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Original Research Article

A Research of Diagnostic Evaluation of Inherited Malformations of Urinary Tract: A Cross Sectional Research at Tertiary HealthCare Center

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Abstract

Background: Pre-natal determination of inherited disease gives information for making pregnancy-related decisions and providing appropriate management to parents, with the goal of improving perinatal and long-term outcomes. Renal abnormalities are frequently detected during pregnancy, accounting for 20-30% of all detectable defects.

Aims & objectives: The goal of this research was to look at methods of diagnosing inherited urinary system abnormalities at a tertiary health care facility.

Material and Methods: A prospective observational research was conducted in neonates who were delivered at our institute or referred within 24 hours of birth, completed 6 months of follow-up, or died within 6 months of birth and whose antenatal anomaly scans revealed the presence of CAKUT (Inherited anomalies of the renal and urinary tract).

Results: 136 instances were studied over the course of the research period. Initially, all abnormalities were discovered during a USG test (86.76 percent antenatally and 13.24 percent post-natally). Only a few patients had further radiological tests, such as an MRI (antenatally 7.35 percent and post-natally 5.88 percent) and a post-natal CT scan (also 5.88 percent) (as antenatally CT is relatively contraindicated). The commonest inherited malformation of the urinary tract was hydronephrosis (30.88%), followed by polycystic renal 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%). (1.47 percent).

Conclusion: CAKUT discovered hydronephrosis most frequently both antenatally and post-natally. Antenatal imaging detected renal collecting system defects better than other renal anomalies.

Keywords: Hydronephrosis, CAKUT, antenatal USG, anomaly scan

Introduction

Pre-natal determination of inherited illness gives information for making pregnancy-related decisions and providing appropriate parental therapy (timed delivery in tertiary care centers), which is thought to improve perinatal and long-term outcomes [1]. Inherited malformations occur at varying rates in different countries, with India accounting for roughly 28% of the worldwide burden of infant mortality owing to inherited defects. Inherited abnormalities are said to cause for 8-15 percent Pre-natal mortality and 16 percent newborn mortality in India. Renal abnormalities are frequently detected during pregnancy, accounting for 20-30% of all detectable defects [2-4]. The severity of Pre-natal pelviectasis can range from mild to bilateral renal agenesis. These diseases affect about 2% of all pregnancies and are frequently linked to other developmental defects or hereditary syndromes [5-7]. The commonest sonographically diagnosed anomalies are genitourinary tract abnormalities, which occur in 1 to 4 out of every 1000 pregnancies. As a result, they account for 15-20% of all inherited malformations detected before birth, with obstructive uropathies accounting for the majority of cases [8,9]. Pre-natal ultrasound has identified roughly 60% of children who have surgery for renal or urinary tract disorders in their first five years of life since the introduction of sophisticated ultrasound screening programs [10].

Aims & objectives: The goal of this research was to look at methods of diagnosing inherited urinary system abnormalities at a tertiary health care facility.

Material and Methods

A prospective observational research was conducted in a tertiary care hospital in India's Department of Paediatrics. The Institutional Ethics Committee gave their approval. The research took place from January 2020 to December 2021. The current research included neonates whose antenatal anomaly scans revealed the presence of CAKUT (Inherited abnormalities of the renal and urinary tract), who were delivered at our institute or referred within 24 hours of birth, completed 6 months of follow-up, or died within 6 months of birth. Maternal information such as age, weight, gravida, parity, and consanguinity, as well as a detailed obstetric history of any risk factors for the development of inherited renal or urinary tract anomalies during pregnancy, such as oligohydramnios, poor weight gain or hypertension, diabetes, obesity, increased salt intake, alcohol consumption, teratogen exposure, and so on, were obtained from the mother's hospital records. Post-natal events were also documented and confirmed, including gestational age, Apgar scores, birth weight, and the presence of another related inherited abnormality. A experienced observer used the ALOKA Prosound alpha 6 ultrasound equipment with a frequency of 3 to 7 MHZ and a curvilinear probe to perform a post-natal ultrasonography. The following parameters were used to evaluate outcomes: post-natal persistence of antenatally diagnosed CAKUT and persistence during the six-month follow-up period, the degree and progression of CAKUT and comparison to previous scans, any surgical intervention, the findings of additional investigations such as MCU, DMSA, and DTPA scans, if done, development of other CAKUT-related complications such as UTI, deranged renal function, and outcome at the end of the research. Microsoft Excel was used to collect and compile data, while SPSS 23.0 was used to analyze it. The descriptive statistics were used in the statistical analysis.

