Original Research Article

# A study of diagnostic evaluation of congenital malformations of urinary tract at tertiary health care center

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## Abstract

**Background:** Prenatal diagnosis of congenital disease provides information for decisions during pregnancy and appropriate treatment parentally, it is assumed to improve perinatal and long-term outcome. Kidney malformations are commonly identified in the antenatal period and account for 20-30% of all detectable anomalies. Present study was aimed to study methods of diagnostic evaluation of congenital malformations of urinary tract at a tertiary health care center. **Material and Methods:** A prospective observational study was carried out in neonates whose antenatal anomaly scans showed presence of CAKUT (Congenital anomalies of the kidney and urinary tract) delivered at our institute or referred within 24 hours of birth, completed 6 months follow-up or died within 6 months of birth were considered for present study. **Results:** During study period 68 cases were studied. initially all anomalies were diagnosed on USG examination (antenatally – 86.76% and postnatally – 13.24%). Few patients underwent additional radiological investigations such as MRI examination (antenatally- 7.35% and postnatally - 5.88 %) and Postnatal CT scan examination in 5.88% (as antenatally CT is relatively contraindicated). In present study hydronephrosis (30.88%) was most common congenital malformations of urinary tract followed by polycystic kidney disease (20.59%), pelviureteric junction obstruction (19.12%), posterior urethral valve (17.65%), ambiguous genitalia (4.41%), epispadius (2.94%), duplicated collecting system (2.94%), hypospadias (1.47%) and bladder exstrophy (1.47%). **Conclusion:** Hydronephrosis was most common antenatally as well as postnatally detected CAKUT. Anomalies of renal collecting system were better detected by antenatal scans than other renal anomalies.

**Keywords:** Hydronephrosis, CAKUT, antenatal USG, anomaly scan.

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## INTRODUCTION

Prenatal diagnosis of congenital disease provides information for decisions during pregnancy and appropriate treatment parentally (timed delivery in tertiary care centers), it is assumed to improve perinatal and long-term outcome. The occurrence of congenital anomalies shows the variance in different countries and India contribute around 28% global burden of neonatal mortality due to congenital anomalies.[1] It has been reported that in India congenital anomalies account for 8-15% perinatal mortality and 16% neonatal mortality.[2] Kidney malformations are commonly identified in the antenatal period and account for 20-30% of all detectable anomalies.[3] Severity can range from mild antenatal pelviectasis to bilateral renal agenesis. These disorders impact approximately 2% of all pregnancies and are often associated with additional developmental abnormalities or genetic syndromes.[4] Congenital abnormalities of the genitourinary tract are the most common sonographically identified malformations, with an incidence of 1 to 4 in

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1000 pregnancies.[5] As such they represent 15-20% of all prenatally diagnosed congenital anomalies, obstructive uropathies accounting for the majority of cases.[6] With the introduction of modern ultrasound screening programs, about 60% of children having surgery for renal or urinary tract problems in their first five years of life are identified by prenatal ultrasound. Present study was aimed to study methods of diagnostic evaluation of congenital malformations of urinary tract at a tertiary health care center.

## MATERIAL AND METHODS

A prospective observational study was carried out at the Department of Paediatrics in a tertiary care hospital in India. Clearance was obtained from Institutional Ethics Committee. Study period was from January 2020 to December 2021. Neonates whose antenatal anomaly scans showed presence of CAKUT (Congenital anomalies of the kidney and urinary tract) delivered at our institute or referred within 24 hours of birth, completed 6 months follow-up or died within 6 months of birth were considered for present study. Maternal details such as age, weight, gravida, parity and consanguinity, detailed obstetric history of any risk factor for development of congenital renal or urinary tract anomalies like history of oligohydramnios, poor weight gain or hypertension, diabetes, obesity, increase salt intake, alcohol consumption, exposure to teratogens, etc. during pregnancy was obtained from mother's hospital records. Details of postnatal events including gestational age, Apgar scores, birth weight presence of another associated congenital anomaly was also documented and was confirmed. A postnatal ultrasonography was performed by a skilled observer using the ALOKA Prosound alpha 6 ultrasound machine having a frequency of 3 to 7 MHZ using curvilinear probe. Outcomes were assessed with the following parameters-Postnatal persistence of antenatally diagnosed CAKUT and persistence during the follow up period of six months, the degree and progression of CAKUT and comparison with previous scans, any surgical intervention, the findings of additional investigations such as MCU, DMSA and DTPA scans, if done, were noted, development of other complications due to CAKUT such as UTI, deranged renal function and outcome at the end of the six months follow up period. Data was collected and compiled using Microsoft Excel, analysed using SPSS 23.0 version. Statistical analysis was done using descriptive statistics.

