

Prenatal Diagnosis and Pediatric Urology

ANTENATAL DIAGNOSIS OF CONGENITAL URINARY TRACT MALFORMATIONS

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ABSTRACT

Congenital urinary tract malformations are the most common congenital malformations in children. Their severity is related to their impact on the kidneys [Macher Ma et al., 1986]. Previously, they were diagnosed when complications developed, generally necessitating intervention because of renal damage. Nowadays, asymptomatic uropathies are detected more frequently, mainly as a result of the widespread introduction of antenatal screening [Bonnin F., Lottman H., 1996]. Management now has two preventive aims: in the short term, to prevent infection, and in the long term, to prevent deterioration of the function of the affected kidney. Fetal ultrasound examination has changed the management of congenital urinary tract malformations. As early as 1958, several authors [Kerzabi K., 1986] published studies on ultrasound examination of the pregnant uterus, the fetus and fetal abnormalities. However, it was not until 1970 that Garret [Garret W. J. et al., 1970] reported the first antenatal diagnosis of a multicystic kidney and fetal ascites.

In our country, congenital urinary tract malformations represent a substantial burden: approximately three-quarters of children receiving dialysis and awaiting transplantation have sequelae of a congenital urinary tract malformation, making this a major public health issue.

Keywords: Uropathies – Congenital malformations – Diagnosis

INTRODUCTION

Why antenatal diagnosis? Failure to recognise these conditions at birth

Only 0.1% of cases are symptomatic at birth. The vast majority become apparent much later, when complications develop, such as pyelonephritis or even renal failure [Dodat H. et al., 2001].

The opportunity to make a therapeutic decision

Medical termination of pregnancy in lethal forms or those associated with severe sequelae, and early neonatal management in other cases [Bonnin F., Lottman H., 1996].

The malformation very often presents as urinary tract dilatation, raising several questions:

- What is the cause of this dilatation?
- Does it reflect simple hypotonia or a genuine obstruction to urinary flow?
- Does it affect fetal development and fetal and postnatal renal function?
- What are the implications for antenatal management?
- Can and should an in utero procedure be proposed?
- Should delivery be brought forward?
- Which postnatal investigations should be performed, and in what sequence?
- In which cases will postnatal surgery be essential?
- In which cases can spontaneous improvement and resolution be expected? [Mahan J. et al., 1982]

The surgical management of congenital urinary tract malformations has evolved in recent years. Treatment is now most often preventive, addressing both infection and the consequences of obstruction. Surgical indications have become more precise, better documented and less routine.

In our country, congenital urinary tract malformations represent a substantial burden: approximately three-quarters of children receiving dialysis and awaiting transplantation have sequelae of a congenital urinary tract malformation, making this a major public health issue.

These considerations prompted us to address this problem and develop a programme covering:

- Antenatal diagnosis of congenital urinary tract malformations in a sample of pregnant women followed at maternal and child health centres (PMI centres), together with screening and follow-up of congenital urinary tract malformations in infants.
- Antenatal diagnosis of congenital uropathies has been widely practised for only about twenty years. Therapeutic decisions require collaboration among paediatricians, surgeons, radiologists, laboratory specialists and bacteriologists. This collaboration should improve a prognosis that remains guarded, making screening a matter of genuine urgency.

RESEARCH METHODOLOGY: SCREENING SURVEY

1. Type of survey

This was a cross-sectional epidemiological survey involving active antenatal screening for congenital urinary tract malformations in a population of pregnant women in the Sétif health district during the year 2000.

2. Objectives

The objectives of the study were:

- To estimate the incidence of urinary tract malformations through antenatal diagnosis in pregnant women during the third trimester of pregnancy.
- To assess the kidneys, monitor their development, and evaluate the progression of lesions and the outcomes of management.
- To establish a protocol for antenatal screening and management of antenatally diagnosed congenital urinary tract malformations.

3. Study method

3.1. Target population

The target population comprised all pregnant women attending primary healthcare facilities in the Sétif health district during the third trimester of pregnancy as part of antenatal follow-up activities in 2010.

3.2. Study population

The study population was a representative sample of the target population, comprising 500 pregnant women in the third trimester of pregnancy.

