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Fetuses and infants with congenital urinary system anomalies: correlation between prenatal ultrasound and postmortem findings

C.V. ISAKSEN*†, S.H. EIK-NES†, H.-G. BLAAS† and S.H. TORP*

**Institute of Morphology and Department of Pathology; †National Center for Fetal Medicine, Department of Obstetrics and Gynecology, Trondheim University Hospital, Norwegian University of Science and Technology, Trondheim, Norway*

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ABSTRACT

Objective *Detection of congenital urinary system anomalies is an important part of the prenatal ultrasound examination. The present study compares prenatal ultrasonographic findings and postmortem examinations of fetuses and infants with renal and urinary tract anomalies.*

Design *Criteria for inclusion were an ultrasound examination at the National Center for Fetal Medicine (Trondheim, Norway) and autopsy performed during the period 1985–94. Results from the ultrasound examination and autopsy regarding urinary system anomalies were categorized according to the degree of concordance.*

Results *Urinary system anomalies were found in 112 (27%) of 408 fetuses with congenital anomalies. The renal and/or urinary tract anomaly was the principal reason for induced abortion or cause of death in 50 cases (45%). In 97 (87%) of the 112 cases there was full agreement between the ultrasound observations and the autopsy findings. In five cases the autopsy revealed minor findings not mentioned in the ultrasound report. The main diagnosis was thus correct in 102 cases (91%). In four cases major autopsy findings had not been found by ultrasound examination; in another four, none of the autopsy findings were suspected by ultrasound, and in two, minor ultrasound findings were not confirmed at autopsy.*

Conclusions *The accordance between ultrasound diagnoses and postmortem examinations proved to be satisfactory. The close co-operation between ultrasonographers and perinatal pathologists is mutually beneficial. In addition to complementing prenatal diagnosis, postmortem examination is of vital importance for the quality control of ultrasonography in fetal diagnosis and plays an important role in genetic counseling.*

INTRODUCTION

Detection of fetal anomalies is an important part of prenatal care. The introduction of routine fetal ultrasonographic examination has improved the detection rate of fetal anomalies, while the technical developments of ultrasound equipment have increased diagnostic accuracy. As a consequence, congenital anomalies are diagnosed earlier than previously. In cases where abortion is elected, the physical and psychological trauma to which these women are exposed may be reduced[1]. The decreasing gestational age at termination of pregnancy (TOP), and thus the small size of the fetus represents a challenge in perinatal pathology which requires a high level of experience and expertise.

The overall prevalence of congenital urinary system anomalies has been reported as two per 1000 live births[2]. Urinary system abnormalities represent a large group of fetal anomalies detected by sonography[3]. Sonographic screening has disclosed an overall frequency of fetal uropathy of 0.28% and about two-thirds of these demonstrated urinary tract dilatation[4,5]. In two different antenatal studies the most frequent cause of significant urinary tract dilatation was obstruction of the ureteropelvic junction[6,7]. Upper urinary tract dilatation and renal dysplasia constitute the largest group of urinary system anomalies[2,4,8,9]. The incidence of bilateral renal agenesis is low, with an incidence of 0.1–0.3/1000 births[10–12].

Autopsy studies comparing the prenatal ultrasound diagnosis with postmortem examination have shown that the prenatal detection of renal anomalies varies from 60% to over 90%. In these studies the urinary system anomaly was part of a general analysis of all types of anomalies and did not focus specifically on particular organ systems[13–19]. A few studies comparing prenatal and postnatal urinary system anomalies have focused on both clinical follow-ups and postmortem examinations, where the detection rate

Correspondence: Dr C. V. Isaksen, Department of Pathology, Trondheim University Hospital, 7006 Trondheim, Norway

