 30% of all antenatally diagnosed fetal congenital anomalies in developed countries.[1]

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The North American Pediatric Renal Trials and Collaborative Studies (NAPRTCS)' report indicated that 30% to 50% of cases of end-stage renal disease are related to congenital anomalies of the kidney and the urinary tract;[2] therefore, it is crucial to have early diagnosis and management, whether medical or surgical, to minimize renal damage and to avoid or delay end-stage renal damage.

The prevalence of congenital anomalies of the kidney and the urinary tract as detected during the antenatal period has been reported as being between 0.1% and 0.7%.[3-5] There is a major paucity of literature related to the prevalence of antenatally diagnosed kidney and urinary tract anomalies in Saudi Arabia. The high rate of consanguinity marriages in Saudi Arabia[6] compared to the developed countries necessitates recognition of the prevalence and pattern of these anomalies in order to take the appropriate steps in prevention and management, thereby reducing end-stage renal disease in Saudi society. This study was designed to determine the prevalence of kidney and urinary tract anomalies, to study the pattern of distribution, and to report the immediate outcome of these anomalies in a local hospital based population. A secondary objective is to report the accuracy of antenatal diagnosis compared to postnatal findings.

MATERIALS AND METHODS

A cross-sectional observational study was designed to review the records of all women who had a routine anatomical ultrasound examination between 18 and 20 weeks of gestation or later in the Maternal-Fetal unit at King Abdulaziz University Hospital in the period between January 2001 and December 2010. Scans are initially carried out as screening tools by technicians, residents in training or, fellows. A board-certified perinatologist reviews the case, documents the findings, and issues reports. All women diagnosed as having fetal CAKUT were included in the study. A data collection sheet was designed to record maternal demographic data including age, gravidity, parity, and consanguinity. The CAKUT were classified into hydronephrosis, renal parenchymal malformation, abnormalities of migration and fusion, abnormalities of the collecting system, and abnormalities of the bladder and the urethra. Cases were divided into isolated urinary tract anomalies or urinary tract anomalies in association with other body systems involvement. Cytogenetic studies were reported when available. Data were collected from the Maternal-Fetal unit, labor and delivery records, neonatal intensive care unit (NICU), special baby care unit (SCBU), and nursery.

Pregnancy loss before 20 weeks or fetal weight less than 500 grams was considered as abortion and excluded from the study. Neonatal death (NND) was defined as death within 28 days of life. The fetal and immediate neonatal outcomes were reported, including intrauterine fetal death (IUFD), NND, and NICU admission. Gestational age at the time of delivery, birth weight, and APGAR score were also collected. Data on postnatal diagnoses were collected to test the accuracy of antenatal diagnosis. The study was approved by the hospital's ethical committee. Parental consanguinity was defined as first or second cousin marriage. Statistical analysis was performed using the Statistical Package for Social Sciences Version 16.0 (SPSS Inc, Chicago, IL, USA) software package for data entry. Analyses included frequency distributions and percentages.

RESULTS

During the study period, 141 case of CAKUT were detected out of 43,209 births at King Abdulaziz University Hospital giving a prevalence of 3, 26 per 1000 births. The mean gestational age at the time of antenatal diagnosis was 26 weeks (ranging from 18 to 36 weeks). The mean maternal age was 29.1 ± 3.6 years, gravidity was 3.8 ± 2.67, and parity was 3.6 ± 2.53. The mean gestational age at the time of delivery was 37.6 ± 3.84 weeks and the birth weight was 2.81 ± 1.01 kg. Maternal and neonatal characteristics of all cases diagnosed antenatally are shown in Table 1. The sociodemographic characteristics of the affected fetuses are shown in Table 2.

The majority of the affected fetuses were males: 94 (66.7%). Females were affected in 44 (31.2%) cases, and 3 (2.1%) cases were diagnosed as having ambiguous genitalia.

Regarding consanguinity, 57 (40.4%) of the affected fetuses were products of consanguineous marriages, while 84 (59.6%) were products of non-consanguineous marriages, and in 5 cases, the consanguinity status was not reported. Out of 141 fetuses diagnosed with urinary tract anomalies, 37 (26.2%) had other associated anomalies including cardiovascular, central nervous system, gastro-intestinal, and facial or anterior abdominal wall

Table 1: Maternal and neonatal characteristics of cases with urinary tract anomalies

Maternal	Range	Mean±2SD
Age	16-44 years	29.1±3.6
Gravidity	1-11	3.8±2.67
Parity	1-10	3.6±2.53
Neonatal		
Gestational age	37.6	28–42±3.84
Birth weight (kg)	0.87-5.2	2.81±1.01
APGAR (1 min)	2.0-10.0	7.5±1.9
APGAR (5 min)	4.0-10.0	9.2±1.21

Table 2: Sociodemographic characteristics of infants with congenital anomalies birth

Characteristics	No (%)
Nationality	
Saudi	70 (49.6)
Non Saudi of Arabic nationality	57 (40.4)
Others	14 (9.9)
Consanguinity	
Yes	57 (40.4)
No	84 (59.6.4)
Missing	5 (3.5)
Gender	
Male	94 (66.7)
Female	44 (31.2)
Ambiguous genitalia	3 (2.1)

defects and 104 (73.8%) had isolated urinary tract anomalies. Chromosomal anomalies were detected in three cases antenatally and two cases postnatally.

