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ORIGINAL ARTICLE

Frequency of Urinary Anomalies in Perinatal Autopsies

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ABSTRACT

Background: Congenital anomalies of urinary system are common and are found in 3-4% of population, and 10% of terminations of pregnancies are attributable to lethal urinary anomalies. Although timely detection of nonlethal anomalies depends on many aspects, for instance equipment quality, sonologist experience and timing of the ultrasound examination. However, to confirm the findings and to attain definite diagnosis, a fetal autopsy after the termination of a pregnancy is of utmost important and is also crucial for genetic counseling.In view of this, the present study aimed to determine the frequency and to identify the types of the urinary anomalies in perinatal autopsies.

Methods: This study includes 391 perinatal autopsies conducted prospectively from July 2016 to June 2018 at M. R. Medical College, Basaveshwar Teaching and General Hospital, Kalaburagi. After obtaining the consent from either parent, each fetus was examined as per the predetermined protocol which included ultrasound diagnosis, photographs, external and internal examination.

Results: The gestational age ranged from 16weeks to 44weeks and the mean weight was 1123.96gm. Out of 391 cases urinary anomalies encountered in 19 cases (4.9%). Out of 19 cases, anomalies of renal parenchyma were seen in 15 cases (78.9%) and anomalies of pelvi-ureters and bladder was seen in 4 cases (21.1%). Three cases of urinary anomalies were associated with syndromes and 8 cases were associated with other system anomalies.

Conclusion: The perinatal autopsy after termination of pregnancy is essential to confirm the ultrasound findings and to make a definitive diagnosis.

Keywords | Autopsy; Congenital anomalies; Ultrasonography; Urinary anomalies.

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INTRODUCTION

In India 8-15% of perinatal deaths and 13-16% of neonatal deaths are due to congenital anomalies. However, multiple congenital anomalies in patients present a reasonably uncommon but extremely difficult task to the pediatrician.[1]

In an around 3-4% of population, congenital anomalies of urinary system are found.[2] Intermediate mesoderm along the posterior wall of the abdominal cavity helps in the development of the urinary system.[3] Although the development begins around the 4th week of post conception; it is completed by the 12th week.[4]

Ten percent of terminations are attributable to lethal urinary anomalies. However, timely detection of nonlethal anomalies depends on many aspects, for instance equipment quality, sonologist experience and timing of the ultrasound examination. In the second half of the pregnancy, normal amniotic fluid volume suggests that at least one kidney is functioning and an intact urinary channel to amniotic cavity. However, oligoamnios if present, suggest strong suspicion of urinary tract anomalies.[5]

Nevertheless ultrasonography (USG) is a valuable examination tool; to confirm the findings and to attain definite diagnosis, a fetal autopsy after the termination of a pregnancy is important. Therefore, a fetal autopsy not only helpful in identifying the cause of the fetal loss but also gives substantial surplus evidence which in practice is essential for genetic counselling.[6]

In view of the above and with limited data on autopsy studies related to urinary system, this study was undertaken to determine the frequency and to identify the types of the urinary anomalies in perinatal autopsies.

MATERIALS AND METHODS

This study includes 391 perinatal autopsies conducted prospectively from July 2016 to June 2018 at M. R. Medical College, Basaveshwar Teaching and General Hospital, Kalaburagi. After obtaining permission from Institutional Ethics Committee (Approval No: HKES/MRMCK/IEC/181021) and the consent from either parent, all fetuses with gestational age greater than 16 weeks and less than 44 weeks, birth weight greater than 350 g were included; while autolysed fetus, fetus with gestational age less than 16 weeks and greater than 44weeks and birth weight less than 350g were excluded. Each fetus was examined as per the predetermined protocol which included ultrasound diagnosis, photographs, external and internal examination.

The autopsy protocol included the removal of cervical, thoracic, abdominal and pelvic organs en block and subsequently dissected into organ blocks. The placenta, fetal membranes and umbilical cord were studied in all the cases. Histological sections were taken from lung, liver, kidney, thymus, brain, placenta and umbilical cord. In cases where the antenatal ultrasonography diagnosis was available, were compared with the postnatal autopsy findings.

RESULTS

A total of 391 perinatal autopsies were conducted with a gestational age ranging from 16weeks to 44 weeks. Majority (30.5%) of the cases were of gestational age 16-20weeks (Table 1). There was male predominance seen with male to female ratio being 1.2 : 1. Birth weight ranged from 350g to 3800g with mean weight of 1123.96±781.46gm (Table 2).