## RESULTS

During study period 68 cases were studied. Most common maternal age group was 21-25 years (32.35 %), followed by 26-30- and 31-35-years age group (22.06 % each). Parental consanguity was noted in 27.94 % cases. Majority were gravida 2/3 (39.71 %) followed by primigravida (32.35%). 72,06 % were male babies, 23.53 % were female and 4.41 % had ambiguous genitalia. 48.53 % babies were delivered at term, 57.35 % were delivered vaginally while 39.71 % required caesarean delivery. Most cases were detected in third trimester (55.88 %), 30.88 % diagnosed in second trimester and 13.24 % were diagnosed after delivery. Though 30.88 % were diagnosed in second trimester, patients did not opted termination.

**Table 1:** General Characteristic.

| Characteristic | No of neonates (n=68) | Percentage (%) |
|---|---|---|
| Maternal age (years) | | |
| <20 | 5 | 7.35 |
| 21-25 | 22 | 32.35 |
| 26-30 | 15 | 22.06 |
| 31-35 | 15 | 22.06 |
| >35 | 11 | 16.18 |
| Parental Consanguinity | | |
| Present | 19 | 27.94 |
| Absent | 49 | 72.06 |
| Order of pregnancy | | |
| G 1 | 22 | 32.35 |
| G 2-3 | 27 | 39.71 |
| G 4 or more | 19 | 27.94 |
| Gender of baby | | |
| Male | 49 | 72.06 |
| Female | 16 | 23.53 |
| Ambiguous | 3 | 4.41 |
| Pregnancy termination according to gestational age | | |
| 21-28 weeks | 12 | 17.65 |
| 29-37 weeks | 23 | 33.82 |
| >37 weeks | 33 | 48.53 |

| | | |
|---|---|---|
| Mode of delivery | | |
| Vaginal delivery | 39 | 57.35 |
| Caesarean section | 27 | 39.71 |
| Instrumental | 2 | 2.94 |
| Time of detection of anomaly. | | |
| 1st trimester | 0 | 0.00 |
| 2nd trimester | 21 | 30.88 |
| 3rd trimester | 38 | 55.88 |
| After delivery | 9 | 13.24 |

Initially all anomalies were diagnosed on USG examination (antenatally – 86.76 % and postnatally – 13.24 %). Few patients underwent additional radiological investigations such as MRI examination (antenatally- 7.35 % and postnatally - 5.88 %) and Postnatal CT scan examination in 5.88 % (as antenatally CT is relatively contraindicated).

**Table 2:** Diagnostic modality

| Diagnostic modality | No of cases | Percentage (%) |
|---|---|---|
| USG examination | | |
| Antenatal | 59 | 86.76 |
| Postnatal | 9 | 13.24 |
| MRI examination | | |
| Antenatal | 5 | 7.35 |
| Postnatal | 4 | 5.88 |
| CT | | |
| Postnatal | 4 | 5.88 |

In present study hydronephrosis (30.88 %) was most common congenital malformations of urinary tract followed by polycystic kidney disease (20.59 %), pelviureteric junction obstruction (19.12 %), posterior urethral valve (17.65 %), ambiguous genitalia (4.41 %), epispadius (2.94 %), duplicated collecting system (2.94 %), hypospadias (1.47 %) and bladder exstrophy (1.47 %).

**Table 3:** Congenital malformations

| CAKUT anomaly | No of cases | Percentage (%) |
|---|---|---|
| Hydronephrosis | 21 | 30.88 |
| Polycystic kidney disease | 14 | 20.59 |
| Pelviureteric junction obstruction | 13 | 19.12 |
| Posterior urethral valve | 12 | 17.65 |
| Ambiguous genitalia | 3 | 4.41 |
| Epispadias | 2 | 2.94 |
| Duplicated collecting system | 2 | 2.94 |
| Hypospadias | 1 | 1.47 |
| Bladder exstrophy | 1 | 1.47 |