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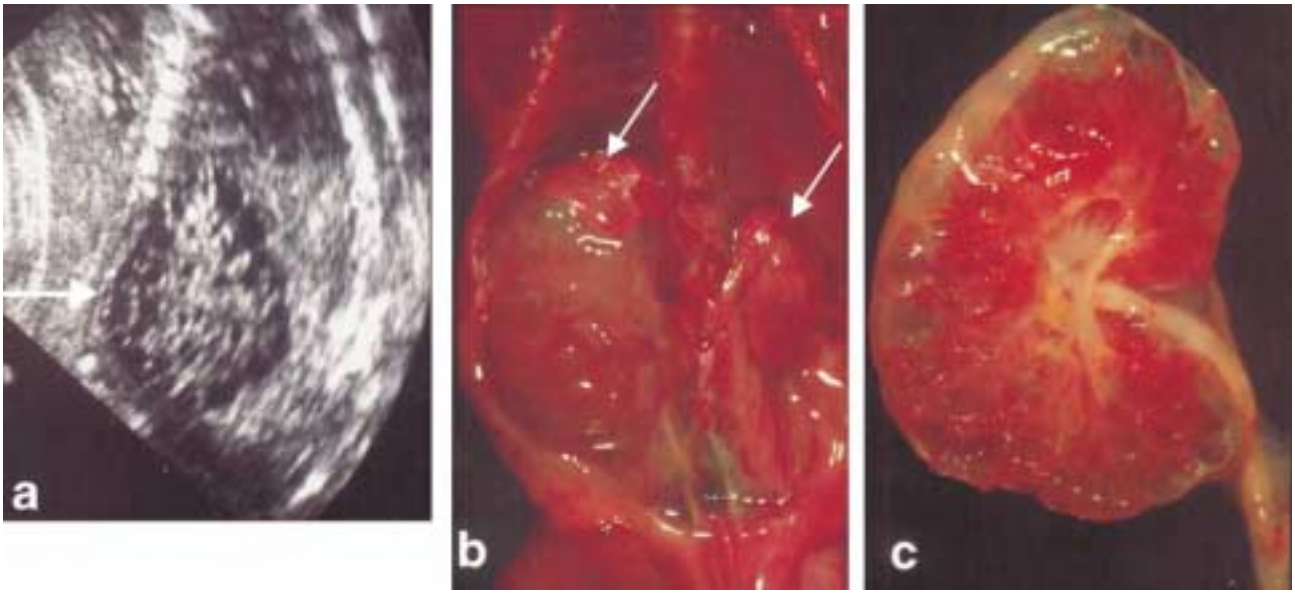

Figure 1 Eighteen-week fetus with cystic dysplastic right kidney and agenesis of the left kidney. (a) Ultrasound scan of right kidney (arrow); (b) *in situ* photograph at autopsy including right kidney and both suprarenal glands (arrows on suprarenal glands); (c) cut surface of right kidney.

has varied from 65% to 100%[4,20–22]. Except for one study[23], they all comprised a limited number of cases, and minor renal anomalies were studied in only one study[22]. To our knowledge, a comprehensive focus on prenatal diagnoses of urinary system anomalies comparing them with autopsy results has not been done.

This study was designed to prospectively evaluate the correlation between prenatal ultrasound and autopsy examination, and at the same time record urinary system anomalies in mixed low-risk and high-risk populations.

MATERIALS AND METHODS

Autopsied fetuses and infants which had been examined prenatally at the Ultrasound Laboratory, Department of Obstetrics and Gynecology, Trondheim University Hospital, and later proved to have a urinary system abnormality, are included in this study. The hospital serves the city of Trondheim (Norway) and surrounding areas with a total of 250 000 inhabitants. The National Center for Fetal Medicine was established in 1990 and is the Norwegian referral centre for pregnant women with suspected or verified fetal anomalies.

Four hundred and eight autopsies of fetuses and infants with developmental anomalies were performed between 1985 and 1994, 365 of these at the Department of Pathology, Trondheim University Hospital, and the rest at other hospitals co-operating with the centre. In 112