4. Study protocol

Antenatal screening will be conducted in a representative sample of pregnant women attending PMI centres.

An ultrasound examination is performed during the third trimester, and the findings are confirmed by a second ultrasound examination 15 days to one month later. If the second examination is positive, an ultrasound examination will be performed after birth, on day 7, together with a diagnostic work-up. If the postnatal examination is negative, another ultrasound examination will be performed at one month to avoid false-negative findings.

5. Conduct of the survey

After prior authorisation had been obtained from the relevant authorities, a work plan was drawn up to launch the survey. Information and explanatory meetings were organised with the participation and coordination of the heads of the PMI centres. A working session was also held with the heads of radiology and obstetrics and gynaecology to agree on a schedule and organise radiological and ultrasound examinations without disrupting routine daily activities. To encourage active participation, a conference on congenital urinary tract malformations was held. At the end of this process, two liaison forms were developed.

Administrative arrangements: Approval from the supervisory authority was obtained following submission of an explanatory request outlining the relevance and value of this two-part survey.

Practical arrangements: Our approach involved contacting PMI centres to raise awareness among pregnant women at 37 weeks of gestation and encourage them to undergo repeated obstetric ultrasound examinations.

RESULTS

6. Results of antenatal screening

A representative sample of 500 women underwent systematic ultrasound examination during the third trimester of gestation. Detection of a urinary tract malformation will be confirmed by a second ultrasound examination 15 days to one month later. If the second examination is positive, the newborn undergoes ultrasound on day 7, followed by a complete work-up if the abnormality is confirmed.

6.1. Parental age

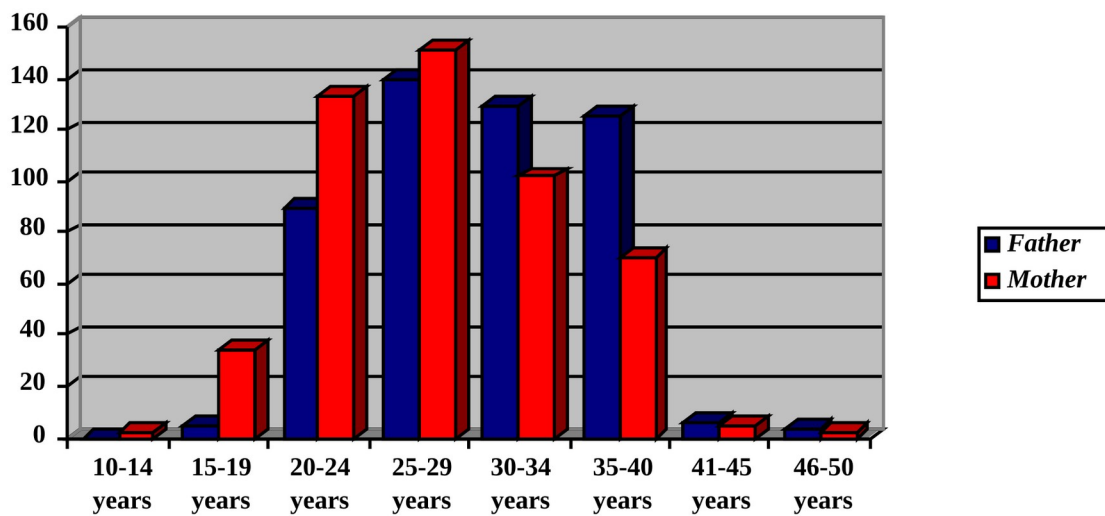


Figure 1: Parental age.

Maternal age lay within the 18–34-year range, corresponding to reproductive age:

- 2 women were aged 14 years.
- 33 women were aged 15–19 years.
- 133 women were aged 20–24 years.
- 152 women were aged 25–29 years.
- 102 women were aged 30–34 years.
- 77 women were older than 35 years.

Paternal age lay within the 25–40-year range:

- 90 men were aged 20–24 years.
- 140 men were aged 25–29 years.
- 130 men were aged 30–34 years.
- 126 men were aged 35–40 years.