Result

136 instances were studied over the course of the research period. The most prevalent maternal age group (32.35 percent) was 21-25 years, followed by 26-30 and 31-35 years (22.06 percent each). Consanguity between parents was observed in 27.94 percent of the instances. The majority were primigravida 2/3 (39.71%), followed by gravida 2/3 (39.71%). (32.35 percent). There were 72,06 percent male births, 23.53 percent female babies, and 4.41 percent with ambiguous genitalia. 48.53 percent of babies were born at full term, 57.35 percent vaginally, and 39.71 percent required a caesarean section. The majority of instances were discovered in the third trimester (55.88 percent), 30.88 percent in the second trimester, and 13.24 percent after delivery. Despite the fact that 30.88 percent of patients were diagnosed in the second trimester, they did not choose abortion.

Table 1: General Characteristic.

Characteristic	No of neonates (n=136)	Percentage (%)
Maternal age (years)		
<20	10	7.35
21-25	44	32.35
26-30	30	22.06
31-35	30	22.06
>35	22	16.18
Parental Consanguinity		
Present	38	27.94
Absent	98	72.06
Order of pregnancy		
G 1	44	32.35
G 2-3	54	39.71
G 4 or more	38	27.94
Gender of baby		
Male	98	72.06
Female	32	23.53
Ambiguous	6	4.41
Pregnancy termination according to gestational age		
21-28 weeks	24	17.65
Mode of delivery		
Vaginal delivery	78	57.35
Caesarean section	54	39.71
Instrumental	4	2.94
Time of detection of anomaly.		
1st trimester	0	0.00
2nd trimester	42	30.88
3rd trimester	76	55.88
After delivery	18	13.24

Initially, all abnormalities were discovered during a USG test (86.76 percent antenatally and 13.24 percent post-natally). Only a few patients had further radiological tests, such as an MRI (antenatally 7.35 percent and post-natally 5.88 percent) and a post-natal CT scan (also 5.88 percent) (as antenatally CT is relatively contraindicated).

Table 2: Diagnostic modality

Diagnostic modality	No of cases	Percentage (%)
USG examination		
Antenatal	118	86.76
Post-natal	18	13.24
MRI examination		
Antenatal	10	7.35
Post-natal	8	5.88
CT		
Post-natal	8	5.88

The commonest inherited malformation of the urinary tract in this research was hydronephrosis (30.88%), followed by polycystic renal 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. (1.47 percent).

Table 3: Inherited malformations

CAKUT anomaly	No of cases	Percentage (%)
Hydronephrosis	42	30.88
Polycystic renal disease	28	20.59
Pelviureteric junction obstruction	26	19.12
Posterior urethral valve	24	17.65
Ambiguous genitalia	6	4.41
Epispadias	4	2.94
Duplicated collecting system	4	2.94
Hypospadias	2	1.47
Bladder exstrophy	2	1.47

Discussion

Because of the early detection and management of crucial blockages and urinary tract infections, Pre-natal determination improves the result of the affected child, preventing additional renal damage and loss of renal function. Pre-natal imaging has seen significant advancements, such as fetal magnetic resonance imaging and Doppler analyses [11]. To find predictive indicators of Pre-natal renal damage, researchers looked at the proteomics of urine and amniotic fluid. High-resolution ultrasonography clearly shows numerous anatomical anomalies, and serial estimation of the amniotic fluid index aids the physician in deciding whether or not to deliver the baby prematurely [12-14]. The use of ultrasonography, magnetic resonance imaging, and nuclear imaging in the determination and subsequent observation of patients is critical. Noninvasive urodynamic investigations, such as uroflow and postvoid residual measurement, are frequently sufficient to assess lower urinary tract function. Renal agenesis is usually unilateral and affects 1 in 1500–3200 live births, with males being more affected. Renal agenesis is one of the most severe renal tract malformations, characterized by the entire absence of renal development as well as a missing ureter. Failure of the metanephric bud to form despite a typically preceding mesonephros, early regression of the metanephros, poor development of the mesonephros, and non-development of the pronephros leading to non-growth of the