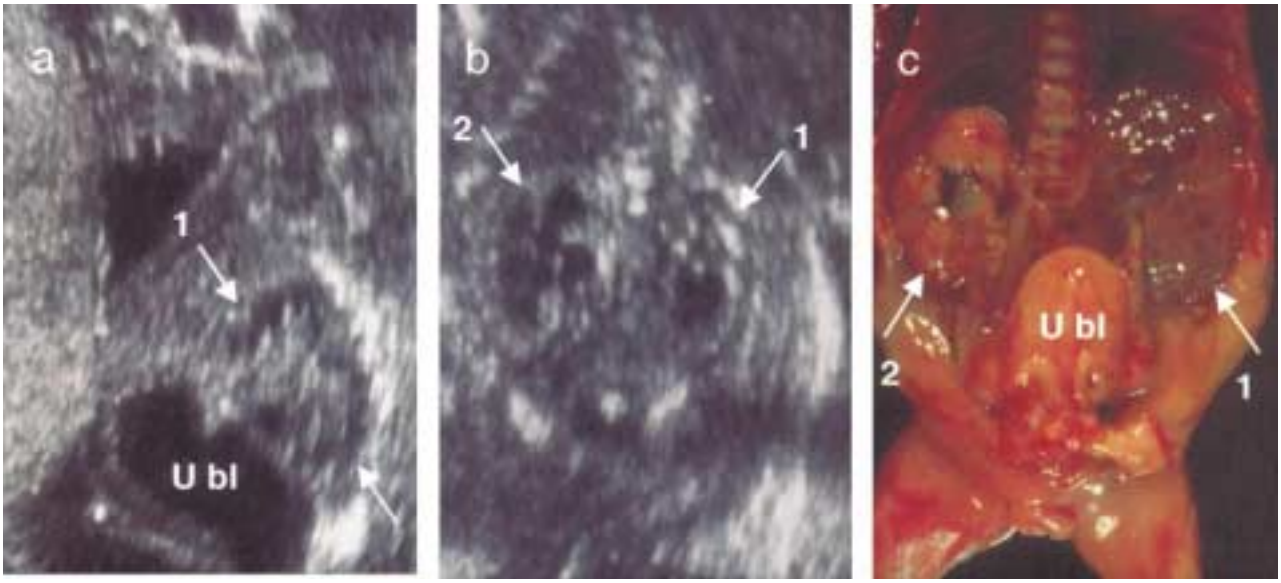

Figure 2 Twenty-week fetus with urethral obstruction, dilated urinary bladder, hydronephrosis of the right kidney and a cystic dysplastic left kidney. (a) Ultrasound scan of urinary bladder (U bl) and left kidney (arrow); (b) ultrasound scan of kidneys (arrows; 1 left, 2 right); (c) *in situ* photograph at autopsy including bladder (U bl) and both kidneys (arrows).

Table 1 Main diagnosis classified according to organ system in 112 cases with urinary system anomalies

Organ system	*Total (n)*	*Proportion of total (%)*
Urinary system	50	45
Central nervous system	20	18
Cardiovascular system	19	17
Diaphragm/abdominal wall defects	12	11
Fetal hydrops/cystic hygroma	7	6
Miscellaneous	4	3
Total	112	100

(27%) of the 408 cases, urinary system anomalies were diagnosed and included in this study.

The anomalies were suspected or diagnosed during the routine fetal ultrasound examination offered to all pregnant women at 18 weeks, or at a selective ultrasound scan performed because of hereditary risk factors or an abnormally developing pregnancy. A thorough description of the ultrasound findings was recorded, including information about supplementary examinations. All data were prospectively stored in a computer database and the comparisons were based on the recorded findings in the ultrasound and autopsy reports. The ultrasound examinations were performed by obstetricians working at the centre using Hitachi EUB 565 (Tokyo, Japan), Dornier AI 3200 (Germering, Germany) and Vingmed Sound CFM 750 (Horten, Norway) machines. The machines were equipped with transducers with frequencies ranging from 3.5 to 7.5 MHz.

At the Department of Pathology, Trondheim University Hospital, a standardized autopsy was carried out. From 1985 to 1989 the autopsies were performed by doctors in training, and from 1990 by a consultant pathologist with experience in perinatal pathology who became part of the team at the National Center for Fetal Medicine. The ultrasound report was available for the pathologist prior to the autopsy. Postnatal radiography of the fetus or infant and photographic documentation of dysmorphic features and anomalies were regularly performed after 1990. At autopsy, all organs were examined. In some cases where special photographic documentation was desirable, the kidneys were left *in situ* after removal of other organs (Figures 1 and 2). When the autopsies were performed at other hospitals, permission to use the report was obtained.

The ultrasound and autopsy findings were categorized as follows[24,25].

1. Full agreement between the ultrasound and autopsy findings.
2. Minor autopsy findings not found or not recorded at the ultrasound examination.
3. Major autopsy findings not detected at the ultrasound examination, although other ultrasound findings indicated termination of pregnancy.
4. No autopsy findings suspected at the ultrasound examination. In these cases the fetus or infant deceased naturally *in utero* or shortly after birth.
5. Minor ultrasound findings not confirmed at autopsy. This category includes findings supplementary to other detected anomalies which were confirmed at autopsy.
6. Major ultrasound findings not confirmed at autopsy. In addition to false positives this category includes cases where postmortem changes interfered with making a proper morphological diagnosis.