Intra uterine death (47.4%), congenital anomalies (21.0%), placental insufficiency (16.8%) and aspiration pneumonia (14.8%) were the cause of death identified in perinatal autopsies. The congenital anomalies were present in 82 cases accounting for the incidence of 21% in perinatal autopsies (Table 3). Among these, majority of the cases showed anomalies of central nervous system (8.7%).

The urinary system anomalies were found in 19 cases (4.9%), of which 15 cases were of renal parenchymal anomalies and found

Table 1: Age and Sex distribution in perinatal autopsies.

Gestational Age	Male		Female		Ambiguous Genitalia	Absent Genitalia	Total	
	Number of Cases	%	Number of Cases	%			Number of Cases	%
16-20wks	66	32.3	44	25.8	07	01	118	30.4
20-24wks	52	25.4	48	27.2	01	03	104	26.3
25-29wks	44	21.5	35	20.5	-	-	79	20.3
30-34wks	24	11.9	28	15.9	-	01	53	13.6
35-39wks	17	07.9	13	07.7	-	-	30	07.5
40wks<	02	01.0	05	02.9	-	-	07	01.9
Total	205		173		08	05	391	

Table 2: Birth weight distribution in perinatal autopsies.

Birth Weight in Grams	No of Cases	Percentage (%)
350-500	85	21.6
500-1000	119	30.5
1000-1500	77	19.5
1500-2000	44	11.4
2000-2500	40	10.3
2500-3000	20	05.2
3000<	06	01.5
Total	391	100

Table 3: Distribution of congenital anomalies in perinatal autopsies.

Type of Anomalies	No. of Cases	Percentage
Nervous system anomalies	34	8.7%
Urinary anomalies	19	4.8%
Cardiovascular anomalies	07	1.9%
Lung anomalies	09	2.4%
Gastrointestinal anomalies	08	2.0%
Musculoskeletal anomalies	05	1.2%
Total	82	21.0%

to be more common than pelvi-ureteral and bladder anomalies which attributed to 4 cases. Urinary anomalies showed male predominance with M:F ratio being 2.3: 1 along with 3 cases of absent genitalia and 2 cases of ambiguous genitalia (Table 4). The gestational age ranged from 16weeks to 34 weeks with maximum cases (47.3%)in 20-24weeks and was found to be statistically significant (p-value=0.032).

Table 4: Age distribution in cases of urinary anomalies.

Gestational Age	Renal Anomalies	Pelvi-ureteric Anomalies	Total	
			No	%
<20wks	02	03	05	26.4
20-24wks	09	-	09	47.3
25-29wks	01	01	02	10.5
30-34wks	03	-	03	15.8
Total	15	04	19	100

Table 5: Urinary anomalies in perinatal autopsies.

Congenital Anomalies of Urinary System	No of Cases
Unilateral renal agenesis	04
Bilateral renal agenesis	03
Bilateral Polycystic renal disease	02
Unilateral Multicystic dysplastic kidney	02
Bilateral Multicystic dysplastic kidney	04
Renal hypoplasia	01
Horse shoe kidney	01
Extrophy of bladder	01
Absent bladder & ureters	03
Distended bladder	03
Total	24

A total of 24 different urinary anomalies were found in the 19 cases. Five had more than one anomaly, while 14 had a single anomaly (Table 5). Five cases with more than one anomaly were Mermaid syndrome with left multicystic dysplastic kidney and right renal agenesis, right multicystic renal dysplasia with left renal agenesis and three cases of bilateral renal agenesis associated with absent bladder and ureters. Three cases were associated with syndromes, one case of

Mermaid syndrome with Potters syndrome and two cases of Mermaid syndrome (Fig 1 & 2). The 8 cases which were associated with other system anomaliesare as follows:

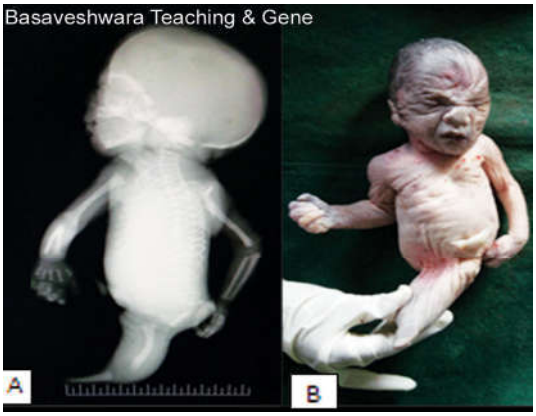

Fig. 1: Mermaid syndrome A) Radiography shows single femur bone, B) Fetus with fused lower limbs with absent feet and absent genitalia.