6.2. Gestational age

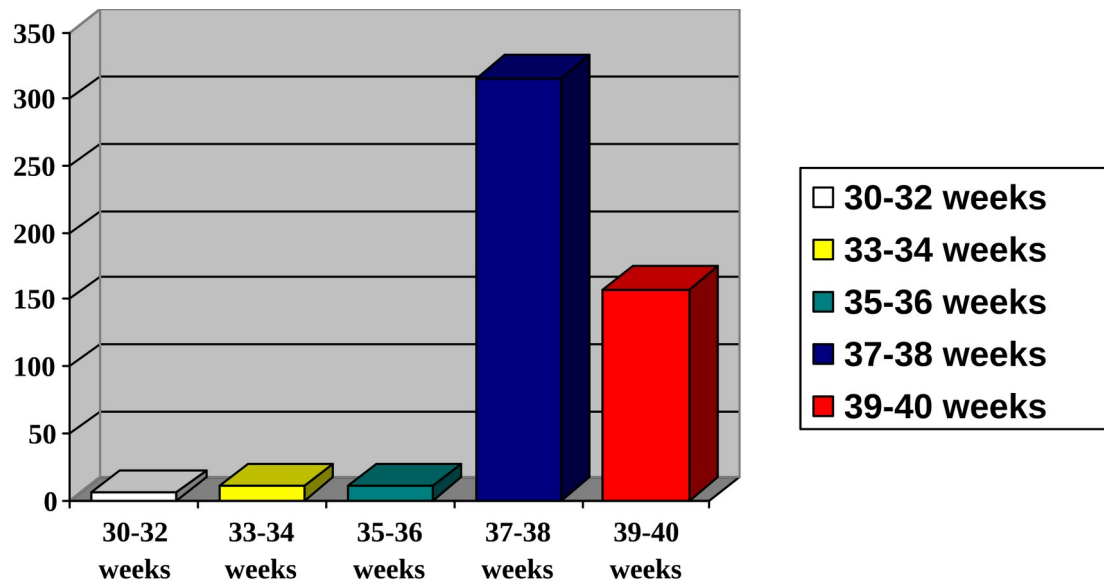


Figure 2: Gestational age.

Screening for congenital urinary tract malformations is performed during routine obstetric ultrasound examinations at the beginning of the third trimester of pregnancy, at 32 weeks of gestation.

6 ultrasound examinations were performed at 30–32 weeks of gestation.

10 ultrasound examinations were performed at 33–34 weeks of gestation.

10 ultrasound examinations were performed at 35–36 weeks of gestation.

To increase the detection rate, screening was intensified during the last month of gestation:

316 ultrasound examinations were performed at 37–38 weeks of gestation.

158 ultrasound examinations were performed at 39–40 weeks of gestation.

6.3. Gestational age and distribution of normal and abnormal ultrasound examinations during antenatal screening

Table I: Gestational age and distribution of normal and abnormal ultrasound examinations.

Gestational age (weeks)	Normal ultrasound	Abnormal ultrasound	Total examinations
30–32	5	1	6
33–34	6	4	10
35–36	5	5	10
37–38	301	15	316
39–40	144	14	158
TOTAL	461	39	500

Antenatal ultrasound detected 39 malformations from the 30th week of gestation onwards:

30–32 weeks: one urinary tract malformation.

33–34 weeks: 4 malformations, comprising 2 urinary tract and 2 neurological malformations.

35–36 weeks: 5 malformations, comprising 3 urinary tract and 2 neurological malformations.

37–38 weeks: 15 malformations, comprising 7 urinary tract and 8 neurological malformations.

39–40 weeks: 14 malformations, all neurological.

Detection of abnormalities of the urinary collecting system was best during the last month of pregnancy. Each abnormality was reassessed by a second ultrasound examination performed 15 days to one month later.

6.4. Malformations detected antenatally

Table II: Malformations detected antenatally.

Malformations	Number	Percentage	95% confidence interval
Neurological malformations	26	5.2	3.3–7.1
Urinary tract malformations	13	2.6	1.3–3.9
TOTAL	39	7.8	6.4–10.2

Thirty-nine abnormalities were detected in 500 fetuses. These comprised 26 neurological abnormalities, including 11 cases of anencephaly and 15 cases of hydrocephalus, giving an incidence of $26/500 = 5.2\%$ (95% confidence interval: 3.3%–7.1%).