Table 2 Cases including urinary system anomalies: main diagnoses and additional findings ($n = 112$)

	Main finding (n)	*Additional finding (n)*	*Total (n)*
Urinary system	50	62	112
Cardiovascular system	19	14	33
Central nervous system	20	4	24
Gastro-intestinal system	2	21	23
Skeletal system	2	21	23
Diaphragm/abdominal wall defects	12	7	19
Fetal hydrops	7	6	13
Genital system		9	9
Lungs		1	1
Total	112		

RESULTS

Eighty-eight (79%) of the 112 cases were referred from all over the country, the rest came from the city of Trondheim. The sex distribution was 47% female and 53% male. The mean age of the mothers at the time of abortion or birth was 28 years (range 18–42). A previous pregnancy loss had been experienced by 32% of the women. The average gestational age of the fetus or infant was 28 weeks in 1985 compared with 25 weeks in 1994 (range 16–40). In 75 (67%) of the 112 cases TOP was carried out, two (2%) were spontaneous abortions, 11 (10%) were intrauterine deaths and 24 (21%) were liveborn.

A urinary system anomaly was the main diagnosis in 50 of the 112 cases (Table 1). The renal or urinary tract anomaly was isolated in 38 (76%) of the 50 cases. Thirty-five were either renal agenesis or various forms of cystic renal disease, two cases were intrauterine fetal deaths with urethral obstruction and dilated bladder, and one case was a fetus with hydroureter and hydronephrosis. The latter was spontaneously aborted after drainage for polyhydramnios. In the other 12 cases where the main diagnosis of urinary system anomaly was associated with abnormalities of other organs, anal atresia was the most frequent associated anomaly (five of 12 cases).

In 62 cases, the renal or urinary tract anomaly was considered less important than an anomaly in another organ system (Table 2). Considering all organ manifestations associated with urinary system anomalies, congenital heart defects were the most common.

More than one urinary system anomaly was registered in 49 (44%) of the 112 cases. The total number of urinary system anomalies found was 171, of these 44 (26%) were multicystic renal dysplasias, 18 of which were associated with ureteral hypoplasia, posterior urethral valves or

Table 3 Classification of urinary system anomalies ($n = 112$)

Diagnosis	*Number* (*n*)	*Proportion* (%)
Renal agenesis		
Bilateral	11	6.4
Unilateral	17	9.9
Renal hypoplasia	2	1.2
Duplication of renal pelvis and ureter	2	1.2
Horseshoe kidney	15	8.8
Multicystic renal dysplasia		
Bilateral	28	16.4
Unilateral	16	9.3
Multicystic renal dysplasia associated with		
Ureteral hypoplasia	10	5.8
Posterior urethral valves	1	0.6
Urethral atresia	7	4.1
Congenital hydronephrosis		
Bilateral	5	2.9
Unilateral	9	5.2
Ureteropelvic obstruction	9	5.2
Anomaly of the ureter		
Duplication	1	0.6
Dilatation	2	1.2
Atresia	3	1.8
Polycystic disease	6	3.5
Meckel–Gruber syndrome	2	1.2
Renal disease uncertain classification	1	0.6
Medullary cystic disorders	2	1.2
Anomalies of the bladder		
Dilatation	6	3.5
Agenesis	9	5.2
Duplication	2	1.2
Anomalies of the urethra		
Posterior valves	2	1.2
Atresia	3	1.8
Total	171	100

urethral atresia. In 15 (13%) of 112 cases a horseshoe kidney was found. In 12 of these 15 cases the karyotype was known; nine were associated with a chromosomal aberration (three Turner syndrome, six trisomy 18). The classification of the urinary system anomalies is shown in Table 3.

In 97 (87%) of the 112 cases there was full agreement between the ultrasound observations and the autopsy findings (Table 4; category 1). Discrepancies between the sonographic observations and the autopsy findings were found in the remaining 15 cases. Different degrees of accordance were registered and are listed in Table 5, cases 1–7 from the period 1985–89 and cases 8–15 from the period 1990–94.