Hydronephrosis was the most commonly diagnosed fetal abnormality, followed by renal parenchyma abnormalities, abnormalities of the bladder and urethra, collecting system abnormities, and migration and fusion anomalies, respectively. Table 3 demonstrates the pattern of distribution and the frequency of different urinary tract anomalies diagnosed antenatally at King Abdulaziz University Hospital in the 10-year period.

Hydronephrosis was diagnosed in 72 (51.1%) fetuses; Table 4 represents the degree and the site of all cases of antenatally diagnosed fetal hydronephrosis. The left kidney is more likely to be affected than the right and the majority of cases had moderate degrees of hydronephrosis. Out of the 72 cases of hydronephrosis, 45 of the mild and moderate degree hydronephrotic cases resolved spontaneously and were not seen on the 6 week follow-up scan. Seventeen (17) cases of the moderate and severe hydronephrosis showed significant improvement without intervention, and 10 cases needed surgical intervention. Of the 10 cases requiring surgical intervention, 5 had cystoscopy and ureteric stent insertion, 3 had cystoscopy and posterior urethral valve resection, and 2 had pyeloplasty for uretero-pelvic obstruction. Renal parenchyma anomalies were the second most frequent type of CAKUT seen in 58 (41.1%) cases.

The major proportion of renal parenchymal anomalies was polycystic kidney disease, accounting for (17.7%) of the total number of CAKUT. This is followed by multicystic dysplastic kidney (12.8%) and renal agenesis (8.5%) in descending order of frequency [Table 3].

The antenatal diagnosis was confirmed postnatally in 128 (90.1%), of all CAKUT cases; 13 cases (9.9%) were missed. The most likely abnormalities to be missed were duplicated urinary collecting system (six cases) followed by hydroureter (four cases) and pelvic kidney (three case). Renal parenchymal anomalies are the most likely anomalies to be diagnosed accurately in the antenatal period.

The perinatal outcome of the 141 fetuses with CAKUT showed that perinatal death occurred in 31 (21.9%) cases. Out of those cases, 5 (3.5%) had intrauterine fetal demise and 26 (18.4%) had NND, of which 21 cases were early NND and 5 late NND.

The calculated perinatal death rate in CAKUT fetuses was 310 per 1000 live births. Twenty-nine newborns (20.7%) were admitted to the NICU. Ten newborns (7.1%) needed early surgical intervention to relive urinary tract obstruction as a salvage procedure to preserve renal function [Table 5].

In the current study, we antenatally detected 10 cases of recurrent CAKUT in 5 families as follows: 1 family had recurrent multicystic dysplastic kidney, 3 families had recurrence of autosomal-recessive polycystic kidney disease, and 1 family had recurrent Meckel Gruber syndrome. One of these unfortunate families had three subsequent pregnancies affected with autosomal-recessive polycystic kidney disease.

DISCUSSION

Antenatal diagnosis of CAKUT may allow early medical therapy or surgical intervention before renal function

Table 3: Types of congenital anomalies of the urinary tract diagnosed antenatally (*N*=141)

Urinary tract malformation	Cases detected antenatally (%)
Hydronephrosis	72 (51.1)
Malformation of the renal parenchyma	58 (41.1)
Renal agenesis	12 (8.5)
Unilateral	3 (2.13)
Bilateral	9 (6.4)
Multicystic dysplastic kidney	18 (12.8)
Unilateral	13 (9.22)
Bilateral	5 (3.5)
Polycystic kidney disease	25 (17.7)
Meckel Gruber syndrome	1 (0.71)
Solitary cyst	1 (0.71)
Hyperechogenic kidney	1 (0.71)
Abnormalities of migration and fusion	2 (1.42)
Pelvic kidney	1 (0.71)
Horseshoe kidney	1 (0.71)
Abnormalities of the urinary collecting system	4 (2.84)
Duplicated system	0
Dilated ureter and hydronephrosis	4 (2.84)
Ureterocele	0
Abnormalities of the bladder and the urethra	5 (3.5)
Posterior urethral valve	3 (2.13)
Bladder exstrophy	1 (0.71)
Prune-Belly syndrome	1 (0.71)
Total	141 (100)

Table 4: Degree and site fetal hydronephrosis cases (*N*=72) diagnosed antenatally

Hydronephrosis	Right side	Left side	Bilateral	Total
Mild (pylactasis) 5-10 mm	8 (11.1)	9 (12.5)	3 (4.2)	20 (27.8%)
Moderate 10-15 mm	12 (16.7)	18 (25%)	5 (6.9)	35 (48.6%)
Severe >15 mm	6 (8.3%)	8 (11.1)	3 (4.2)	17 (23.6)
Total	26 (36.1)	35 (48.6%)	11 (15.3%)	72 (100%)