Thirteen urinary tract abnormalities were detected, giving an incidence of $13/500 = 2.6\%$ (95% confidence interval: 1.3%–3.9%).

The overall incidence of the malformations encountered was $39/500 = 7.8\%$ (95% confidence interval: 6.4%–10.2%).

Among the 13 urinary tract abnormalities, 2 deaths occurred at birth, and postnatal confirmation was not possible.

One false-positive finding was recorded, with a normal postnatal ultrasound examination confirmed by repeat examination one month later.

The incidence of confirmed urinary tract malformations was $10/500 = 2\%$ (95% confidence interval: 0.8%–3.4%).

With 1,048 births per year in Sétif, approximately 21 urinary tract malformations per year can be expected.

This antenatal screening survey raised awareness among the population and obstetricians. Since then, obstetricians have referred all pregnant women whose ultrasound examination reveals a urinary tract abnormality.

A further 20 cases were detected in this way. Our study therefore covers all antenatally detected congenital urinary tract malformations, comprising 33 cases.

6.5. Urinary tract malformations detected at the first ultrasound examination

Table III: Urinary tract malformations detected at the first ultrasound examination (N = 33).

Urinary tract malformations	Number
Hydronephrosis	15
Hydroureteronephrosis	13
Multicystic kidneys	5
TOTAL	33
Amniotic fluid	
Oligohydramnios	16
Polyhydramnios	2

The first antenatal ultrasound examination detected 33 cases of congenital urinary tract malformations: hydronephrosis in 15 cases, hydroureteronephrosis in 13 cases and multicystic kidneys in 5 cases. Assessment of amniotic fluid most often revealed oligohydramnios, which was identified in 16 cases.

6.6. Progression of renal lesions on antenatal ultrasound examinations

Table IV: Progression of renal lesions on antenatal ultrasound examinations.

Progression of lesions	Number
Stable lesions	26
Worsening hydronephrosis	1
Worsening hydroureteronephrosis	3
Increasing bladder dilatation	3
Amniotic fluid	
Worsening oligohydramnios	5
Worsening polyhydramnios	1
New-onset oligohydramnios	1

Follow-up ultrasound performed 15 days to one month later showed stable findings in 26 cases, worsening hydroureteronephrosis in 3 cases, worsening hydronephrosis in 1 case and increasing bladder dilatation in 3 cases.

Amniotic fluid findings were abnormal in 18 cases and remained stable in 11. The following changes were observed:

- Worsening oligohydramnios in 5 cases.
- Worsening polyhydramnios in 1 case.

In addition, one new case of oligohydramnios was recorded.

6.7. Antenatally diagnosed hydronephrosis

Table V: Hydronephrosis (N = 15).

Unilateral involvement	11 cases
Bilateral involvement	4 cases
Oligohydramnios	10 cases
Polyhydramnios	0 cases

Antenatal ultrasound detected 15 cases of pelvicalyceal dilatation, including 11 with unilateral involvement.

Amniotic fluid was normal in 5 cases. Oligohydramnios was the most frequent finding, occurring in 10 cases.

6.8. Antenatally diagnosed hydroureteronephrosis

Table VI: Hydroureteronephrosis (N = 13).

Unilateral involvement	4 cases
Bilateral involvement	9 cases
Oligohydramnios	9 cases
Polyhydramnios	2 cases

Antenatal ultrasound detected 13 cases of ureteropelvic dilatation, including 9 with bilateral involvement. Oligohydramnios was the most frequent amniotic fluid abnormality, occurring in 9 cases. Polyhydramnios was found in 2 cases, and amniotic fluid was normal in 2 cases.

6.9. Antenatally diagnosed multicystic kidneys

Table VII: Multicystic kidneys (N = 5).

Unilateral involvement	0 cases
Bilateral involvement	5 cases
Oligohydramnios	5 cases

Five cases of multicystic kidneys were diagnosed antenatally. Bilateral involvement and oligohydramnios were present in all cases.