In five cases the autopsy revealed minor findings not mentioned in the ultrasound report (Table 5; category 2) and in four cases major urinary system anomalies registered at autopsy were not mentioned in the ultrasound report (Table 5; category 3). Four other fetuses and infants had autopsy findings not observed at the routine ultrasound examination (Table 5; category 4). These died *in utero* between 23 and 38 weeks' gestation.

Two cases were classified in category 5; dysplastic kidneys were suspected at the ultrasound examination in case 7, but autopsy revealed bilateral renal agenesis. A horseshoe kidney in case 15 was interpreted on ultrasound as agenesis of the right kidney with a dysplastic left kidney. In this case other anomalies led to TOP. None of the 15 horseshoe kidneys was diagnosed at the ultrasound examination; they are listed separately in Table 6.

The main diagnosis was correct in 102 cases (91%). This includes cases with full agreement (category 1) and cases with minor autopsy findings not detected prenatally (category 2) see Table 4. If we compare the two time periods 1985–89 and 1990–94, the overall accordance between ultrasound and autopsy diagnoses was approximately the same during the two periods; 85% and 88%, respectively. As for the types of anomalies not detected, there seem to be no major differences.

Of the 112 cases with urinary system anomalies, an amniocentesis or fetal blood sampling for chromosomal analysis was performed in 82 cases. Karyotyping was not successful in seven of them due to cell culture problems. A chromosomal abnormality was detected in 28 (37%) of the

Table 4 Correlation of prenatal ultrasound findings and autopsy; renal and urinary system anomalies ($n = 112$)

	1985–1989		*1990–1994*		*1985–1994*	
Category	(*n*)	(%)	(*n*)	(%)	(*n*)	(%)
(1) Full agreement	39	85	58	88	97	87
(2) Minor autopsy findings not found by ultrasound	3	7	2	3	5	4
(3) Major autopsy findings not found by ultrasound	2	4	2	3	4	3.5
(4) None of the autopsy findings suspected by ultrasound	1	2	3	4	4	3.5
(5) Minor ultrasound findings not confirmed at autopsy	1	2	1	2	2	2
(6) Major ultrasound findings not confirmed at autopsy	0	0	0	0	0	0
Total	46	100	66	100	112	100

Table 5 Kidney and urinary tract anomalies: survey of discrepancies between ultrasound and autopsy findings ($n = 15$)

Case	*Prenatal diagnosis*	*Sex*	*GA weeks*	*Mode of death/birth*	*Final diagnosis following autopsy*	*Category*
1	Omphalocele, skeletal deformities in spinal column and lower extremities	F	33	TOP	LBWC with hypoplastic right kidney, aplasia of right ovary and fallopian tube, skeletal deformities in spinal column, pelvis and lower limbs	2
2	Right kidney and urinary bladder not localized, possibly a kidney on the left side; lung hypoplasia and Potter's syndrome	M	36	TOP (LB)	Multicystic renal dysplasia with hypoplasia (0.8 g) of right kidney (birth induced on suspicion of Potter type II, lived 2 h)	2
3	Myelomeningocele, omphalocele, skeletal deformities in spinal column and amelia one lower extremity, ARS?	F	37	IUFD (wanted to continue pregnancy)	LBWC, anal atresia, spinal deformities, amelia right lower extremity, urethral atresia with dilated bladder, hydroureter and hydronephrosis in a horseshoe kidney	2
4	Hydrocephaly and oligohydramnios	F	40	TOP	Hydrocephaly, right renal agenesis	3
5	Hydrocephaly	M	22	TOP	Hydrocephaly, bilateral radial aplasia, left renal agenesis, two ureters right kidney	3
6	None	M	29	IUFD	IUGR, skeletal deformities upper limbs, right renal agenesis	4
7	Oligohydramnios and suspected dysplastic kidneys	F	18	TOP	Bilateral renal agenesis	5
8	Polyhydramnios	M	24	LB	Bilateral hydronephrosis/hydroureter; polyhydramnios, spontaneous birth after amniotic fluid drainage, hyaline membrane disease	2
9	Polyhydramnios, hypoplastic right ventricle, VSD, oligodactyly right foot	F	33	LB	Trisomy 18, hypoplastic right ventricle, VSD, esophageal atresia and tracheo-esophageal fistula, anal atresia, ureteropelvic junction atresia, syndactyly right foot (Cesarean section, lived 1 day)	2
10	Holoprosencephaly, abdominal wall defect, dilated rectum with anal atresia and fistula	M	18	TOP	Holoprosencephaly, cleft lip/palate, cystic renal dysplasia, urethral atresia, anal atresia with fistula to bladder	3
11	Holoprosencephaly, lumbosacral meningocele, dysplastic right kidney	F	22	TOP	Triploidy, IUGR, holoprosencephaly, lumbosacral meningocele, omphalocele, VSD, left renal agenesis, ureteropelvic atresia right side with cystic renal dysplasia, syndactyly left hand	3
12	None	M	23	IUFD	Urethral atresia with megacystis	4
13	None	M	38	IUFD	Dysplastic and hypoplastic left kidney, incomplete duplication of bladder, IUGR	4
14	None	F	36	IUFD	Posterior urethral valves with dilated bladder; intrauterine asphyxia (placental infarction and retroplacental hemorrhage)	4
15	Trisomy 18, Arnold-Chiari malformation, VSD, clenched fingers, bilateral clubfeet, right renal agenesis and dysplastic left kidney	M	29	TOP	Trisomy 18, IUGR, Arnold-Chiari malformation, VSD, clenched fingers, bilateral clubfeet, horseshoe kidney	5