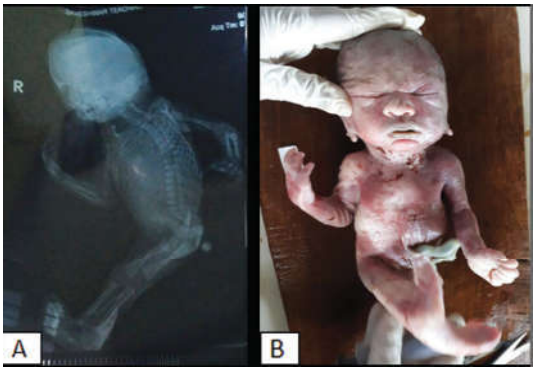

Fig. 2: Mermaid syndrome A) Radiography shows two femur bones, B) Fetus shows fused lower limbs with fused feet and absent genitalia.

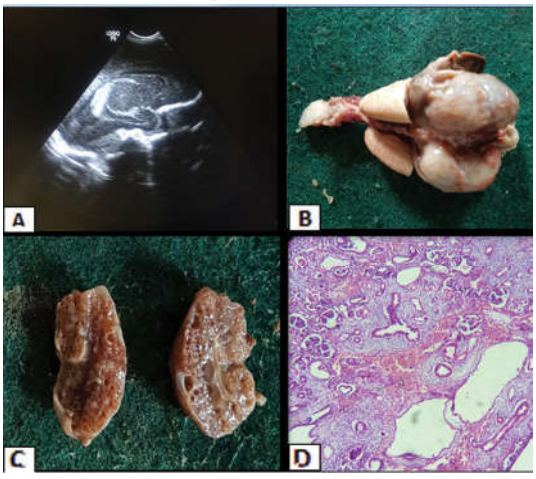

Fig. 3: Bilateral multicystic renal dysplasia. A) USG shows enlarged cystic kidneys, B) Enblock shows bilateral enlarged kidneys, C) Cut section shows multiple cystic spaces, D) H&E 10x Section shows varying sized cysts lined by flat epithelium surrounding fibrous stroma.

Fig. 4: (a) Fetus with distended abdomen, (b) Megacystis.

Fig. 5: Bilateral kidneys with distended bladder (megacystis).

- Bilateral multicystic renal dysplasia with CCAM type-2 (Fig 3).
- Megacystis with imperforate anus & single umbilical artery (Fig 4).
- Megacystis with situsin versus & dextrocardia (Fig 5).
- Renal-hepatic-pancreatic dysplasia (RHPD) with asplenia.
- Left renal agenesis with diaphragmatic hernia & CCAM type-2 with single umbilical artery.
- Mermaid syndrome with single umbilical artery.
- Left renal hypoplasia with ventriculomegaly& single umbilical artery.
- Megacystis with pentology of Cantrell, diaphragmatic hernia, omphalocele & imperforate anus.

Out of 19 cases, USG findings were available in 12 cases. In 5 cases the USG findings were confirmed by autopsy and there were no changes in the diagnosis and in

7 cases additional findings were found in the autopsy (Table 6).

Table 6: Comparison of prenatal USG findings with autopsy diagnosis.

USG Findings	No of Cases
A) USG findings were confirmed in autopsy	5
B) Change in autopsy diagnosis	7
USG findings	Autopsy diagnosis
Oligohydromnios	Bilateral renal agenesis
Oligohydromnios	Mermaid syndrome with bilateral renal agenesis
Diaphragmatic hernia	Left renal agenesis, CCAM type 2 with diaphragmatic hernia
Single live intrauterine fetus of gestational age 17weeks	Left renal hypoplasia
Oligohydromnios	Mermaid syndrome with right renal agenesis and left renal dysplasia
Oligohydromnios with bilateral renal agenesis	Mermaid syndrome with bilateral renal agenesis and potters syndrome
Single live intrauterine fetus of gestational age 23weeks	Left renal agenesis

DISCUSSION

Congenital malformations are an important cause of perinatal mortality and morbidity of great deal in childhood. Due to use of advanced diagnostic technology in particularly USG; the detection of birth defects is increasing antenatally and during the neonatal period.[7]

Multifaceted interaction of racial and ethnic factors, socio-cultural and known and unknown genetic and environmental factors, may have an impact on the prevalence pattern and the array of congenital anomalies.[8]

In the present study the incidence of congenital malformation is 21.0% which correlated with Naik et al[11] (17.4%) (Table 7).

Table 7: Congenital anomalies compared with other studies.