6.10. Agreement between antenatal and postnatal ultrasound examinations

Any urinary tract abnormality detected in utero should lead to an ultrasound examination after birth to confirm or exclude the finding. If the ultrasound examination is positive, further urological and radionuclide investigations allow more precise characterisation of the abnormalities.

Table VIII: Agreement between antenatal and postnatal ultrasound examinations.

Antenatal ultrasound	Postnatal ultrasound	Number of cases
Hydronephrosis	Same appearance	9
Hydronephrosis with contralateral renal agenesis	Same appearance	1
Hydroureteronephrosis	Same appearance	9
Hydroureteronephrosis with contralateral renal agenesis	Same appearance	1
Bilateral multicystic kidneys	Bilateral hydroureteronephrosis	3
Bilateral multicystic kidneys	Bilateral hydronephrosis	1
Bilateral multicystic kidneys	Right hydronephrosis	1
Left hydronephrosis	Left hydroureteronephrosis	2
Unilateral hydronephrosis	Bilateral hydroureteronephrosis	2
Right hydronephrosis	Right hydroureteronephrosis	1
Hydroureteronephrosis	Normal kidney	1
Bilateral hydroureteronephrosis	Death at birth	2

The antenatal diagnosis of urinary tract dilatation was confirmed postnatally in 20 cases. In 10 cases, the diagnosis was incomplete or inaccurate. The most frequent diagnostic confusion was between multicystic kidneys and hydroureteronephrosis, reflecting the severity of the uropathy. Two deaths occurred at birth, and postnatal confirmation was not possible. A single false-positive finding was recorded: a case of hydroureteronephrosis in which postnatal ultrasound was normal, as confirmed by repeat examination one month later.

Among the 31 newborns assessed postnatally, agreement between the ultrasound findings was reported in 60.6% of cases.

6.11. Definitive postnatal aetiological diagnosis

The definitive diagnosis was established within a few days to a few weeks after birth.

Table IX: Definitive aetiological diagnosis.

Definitive aetiological diagnosis (30 cases)	Number
Ureteropelvic junction obstruction	12
Megaureter	9
Vesicoureteral reflux	2
Right bladder diverticulum with hydroureteronephrosis	1
Posterior urethral valves	3
Duplex collecting system	2
Megacystis–megaureter	1

DISCUSSION

The detection of a uropathy in utero primarily raises the issues of identifying the abnormality and determining its severity. Ultrasound examination seeks to answer three questions:

- At what anatomical level is the abnormality located?
- What type of abnormality is present?
- What is the functional status of the kidneys?

Antenatal ultrasound allows visualisation of 90% of fetal kidneys between 17 and 20 weeks of gestation, and 95% by 22 weeks. The urinary tract abnormalities identified involve either the renal parenchyma itself, with dysplastic lesions of the renal cortex, or the urinary collecting system. Renal function is assessed indirectly, particularly through the characteristics of the amniotic fluid, which must be interpreted in relation to gestational age. Severe uropathies are characterised by the development of early renal dysplasia upstream of an obstructive uropathy. Conversely, dilatation of the collecting system may regress without apparent sequelae. Although antenatal ultrasound allows population screening, it does not always provide a precise diagnosis: observation of urinary tract dilatation before birth does not necessarily establish a diagnosis of true uropathy. Antenatal diagnosis nevertheless has limitations. Ultrasound diagnostic errors have included confusion between dilatation of the gastrointestinal tract and pelvicalyceal dilatation, and imprecision regarding the severity, extent and unilateral or bilateral nature of lesions of the urinary collecting system.

Published series report ultrasound diagnostic error rates ranging from 23% to 61%, with sometimes serious consequences. In a series of 38 terminations of pregnancy for major urinary tract abnormalities, Gauderer et al. [1984] reported 3 unjustified terminations. The series of Huston et al. [1990] showed that 39% of prophylactic caesarean deliveries were followed by early infant death due to respiratory or renal failure. These poor outcomes led the authors to seek a more precise assessment of fetal renal function by evaluating the ultrasound appearance of the renal parenchyma, the volume of amniotic fluid and the amount of urine produced by the fetal kidneys. Oligohydramnios or anhydramnios in the context of a uropathy is a definite marker of severity. A marked reduction in amniotic fluid volume indicates impaired renal function and may provide information about disease progression. Renal function can also be assessed through direct evaluation of renal parenchymal thickness. These two criteria seek to reflect the extent of distension of the urinary tract.