ARS = amnion rupture sequence; IUFD = intrauterine fetal death; IUGR = intrauterine growth restriction; LB = liveborn; LBWC = limb-body-wall-complex; TOP = termination of pregnancy; VSD = ventricular septal defect.

Table 6 Urinary system anomalies: survey of cases with horseshoe kidneys ($n = 15$)

Case	*Prenatal diagnosis*	*Sex*	*GA weeks*	*Mode of death/birth*	*Final diagnosis following autopsy*
1	Polyhydramnios, omphalocele	M	40	LB	Trisomy 18?, IUGR, omphalocele, VSD, aortic coarctation, clubfeet, horseshoe kidney
2	Omphalocele, hypoplastic right ventricle with tricuspid atresia, VSD, choroid plexus cysts, low set ears, rockerbottom feet	M	18	TOP	Trisomy 18?, omphalocele, hypoplastic right ventricle with tricuspid atresia, VSD, low set ears, horseshoe kidney
3	Polyhydramnios, IUGR	F	30	LB	Trisomy 18?, IUGR, VSD, dysmorphic facial features, malrotation of gut, horseshoe kidney
4	Anhydramnios, cystic hygroma	F	21	IUFD	Turner syndrome (mosaic) cystic hygroma, horseshoe kidney
5	Trisomy 18, Arnold-Chiari malformation, cystic hygroma, fetal hydrops, clenched fingers	F	19	TOP	Trisomy 18, Arnold-Chiari malformation, cystic hygroma, fetal hydrops, clubfeet, syndactyly, horseshoe kidney
6	Trisomy 18, choroid plexus cysts, omphalocele, bilateral radial aplasia	M	20	TOP	Trisomy 18, omphalocele, VSD, bilateral radial aplasia, horseshoe kidney
7	Fetal hydrops	F	28		Fetal hydrops, horseshoe kidney
8	Myelomeningocele, omphalocele, skeletal deformities in spinal column and amelia one lower extremity, ARS?	F	37	IUFD	LBWC, anal atresia, spinal deformities, amelia right lower extremity, urethral atresia with dilated bladder, hydroureter and hydronephrosis in a horseshoe kidney
9	Trisomy 18, omphalocele, VSD, deformities right hand	F	24	TOP	Trisomy 18, omphalocele, VSD, dysmorphic facial features, deformities right hand, horseshoe kidney
10	Trisomy 18, Arnold-Chiari malformation, VSD, clenched fingers, bilateral clubfeet, right renal agenesis and dysplastic left kidney	M	29	TOP	Trisomy 18, IUGR, Arnold-Chiari malformation, VSD, clenched fingers, clubfeet, horseshoe kidney
11	LBWC with deformities in spinal column and lower extremities	M	34	IUFD	LBWC with extensive skeletal deformities, ASD, horseshoe kidney
12	IUGR, cystic hygroma and fetal hydrops	F	21	IUFD	Turner syndrome, IUGR, cystic hygroma, hydrops, ASD, horseshoe kidney
13	IUGR, VSD, dysmorphic features, clenched finger, rockerbottom feet	F	36	LB	Trisomy 18, IUGR, VSD, dysmorphic facial features, clenched fingers, rockerbottom feet, Meckel's diverticula, horseshoe kidney
14	Trisomy 18, IUGR, hypoplastic left ventricle, VSD, clenched fingers	F	41	IUFD	Trisomy 18, IUGR, hypoplastic left ventricle, VSD, aortic coarctation, CNS dysplasia, clenched fingers, horseshoe kidney
15	Fetal hydrops, cystic hygroma	F	20	TOP	Turner syndrome, cystic hygroma, fetal hydrops, horseshoe kidney

