Original Research Article

A study of prevalence of various types of congenital malformations of urinary tract at tertiary health care center

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Abstract

Background: Congenital anomalies of the kidney and urinary tract (CAKUT) are observed in three to six per 1000 live births and account for 40–50% of the etiology of chronic kidney disease (CKD) in children worldwide. They has the potential to impact fetal lung maturity which in turn has important prognostic implications. Present study was aimed to study prevalence of various types of congenital malformations of urinary tract at tertiary health care center. **Material and Methods:** Present study was institution based, cross-sectional, observational study, conducted in neonates with antenatal ultrasound suggestive of an abnormality of the urinary tract, delivered at our hospital or referred from outside within 24 hours of birth. **Results:** During study period 68 cases were studied. 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. Abnormalities of AFI (52.94 %), family history of congenital anomaly (4,41 %), were maternal risk factors noted. 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 %). In present study 42.65 % cases required observation only, while 26.47 % required surgery in neonatal period and 19.12 % were referred to higher centre for the same. Neonatal death was noted in 11.76 % cases. **Conclusion:** Hydronephrosis was most followed by polycystic kidney disease, pelviureteric junction obstruction, posterior urethral valve were common congenital malformations of urinary tract noted. Majority required observation only while few required surgical intervention.

Keywords: congenital malformations of urinary tract, hydronephrosis, polycystic kidney disease, pelviureteric junction obstruction,

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Received Date: 04/10/2021 Revised Date: 10/11/2021 Accepted Date: 21/10/2021

Access this article online

Quick Response Code:

Website: www.medpulse.in

DOI: https://doi.org/10.26611/10142031

INTRODUCTION

Congenital anomalies of the kidney and urinary tract (CAKUT) are observed in three to six per 1000 live births and account for 40–50% of the etiology of chronic kidney disease (CKD) in children worldwide.[1,2] Congenital renal anomalies can be sporadic or familial, syndromic (also affecting non-renal tissues) or non-syndromic.[3] The primary insults believed to be associated with the development of CAKUT are environmental factors and genetic mechanisms. Various genetic loci associated with syndromic CAKUT are identified. In animal models, deletion of several of these genes results in low nephron endowment including PAX2, p53, GLI3R, GDNF, FGF 7, FRS2 and Six2.[4,5] Congenital anomalies of the kidney and urinary tract (CAKUT) comprises of a spectrum of defects involving kidney (hypoplasia and dysplasia), ureters and bladder (vesico-ureteral reflux, duplex system) and urethra

How to cite this article: Santosh Bajaj, Kalpana Jaju. A study of prevalence of various types of congenital malformations of urinary tract at tertiary health care center. *MedPulse International Journal of Pediatrics.* December 2021; 20(2): 43-47.
http://medpulse.in/Pediatrics/index.php

(posterior urethral valves) that results from perturbations in the development of the kidney and urinary tract during fetal life. [6,7] The malformations may affect the kidney and urinary tract unilaterally and bilaterally or may occur in syndromic forms with other organ defects.[8] In addition, urinary tract disease has the potential to impact fetal lung maturity which in turn has important prognostic implications. An accurate diagnosis of these anomalies is therefore of paramount importance and can prove to be valuable in formulating management strategies and influencing parental counseling.[9] Present study was aimed to study prevalence of various types of congenital malformations of urinary tract at tertiary health care center.

MATERIAL AND METHODS

Present study was institution based, cross-sectional, observational study, conducted in department of pediatrics in collaboration with department of obstetrics and gynecology at XXX medical college and hospital, XXX, India. Study duration was of 2 years (January 2020 to December 2021).

Inclusion criteria: Neonates with antenatal ultrasound suggestive of an abnormality of the urinary tract, delivered at our hospital or referred from outside within 24 hours of birth

Exclusion criteria: Neonates not completed 6 months follow-up

The written informed consent from the parents and approval from the institutional ethical committee was obtained. The demographic variables which included age, weight, gravida, parity and consanguinity were recorded. A 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. Antenatal Details such as Age of diagnosis, biometry and amniotic fluid, morphology of the urogenital system, diagnoses suspected by ultrasound, topographic distribution, associated extra-renal malformations and syndromic approach, outcome of pregnancy and gestational age, fetal sex, Apgar score and the Weight at birth was noted. Outcomes were assessed with any 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 CT scan, MRI, MCU, DMSA and DTPA scans, if done. Data was collected and compiled using Microsoft Excel, analysed using SPSS 23.0 version. Statistical analysis was done using descriptive statistics. Frequency, percentage, means and standard deviations (SD) was calculated for the continuous variables, while ratios and proportions were calculated for the categorical variables.

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

Abnormalities of AFI (52.94 %), family history of congenital anomaly (4,41 %), maternal hypothyroidism (2.94 %), uncontrolled maternal diabetes mellitus (2.94 %), history of fever with rash in ANC period (2.94 %), history of unknown drug exposure (2.94 %) and history of exposure to X-ray antenatally (1.47 %) were maternal risk factors noted in present study.