	Total no of autopsies	Congenital anomalies	-Percentage
(Ahuja et al[9](2013	140	58	41.4%
(Potekar et al[10](2014	54	35	54.3%
(Naik et al[11](2015	46	8	17.4%
(Kupati et al[12](2016	200	54	27%
(Present study(2018	391	82	21%

The detection of the foetal urinary anomalies incidence was also influenced by the time of examination. In the present study, thorough examination of foetuses showed incidence of 4.43% in 16-18 weeks of gestational age and this increased to 12.83% by 28-30 gestational weeks. Therefore, a single scan at 16-18 weeks of gestation could have unnoticed many of these anomalies. However, only 9% of renal abnormalities were detected by 17 weeks of gestation during a screening program held in Sweden for detecting renal foetal malformation; while 91% could be detected by 33 weeks of gestation.[6] Therefore, the present study shows the importance of timing of antenatal scan for the detection of urinary anomalies.

Table 8: Incidence of urinary anomalies compared with other studies.

Study	Incidence of Urinary System Anomalies
Gupta et al[6] (2012) n= 226	4.43%
Ahuja et al[9] (2013) n= 140	7.85%
Naik et al[11](2015) n=46	2.1%
Kupati et al[12](2016) n=200	8.5%
Present study (2018) n= 391	4.85%

The incidence of urinary system anomalies in the present study is 4.94%, which corroborated with the earlier study conducted by Gupta et al[6] which showed 4.43% (Table 8). Urinary anomalies in the male foetuses were twice reported as that in the female foetuses indicated by Cortes et al.[13] Although the incidence of the sexual dimorphism varies in different series, a similar male preponderance was reported by

Sanghvi et al.[14] and Menasinkai et al[2] which relates with the present study (M:F as 2.3:1).

As early as, in 16-18 weeks of gestation, the abnormalities in the development of the urinary system could be detected. However, foetuses in the present study showed the maximum incidence in between 20-24 weeks of gestation, which correlated with the studies conducted by Gupta et al[6], Ahuja et al[9] and Kupati et al[12] (20-24wks of gestation).

The pelvi-ureteral and the bladder anomalies were more frequent in the 16-24 weeks of gestation, while the renal anomalies were found to be more in the 24-32 weeks of gestation. The renal anomalies were more common than the pelvi-ureteral and combined renal pelvi-ureteral anomalies, which is closely correlated with Gupta et al[6] and Kupati et al.[12]

The incidence of renal agenesis is between 1/200–1/4000 births and is one of the most frequent renal abnormalities.[15] In our study, most frequent abnormality seen was renal agenesis attributing to 7 cases (41.2%) which is comparable with Gupta et al.[6] However, unilateral renal agenesis (4 cases) was found to be slightly common than bilateral renal agenesis (3 cases). Isolated unilateral kidney lesions such as agenesis, hypoplasia or dysplasia escape detection more often than bilateral lesions, probably because they will not cause amniotic fluid alterations and thus will not trigger the awareness for a renal anomaly.[16] The renal agenesis was most frequent in between 20-24weeks of gestational age and males affected more than females. The second most common anomaly was multicystic renal dysplasia. Other renal anomalies found were polycystic renal