## DISCUSSION

Prenatal diagnosis improves the outcome of the affected child because of early recognition and treatment of critical obstructions and urinary tract infections, preventing further renal damage and loss of renal function. Important developments have taken place in prenatal imaging including fetal magnetic resonance imaging and Doppler analysis. Proteomics of urine and amniotic fluid have been studied to determine prognostic biomarkers of prenatal renal injury. High resolution ultrasonography identifies various structural abnormalities with clarity, and serial estimation of the amniotic fluid index helps the obstetrician to decide about conducting preterm delivery. Ultrasonography, magnetic resonance imaging, and nuclear imaging are crucial in diagnosis and subsequent observation. Noninvasive urodynamic studies including uroflow and postvoid residual measurement are often sufficient to evaluate the function of lower urinary tract.[7] Renal agenesis is usually unilateral with the prevalence of 1 in 1500–3200 live births and more common in males.[8] Renal agenesis is one of the most profound renal tract malformation characterized by complete absence of kidney development and is often accompanied by an absent ureter. Four theories have been proposed as the cause as failure of the metanephric bud to appear in spite of a normally preceding mesonephros, early regression of the metanephros, imperfect development of mesonephros and non-development of pronephros leading to non-growth of mesonephros.[9] Renal agenesis has also been associated with chromosomal abnormalities such as 21, 22, 7 and 10 trisomy's, 45 X mosaicism and 22q11 microdeletion.[10] Hekmat C *et al.*[11] studied antenatally diagnosed renal and excretory tract abnormalities, noted that MCDK was the most frequently diagnosed fetal abnormality (44.44%), followed by Posterior urethral valves (PUV) (22.22%),

renal agenesis (13.89%), Autosomal recessive polycystic kidney disease (ARKD) (11.11%), Ureteropelvic junction (UPJ) obstruction (11.11%), duplicated collecting system (8.33%) and isolated fetal pyelectasis observed in two cases with a percentage of 5.56% with a predominance on the left side. In study by Saryu Gupta *et al.*,[12] mean age of study participants was 29 ± 3 years. Amniotic fluid was reduced or absent in 41% (N = 13) and normal in 59% (N = 18) of participating mothers. Overall, urinary obstruction was the commonest anomaly encountered (29%) followed by the multicystic dysplastic kidney (MCDK) (22%). Bilateral renal disease was seen in all mothers having features of anhydramnios {B/L MCDK (N = 3), autosomal recessive polycystic kidney disease (ARPKD) (N = 2), posterior urethral valves (PUV) (N = 2), B/L renal agenesis (N = 3), and megacystis (N = 1)}. Fusion anomalies (horseshoe kidney) and rotation anomaly (malrotation) were detected in one case each. Additional extrarenal findings were seen on fetal MRI in 35% (N = 11) cases. Fetal MRI improves diagnostic accuracy in anomalies affecting the fetal kidney and genito-urinary systems by better morphological delineation. Chougule A, *et al.*,[13] noted that CAKUT was more common among males. Hydronephrosis was the most common CAKUT in antenatal scans. Anomalies of the renal collecting system formed 93.9% of all CAKUT detected on antenatal anomaly scan and 57.4% of these resolved by six months of age. Abnormalities of the renal collecting system together formed 93.9% of all antenatally diagnosed CAKUT and were more common than abnormalities of the renal parenchyma which formed 6.1%. Postnatal resolution on day three USG was seen in seven out of 22 (31.8%) cases of antenatally diagnosed mild hydronephrosis irrespective of their site. Out of the 28 antenatally diagnosed hydronephrosis, 11 (39.3%) resolved at some point during the follow up period of six months. Kumar BH *et al.*,[14] noted that primary VUR was the commonest CAKUT followed by PUJO, MCDK, non-obstructive hydronephrosis and PUV. In contrast, Soliman, *et al.*,[15] reported PUV (36.4%) as the commonest followed by primary VUR (19.6%) and PUJO (18.7%). Aksu, *et al.*, [16] reported PUJO in 62.7% followed by VUR in 16.6%. A meta-analysis by Lee *et al.*[17] demonstrated that every dilatation of the urinary tract regardless the degree, was associated with an overall increased risk (36%) of an underlying uropathy. The risk was depending on the severity of the hydronephrosis, increasing from 12% in cases for mild hydronephrosis (APPd > 7 mm in the 2nd trimester and < 9 mm in the 3rd trimester), to 45% in cases of moderate hydronephrosis (APPd 7-9 mm in the 2nd trimester, 9- 15 mm in the 3rd trimester) and up to 88% in cases of severe hydronephrosis (> 10 mm in the 2nd and > 15 mm in the 3rd trimester). MR imaging is mandatory in the evaluation of associated spinal anomalies. MR urography can demonstrate ectopic extravesical ureteric insertions, thereby providing a global view of the malformation. Familiarity with anomalies of the lower urinary tract is essential for correct diagnosis and appropriate management.

## CONCLUSION

Hydronephrosis was most common antenatally as well as postnatally detected CAKUT. Anomalies of renal collecting system were better detected by antenatal scans than other renal anomalies. Computed tomography and magnetic resonance (MR) imaging are unsuitable for general screening but provide superb anatomic detail and added diagnostic specificity.

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