ARS = aminion rupture sequence; ASD = atrioseptal defect; CNS = central nervous system; IUFD = intrauterine fetal death; IUGR = intrauterine growth restriction; LB = liveborn; LBWC = limb-body-wall-complex; TOP = termination of pregnancy; VSD = ventricular septal defect.

Table 7 Frequency of chromosomal abnormalities

Years	Cases karyotyped (*n*)	Normal (*n*)	Normal (%)	Abnormal (*n*)	Abnormal (%)
1985–89	23	15	65	8	35
1990–94	52	32	62	20	38
1985–94	75	47	63	28	37

75 cases that were successfully karyotyped (Table 7). The karyotype was known in 23 (50%) of all 46 cases and 52 (79%) of all 66 cases during the earlier and later time periods, respectively.

DISCUSSION

The aim of this study was to register renal and urinary tract anomalies and to compare the findings of ultrasonographic examination with the postmortem findings in 112 fetuses and infants with urinary system anomalies. The frequent occurrence of serious urinary system anomalies such as multicystic renal dysplasia reflects that this is a selected population in which almost 80% of patients were referred from elsewhere for examination. The possibility of bias will always be present. Some anomalies may be missed when other more important anomalies are sufficient for further management, or the finding of one anomaly may trigger the attention to look for other anomalies. This may influence the diagnosis of subtle lesions.

In our study, full agreement between the ultrasound examination and the autopsy report was found in 87% of cases. In 91% the main diagnosis was correct (Table 4). Other follow-up studies of ultrasonography of urinary system anomalies include, for the most part, a combination of clinical and postmortem cases. Postnatal confirmation of the prenatal diagnoses is described in 50% to 78% of cases from the late 1980s[20,21,23]. In a recent study of 55 cases, there was an agreement of 81% between antenatal and postnatal diagnoses[4]. The major discrepancies consisted of difficulties in discriminating dysplastic kidneys from hydronephrosis. In an early second-trimester sonographic screening, 21 anomalies consisting of unilateral renal agenesis, pelvic kidney and double collecting system were all confirmed postnatally or at postmortem examination[22].

Oligohydramnios or anhydramnios will usually trigger the attention towards a thorough examination of the urinary system. The presence of oligohydramnios or anhydramnios makes the interpretation of the ultrasound findings more difficult and can therefore be responsible for inaccuracies[11,21,26].

In our series, a better agreement was found between the prenatal and postnatal diagnoses in cases with central nervous system (CNS) anomalies[24] than with urinary tract anomalies. The opposite was true for congenital heart defects; in this study the correlation between the two methods was better for urinary system anomalies than for congenital heart defects[25]. This has also been found by others[16,27]. Unlike small ventricular septal defects and aortic coarctations which are difficult or impossible to detect ultrasonographically, even moderate obstructions of the urinary tract can be detected as they may lead to dilatation of the renal pelvis. While early obstruction is more likely to cause renal dysplasia, late obstruction may lead to hydronephrosis, which is therefore more likely to be observed at a later stage in pregnancy[28,29].

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. Focal dysplastic lesions, especially if subtle, can also be difficult to detect. The importance of detecting minor renal abnormalities has been emphasized, and of these unilateral agenesis is the most common[22].

Bilateral dysplastic lesions do not pose any great diagnostic problems whereas bilateral renal agenesis can be difficult to discern both because of reduced amniotic fluid and also the adrenals in these cases may mimic kidneys[2,30]. Later in pregnancy, the kidneys become more hypoechogenic which makes them even more difficult to differentiate from the adrenal glands[22,30]. In one case from the first time period, bilateral renal agenesis was falsely interpreted on ultrasound as dysplastic kidneys. This did not have any consequences for management.