Table 3: Maternal risk factors

Risk Factors	No of cases	Percentage (%)
Abnormalities of AFI	36	52.94
Family h/o of cong. Disease	3	4.41
Hypothyroidism	2	2.94
Uncontrolled maternal diabetes mellitus	2	2.94
H/o Fever with rash in ANC period	2	2.94
H/o unknown drug exposure	2	2.94
H/o exposure to X-ray antenatally	1	1.47

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 4: 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

In present study 42.65 % cases required observation only, while 26.47 % required surgery in neonatal period and 19.12 % were referred to higher centre for the same. Neonatal death was noted in 11.76 % cases.

Table 5: Neonatal outcome

Outcome	No. of cases	Percentage
Observation only	29	42.65
Surgery required	18	26.47
Referred to higher center	13	19.12
Neonatal death	8	11.76

DISCUSSION

Major structural anomalies appear in 2-3% in live newborns and other 2-3% are discovered in children up to 5 years old, summarizing 4-6%. Minor anomalies appear in approximately 15% out of the total of newborns.[10] These anomalies do not alter the individual's health status, but they are associated with major defects in some cases,

therefore they can serve as key elements for the diagnosis of more serious, hidden defects. CAKUT is a clinically heterogeneous phenotype that encompasses renal agenesis, renal hypoplasia/dysplasia, multicystic kidney dysplasia, cross-fused ectopia, duplex renal collecting system, ureteropelvic junction obstruction, mega-ureter, posterior urethral valves, and vesicoureteral reflux (VUR). High resolution ultrasonography, magnetic resonance imaging, and nuclear imaging are crucial in diagnosis and subsequent observation.[11] Ameen, S.K.*et al.*,[12] at Maternity Teaching Hospital, Erbil noted rate of congenital anomaly as 3.63/1000 deliveries. The most common area for anomalies was the central nervous system (37.7%) followed by the musculoskeletal (23.1%) and gastrointestinal systems (20.8%). There was a statistically significant association between having a child with congenital anomalies and a maternal history of previous congenital anomalies (odds ratio [OR] 59.0, 95% CI 5.74–607.0), parental consanguinity (OR 6.26, 95% CI 2.42–16.19), and history of medical disorders (OR 153.2, 95% CI 25.9–905.4). While Hekmat Chaara *et al.*, [13] 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. Chougule A, *et al.*,[14] 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. Veerabhadra R *et al.*,[16] studied 81 patients (neonates and children below 13 years of age) with CAKUT, Twenty-two (27%) patients were underweight, 4 (5%) patients were stunted, and 26 (32%) were both underweight and stunted. Children with bilateral disease had a higher incidence of underweight (21/44 vs. 8/37; $p = 0.04$), and both underweight and stunted (25/44 vs. 10/37; $p = 0.006$) compared to children with unilateral disease. Hypertension was found in 27% cases. Bilateral disease in CAKUT was significantly associated with poor somatic growth. Unilateral renal agenesis occurs in approximately 1/1,300 pregnancies, the majority of which are probably cases of renal aplasia.[17,18] The difference between both cannot be made by ultrasound. Although unilateral renal agenesis is mostly isolated and sporadic, it might be part of a genetic syndrome, or occur in association with chromosomal or developmental defects (VACTeRL association) and genital abnormalities. The etiology of antenatal hydronephrosis is very diverse and majority of cases are transient (48%) or physiological (15%). Only in a minority of cases, a significant underlying pathology of the urinary tract is found. Rare causes are an ectopic ureter, prune belly syndrome, urachal cysts and urethral atresia.[19] The likelihood of an underlying abnormality correlates with the severity of the hydronephrosis. Transient or physiological hydronephrosis will mostly be mild, while ureteropelvic junction (UPJ) stenosis almost always presents with severe hydronephrosis. Tertiary care hospital usually do not have definite catchment area and complicated cases are more commonly encountered. Hence, prevalence calculated in this type of hospital-based study cannot be projected to the total population. Community based study should be ideal for true estimation of incidence of congenital anomalies in a population. Although congenital anomalies may be the result of one or more genetic, infectious, nutritional or environmental factors, it is often difficult to identify the exact causes. Some congenital anomalies can be prevented by vaccination, adequate intake of folic acid or iodine through fortification of staple foods or supplementation, and adequate antenatal care.

CONCLUSION

Antenatally diagnosed CAKUT were more common among male fetuses. Hydronephrosis was most followed by polycystic kidney disease, pelviureteric junction obstruction, posterior urethral valve were common congenital malformations of urinary tract noted. Majority required observation only while few required surgical intervention.

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Source of Support: None Declared
Conflict of Interest: None Declared