In our series, our initial objective was to involve obstetricians in antenatal diagnosis in general and in the diagnosis of congenital urinary tract malformations in particular. This was not easy, and we therefore sought the assistance of the most senior practitioners with the greatest experience in obstetric ultrasound. Our aim was to detect uropathies antenatally and, above all, to ensure early management in the immediate postnatal period.

In our series, 10 uropathies were detected and confirmed postnatally among 500 pregnant women. Urinary tract malformations ranked second after central nervous system malformations, as reported in the literature. The incidence of antenatally detected uropathies is closely related to the criteria used to define a urinary tract abnormality, namely the appearance of dilatation of the urinary collecting system. Approximately 1% of pregnancies are estimated to be affected.

Our series reports an incidence of 2% of live births, whereas the theoretical incidence is 0.9%–1% [100].

In a study of 6,292 pregnancies, Livera et al. [1989] reported that 1.46% of fetal ultrasound examinations performed at 28 weeks of gestation suggested a uropathy. After birth, only 48% of suspected cases, representing 0.71% of the pregnancies studied, had a confirmed uropathy. In 1992, Johnson et al. studied 7,530 pregnancies and identified suspected fetal uropathy in 0.74%. Only 38% of the infants had a uropathy confirmed after birth, corresponding to 0.28% of the pregnancies studied. Reported incidence varies among authors, from 0.18% in the study by Fugelseth [1994] involving 22,310 pregnancies, to 1% in the study by Mouriquant [2004] involving 1,200 pregnancies.

Our prospective study found an incidence of 2%, comparable with other studies. Antenatal diagnosis is still in its early stages in our country, and no similar screening study had previously been performed.

The probability of detecting a urinary tract abnormality depends strongly on the experience of the ultrasonographer, for whom detection of a urinary tract malformation is easier in the second half of pregnancy. In our series, detection was substantially better towards the end of pregnancy, a finding also reported by other centres [Cambell S., Pearce J. M., 1995]. This observation is important because, in an era of budget restrictions and healthcare cost control, it supports provision of more than two fetal ultrasound examinations during pregnancy. The number of false positives could not be assessed accurately. We recorded only one case among the 500 pregnancies; subsequently, however, we received a further 20 confirmed cases of uropathy referred by obstetricians who had not participated in the screening programme.

Hydronephrosis due to ureteropelvic junction abnormalities and megaureters account for most antenatally detected renal dilatations, representing 66% of cases in our series. In recent years, immediate management has become increasingly non-surgical. This specific issue is the focus of much of the current research and debate.

Our antenatal screening survey therefore detected 30 cases of congenital urinary tract malformations confirmed postnatally. Ureteropelvic junction obstruction and megaureters remained the uropathies most frequently detected antenatally.

CONCLUSION

Antenatal and postnatal ultrasound play a fundamental role in the early management of congenital urinary tract malformations. Although the diagnostic and functional assessment of uropathies does not rely exclusively on ultrasound, it remains the first-line investigation in the immediate postnatal period, guiding subsequent assessment.

Accurate perinatal diagnosis of obstructive uropathy nevertheless remains a matter of debate. Although screening has already reduced the number of neonatal urological emergencies through improved postnatal management, its value in unilateral obstructive uropathies has yet to be demonstrated, because not every antenatal dilatation signifies obstruction. The postnatal work-up should not be undertaken in haste. Surgery is rarely required during the neonatal period, and many dilatations observed in utero regress spontaneously during the first months or years of life. Regular surveillance is nevertheless necessary until the ultrasound findings have normalised.

Decisions and the organisation of antenatal and postnatal care should be based on multidisciplinary discussion involving obstetricians, radiologists, laboratory specialists, neonatologists, nephrologists and surgeons. Effective cooperation among the members of the multidisciplinary team provides the best assurance of prompt and efficient management.

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