The level and severity of urinary tract obstruction and the time of its onset influences the morphologic changes[29,31,32]. Of five discrepant cases with urinary tract obstruction, two had hydronephrosis and hydroureter, one had ureteropelvic junction atresia, and two had obstructive lesions of the urethra (Table 5). In the above mentioned cases, the fetus or infant was either live born or died *in utero*, but the obstructive lesions were not considered as the cause of death. These urinary tract obstructions may have occurred later in pregnancy thus accounting for the diagnosis being missed at the 18th week routine ultrasound examination.

Horseshoe kidneys are difficult to detect because the connection may be missed on the two-dimensional plane[2]. They occur in one in 600 individuals and represent one of the more frequent renal anomalies[2]. The horseshoe kidneys in our material were all associated with other anomalies; none of them was detected by ultrasound examination. During the later years the ability to diagnose horseshoe kidneys has improved considerably.

The percentage of overlooked minor ultrasound findings was reduced from the first (1985–89) to the second (1990–94) time period (Table 4). The numbers are small, but we believe this is an expression of improved ultrasound expertise. The downward shift in gestational age at abortion or birth from 28 weeks during the first year of registration to 25 weeks during the last year acknowledges both the introduction of the routine ultrasound examination at 17–18 weeks' gestation and the technical improvements of ultrasound equipment[16].

In our study, uni- or bilateral multicystic renal dysplasia was the most common anomaly and occurred in 39% of

the fetuses. Except for one intrauterine fetal death with unilateral dysplastic and hypoplastic kidney and one fetus with holoprosencephaly and cystic renal dysplasia, the renal dysplasias were either suspected or correctly diagnosed prenatally, giving a detection rate of 95%. Ureteropelvic obstruction and duplication anomalies were less common, which was expected considering the examinations were postmortem. The detection rate for hydronephrosis and ureteropelvic obstruction was 87%. Obstructive lesions were most often found in connection with other more serious anomalies.

Central nervous system anomalies and congenital heart defects were the most frequently associated conditions, followed by gastro-intestinal anomalies, diaphragmatic hernia and abdominal wall defects. When the urinary system anomaly was associated with a CNS anomaly, the latter was usually the reason for TOP or cause of death. Multiple organ anomalies with combinations of renal anomalies, CNS anomalies and congenital heart defects have previously been described[15–17,31,33–35]. An abnormal karyotype is often present in such cases and was demonstrated in 25% of all the urinary system anomalies in this study. According to other authors, between 2% and 33% of renal anomalies are associated with chromosomal aberrations[11,36] and approximately 50% are associated with other anomalies[10,11,37]. The combination of urinary system anomalies and anal atresia is understandable considering that during embryonic development the distal urinary tract and the distal gut have a common origin in the cloaca[38,39].

At autopsy, renal anomalies, if not subtle like some medullary cystic disorders, are usually easy to diagnose. Distinguishing between polycystic disease of the kidneys and multicystic renal dysplasia may be difficult on gross examination. The examination by light microscopy usually renders the correct diagnosis. Dilatations of the urinary tract are easy to discern, though it may be difficult to determine the level of obstruction. The diagnosis of urethral obstruction, specifically posterior urethral valves, may be missed at gross inspection, even with careful dissection.

Apart from complementing prenatal diagnosis, postmortem examination is considered important for the quality control of ultrasonography in fetal diagnosis. Considering the variety of etiologic factors governing renal cystic lesions, the morphologic diagnosis classifying them into non-hereditary and hereditary forms is important not only for epidemiologic studies but also for the genetic guidance of the parents[40].

CONCLUSION

In our project consisting of 408 fetuses and infants with developmental anomalies, renal and urinary tract anomalies occurred with a frequency of 27%, next after CNS anomalies (34%). The prenatal ultrasound findings from this tertiary centre were compared with the results of postmortem examination. The main diagnosis in the 112 cases with urinary system anomalies was correct in 91%, thus showing a good correlation between the prenatal and postmortem examination. This comparison of ultrasound and autopsy diagnoses does not differ greatly from the results observed in cases with CNS anomalies and congenital heart defects[24,25].

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