Table 5: Perinatal outcome of 141 cases of urinary tract anomalies

Intrauterine fetal demise (*N*/%)	Neonatal death (*N*/%)	Total perinatal death (*N*/%)	NICU admission (*N*/%)	Surgical intervention (*N*/%)
5 (3.5)	26 (18.4)	31 (21.9)	29 (20.7)	10 (7.1)

impairment occurs. In developed countries, the prevalence of CAKUT in live and stillborn infants is 0.3 to 1.6 per 1000.[4,7]

A large multicenter study of renal anomalies in 20 registries of 12 European countries covering 709,030 live births and stillbirths that was performed to evaluate the prevalence of antenatal ultrasonography diagnoses of all types of renal malformation reported a mean prevalence of 1.6 per 1000 births. The most frequent diagnosis reported was upper urinary tract dilatation in 309 (27%) of patients, with 259 (84%) being detected prenatally.[7]

The available data on CAKUT from Saudi Arabia are very scanty and extracted from studies on fetal congenital anomalies in general, with no particular attention to the prevalence or pattern of the CAKUT anomalies. Salute *et al.*[8] showed that genitourinary anomalies were the most common fetal anomalies diagnosed in the antenatal period constituting 38.6% of all anomalies diagnosed in that center. However, the prevalence and pattern of CAKUT was not reported. The prevalence of antenatally diagnosed CAKUT in the current study is 3.26 per 1000 births. This figure is higher than that reported from the western countries. The higher rate in this study can be explained by the facts that we are a tertiary care referral center for perinatal diagnosis in the western region of Saudi Arabia and that we have a higher number of consanguinity marriages in our local population. Excluding hydronephrosis cases from our study decreases the prevalence to 1.56 per thousand births.

Fetal hydronephrosis is a common finding on antenatal ultrasound examination occurring in 0.5% to 1% of pregnancies.[9] In a meta-analysis by Lee *et al.*[10] that included 17 studies, antenatal hydronephrosis was identified in 1678 fetuses of 104,572 women (1.6%). However, the criteria for the diagnosis of hydronephrosis differed among the included studies.

Of all antenatal CAKUT anomalies in the current study, hydronephrosis is the most frequent abnormality seen (51.1%) representing 1.7 per 1000 births. The majority of hydronephrosis cases occurred in male fetuses (66%) and unilateral hydronephrosis was detected in 84.7% of the cases. This finding is in concordance with that reported by González *et al.*[11]

Our data showed that renal parenchymal malformation comprises 41.1% the antenatally diagnosed CAKUT. Polycystic kidney disease was the most frequent diagnosis, followed by multicystic kidney and renal agenesis, respectively [Table 3]. Polycystic kidney disease is one of the major causes of end-stage renal disease in children and adults. Polycystic kidneys in a fetus should prompt investigation of the kidneys in the rest of the family.

Because of the similarity of fetal ultrasound features between autosomal recessive polycystic kidney and autosomal dominant polycystic kidney, we reported the two anomalies as polycystic kidney disease. The reported prevalence in our study is 0.57 per 1000 births.

The prevalence of multicystic dysplastic kidney in the current study is 0.4 per1000 births with the majority of cases being unilateral. This finding concurs with what was reported from the developed countries.[12] Renal agenesis ranked third in the renal parenchymal anomalies in our institution with a prevalence of 0. 27 per1000 births, which is almost the same figure as the one reported in the United States by Parikh *et al.*[13]

Anomalies of migration and fusion of the urinary collecting system and anomalies of the bladder and urethra were less frequent than renal parenchymal lesions, constituting 11% of the diagnoses of CAKUT in the current study. Although previous literature reports that complete or partial duplication of the renal collecting system is a common congenital anomaly of the urinary tract in children,[14] we believe that the low detection of these anomalies in our study is due to false negative antenatal diagnosis. Four of our cases were diagnosed as hydronephrosis and postnatal diagnosis showed that they were in fact duplicate renal collecting system.

In the multi-center European study by Wiesel *et al.*,[7] the association of CAKUT and non-renal congenital anomalies were seen in about 30% of cases. Our data showed the association in 26.2% of the cases. The perinatal loss was significantly higher among fetuses with CAKUT, with a perinatal mortality rate of 310:1000 births compared to 17.7:1000 births in non-affected fetuses in our institution during the 10-year study period. The majority of perinatal deaths in our study occurred renal parenchymal abnormalities, including all cases with bilateral renal agenesis, bilateral multicystic dysplastic kidney, and some cases of polycystic kidney disease.

Similar findings were reported by Damen *et al.*[15] who studied the outcome of 402 cases of congenital renal tract anomalies and found that in 106 out of 402 (26.4%) deceased children, the cause of death was directly related to the renal tract anomalies.

CONCLUSIONS AND RECOMMENDATIONS

The prevalence of kidney and urinary tract anomalies is relatively higher than that reported in developed countries. However, the pattern of distribution of CAKUT is similar. Antenatal ultrasound examination should be performed routinely for all pregnant women to diagnose fetal congenital

anomalies and help in establishing plans for post natal care to optimize the outcome, and reduce or delay the progress of renal function deterioration and the end-stage disease.

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