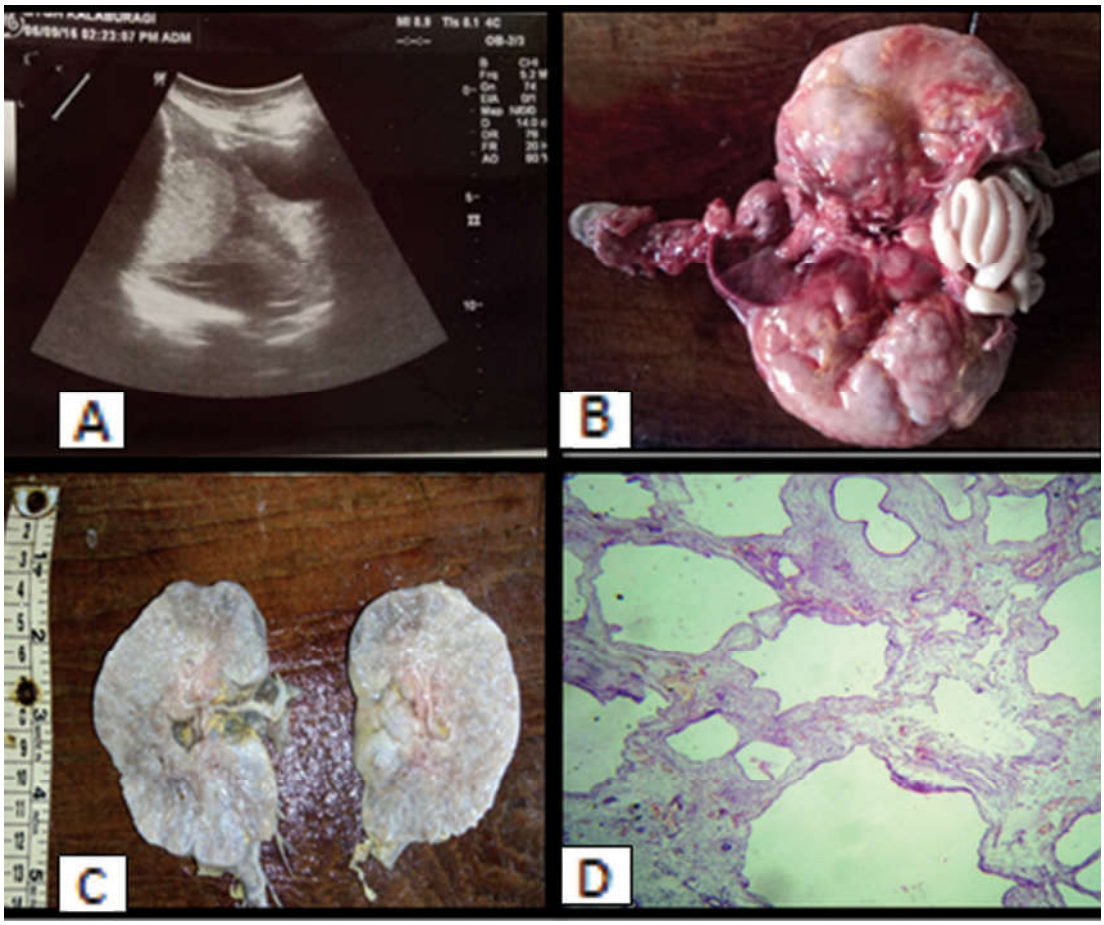

Fig. 6: Bilateral polycystic kidney disease A) USG image showing enlarged kidney, B) Enblock showing bilateral enlarged cystic kidneys, C) Cut section shows multiple cystic spaces, D) H&E 10x Section shows varying sized cysts lined by flat epithelium.

disease (Fig. 6), horse shoe kidney (Fig. 7) and hypoplastic kidney. In pelvi-ureteral bladder anomalies more common were megacystis, extrophy of bladder (Fig. 8) and absent bladder and ureters.

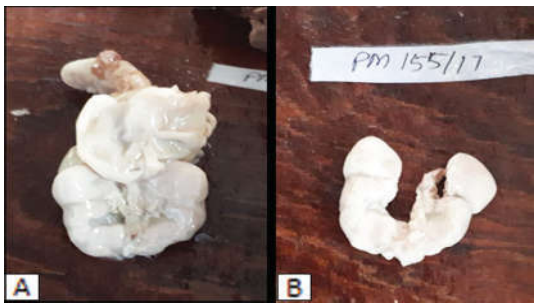

Fig. 7: A) Enblock shows fused kidneys, B) Horse shoe shaped kidney.

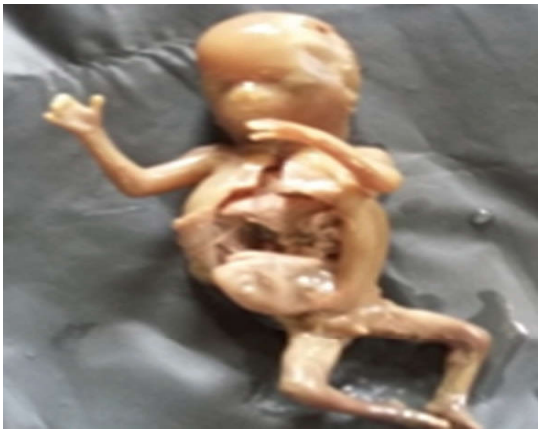

Fig. 8: Fetus with extrophy of bladder.

This study has prospectively evaluated the frequency and patterns of urinary anomalies in perinatal autopsies along with the comparison of prenatal ultrasound findings with postmortem diagnosis.

The limitation of this study is unavailability of karyotyping in our study setup to demonstrate the chromosomal aberrations which is most commonly associated with congenital anomalies and it is important not only for epidemiologic studies but also for the genetic guidance of the parents.

CONCLUSIONS

Early antenatal detection of urinary anomalies has significance, as this may help in postnatal management and will also have a bearing on the decision of the termination of the pregnancy. Although oligohydramnios usually trigger the attention towards a thorough examination of the urinary system but the presence of oligohydramnios makes the interpretation of the ultrasound findings more difficult and can therefore be responsible for inaccuracies. Thus the fetal autopsy after termination of pregnancy is essential to confirm the findings for a definitive diagnosis.

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