mesonephros have all been postulated as causes. Renal agenesis has also been linked to chromosomal anomalies such as trisomy 21, 22, 7, and 10, 45 X mosaicism, and microdeletion of 22q11. MCDK was the most frequently diagnosed fetal abnormality (44.44 percent), followed by Posterior urethral valves (PUV) (22.22 percent), renal agenesis (13.89 percent), Autosomal recessive polycystic renal disease (ARKD) (11.11 percent), Ureteropelvic junction (UPJ) obstruction (11.11 percent), duplicated collecting system (11.11 percent), isolated fetal pyelectasis Saryu Gupta et al. found that the average age of research participants was 29 3 years. Amniotic fluid was reduced or nonexistent in 41% (N = 13) of participating moms, while it was normal in 59 percent (N= 18). The commonest anomaly discovered was urinary blockage (29 percent), followed by multicystic dysplastic renal (MCDK) (22 percent). All mothers with anhydramnios B/L MCDK (N = 3), autosomal recessive polycystic renal disease (ARPKD) (N = 2), posterior urethral valves (PUV) (N = 2), B/L renal agenesis (N = 3), and megacystis (N = 1) had bilateral renal illness. One patient each had fusion anomalies (horseshoe renal) and rotation anomalies (malrotation). On fetal MRI, additional extrarenal abnormalities were identified in 35% (N = 11) of the cases. By greater morphological delineation, fetal MRI enhances determination accuracy in abnormalities affecting the fetal renal and genito-urinary systems. CAKUT was shown to be more prevalent in males, according to Chougule A, et al. In Pre-natal imaging, hydronephrosis was the most frequent CAKUT. 93.9 percent of all CAKUT found on a Pre-natal anomaly scan were renal collecting system anomalies, and 57.4 percent of these resolved by six months of age. The renal collecting system abnormalities accounted for 93.9 percent of all antenatally diagnosed CAKUT, which was higher than the renal parenchyma abnormalities, which accounted for 6.1 percent. Regardless of the site, post-natal resolution on day three USG was noted in seven out of 22 (31.8 percent) patients of antenatally diagnosed mild hydronephrosis. 11 (39.3%) of the 28 antenatally diagnosed hydronephrosis cases resolved at some time throughout the six-month follow-up period. Primary VUR was the most prevalent CAKUT, according to Kumar BH et al., followed by PUJO, MCDK, non-obstructive hydronephrosis, and PUV. PUV (36.4 percent), main VUR (19.6 percent), and PUJO were the commonest, according to Soliman et al (18.7 percent). PUJO was found in 62.7 percent of cases, followed by VUR in 16.6 percent, according to Aksu et al. According to Lee et almeta-analysis, .'s every dilatation of the urinary tract, regardless of degree, is associated with a 36 percent greater risk of an underlying uropathy. The risk varied depending on the severity of the hydronephrosis, ranging from 12% in cases of mild hydronephrosis (APPd > 7 mm in the 2nd trimester and 9 mm in the 3rd trimester) to 45 percent in cases of moderate hydronephrosis (APPd 7-9 mm in the 2nd trimester, 9- 15 mm in the 3rd trimester) and up to 88 percent in cases of severe hydro In the case of concomitant spinal abnormalities, MR imaging is required [15-18]. MR urography can reveal ectopic extravesical ureteric insertions, giving a comprehensive picture of the abnormality. A thorough understanding of lower urinary tract abnormalities is required for accurate determination and management.

Conclusion

CAKUT discovered hydronephrosis most frequently both antenatally and post-natally. Antenatal imaging detected renal collecting system defects better than other renal anomalies. CT and MR imaging are not ideal for broad screening, but they provide excellent anatomic detail and increased diagnostic specificity.

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