Original article

Value of autopsy in renal malformations: comparison of clinical diagnosis and post-mortem examination

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Abstract. *Background and aim:* This 6-year retrospective study reports 14 cases of renal malformations, observed in foetuses and newborns. The objective was to evaluate the usefulness of post-mortem examination in cases of renal malformations by a comparison of the post-mortem findings with clinical diagnosis. *Methods:* This study included laboratory tests, ultrasonography, karyotype and detailed pathological evaluation of the foetuses and newborns by external, macroscopic, microscopic and placental examination. *Results:* The results of post-mortem examination were of paramount importance: they disclosed renal malformations escaped on prenatal studies (42,86%), provided extensive additional informations (50%), or confirmed the diagnosis hypothesis (7,14%). *Conclusions:* Thus, this study confirmed the need of pathological examination for foetuses and newborns, after medical abortion and neonatal death. The pathologist's contribution to the multidisciplinary management of prenatal or postnatal abnormalities is important in particular for further genetic counseling. (www.actabiomedica.it)

Key words: amniotic fluid, renal adysplasia, renal agenesis, cystic dysplasia, oligohydramnios, renal tubular dysgenesis

Introduction

A post-mortem examination in foetuses and newborns may be made for two purposes.

First, to obtain as much informations as possible to determine various factors responsible of foetal and infant death.

Second to confirm prenatal malformations after medical abortion.

The systematic utilization of obstetric ultrasound in pregnancy has resulted in early and more frequent detection of malformations.

However, accurate echographic diagnosis of malformations is correlated with various factors, including experience of operator, the resolution of echographyst, foetal position during ultrasound examination, the thickness of maternal abdominal wall and type of malformation.

Regarding malformations of kidney and urinary tract, prenatal diagnosis is yet more difficult because they are associated with absence of amniotic fluid or reduction (oligohydramnios), which make foetal slight distinct image and doubtful examination of organs.

The objective of this investigation was to evaluate the usefulness of autopsy in a series of renal malformations by a comparison of clinical diagnosis with post-mortem findings.

Methods

The foetal and neonatal autopsy files from our institution were reviewed for 6-years period

We selected all cases of renal malformations.

We checked the clinical summaries and recorded the number that included the clinical impression of renal malformations.

Clinical data were also reviewed to determine:

1) the date of last menstrual period for assessment of length of gestation
2) time and mode of delivery
3) Exposition to teratogenic agents
4) Abnormalities observed on ultrasound examination
5) Karyotype of the foetuses and newborns.

For cases of livebirths, we also evaluated conditions at birth, appearance, clinical estimation of maturity, together with a brief postnatal history comprising problems and procedures undertaken.

Post-mortem examination was performed according the necropsy technique suggested by Langley (1).

Thus, body weight and external measurement, including crown-rump, crown-heel, foot lengths, head, thorax and abdominal circumferences were estimated and compared with standard values for assessment of foetal growth.

External dysmorphic features were evaluated prior of the evisceration and to alert to the range of internal malformations.

The brain is removed and weighted. It was examined evaluating cranial and ventral aspect, the sizes of cerebellum hemispheres. The gyral pattern was compared with normal for gestational age.

Then serial coronal sections trough the cerebrum were observed to reveal a normally positioned, normal-sized ventricular system and the presence of lesions in basal ganglia, pons and medulla and cerebellar hemispheres.

A whole-body radiograph was mandatory when skeletal disorders are suspected.

On internal examination the location and shape of every organ were evaluated.

The heart was removed after an internal inspection to evaluate morphology, sequence of cardiac chambers, the origin of greater arteries and pulmonary and systemic outflow and moreover to note the presence of septal defects.

When lower urinary tract obstruction were suspected, the whole urinary tract was removed in continuity, with adrenal glands, kidneys and genital tract.

Histological examination was performed for all organs, after fixation in 10% buffered formalin solution and embedding in paraffin. The sections were stained with hematoxylin-eosin.

Furthermore, to detect anomalies of differentiation or quantitative alterations of microscopic components in every organ immunohistochemical analysis was been essential.

Macroscopic and microscopic examination of the placenta were part of necropsy examination in all cases of foetalis malformations to discover the presence of other diseases which were associated with renal malformations such as infections, chromosome abnormalities, alterations of foetal-placental circulation and alterations of the number of cord vessels.

Results

Clinical data

Clinical data are summarized in Table 1.

Out of 308 autopsies performed during 6-year period, from January 1995 to December 2001, 14 cases were characterized by renal malformations (4,5%) (Graphic 1).

The range of maternal age was 25-42 years.

Two cases were newborns (table 1: case 5 and 10).

The first newborn was delivered at the age of 33 weeks by cesarean section when ultrasound revealed oligohydramnios and intestinal perforation.

During its postnatal life was observed anuria and he died 21 days after birth.

The second newborn was delivered at the age of 38 weeks and he died few minutes after the birth for intrapartum asphyxia. In this case during intrauterine life there is an history of oligohydramnios and the exposure to non steroidal antinflammatory agents.

The twelve remaining cases of our series were foetuses, with gestational age ranging from 15 to 23 weeks.

One case of them (case 3) was spontaneous intrauterine death, with premature rupture membranes, absence of amniotic fluid and foetal morphology not estimated by ultrasound.

Other eleven cases of this study were medical abortions performed for:

1) presence of chromosomal aberrations, detect-

Table 1. Renal malformations. Clinical data

Case	Year	Foetus Newborn	Sex	Age (wks or days)	Maternal age	Clinical data
1	1995	foetus	M	22th wks	30 years	MA. Trisomy18 by amniocentesis
2	1995	foetus	F	18th wks	26 years	MA Cystic hygroma of neck on ultrasound. X monosomy by biopsy of villi
3	1997	foetus	F	17th wks	28 years	IUD. Probable renal malformation for oligohydramnios Foetal morphology not valutable
4	1998	foetus	M	22th wks	28 years	MA Karyotype not valutable. On ultrasound absence of amniotic fluid, R cystic kidney, L kidney not observed.
5	1998	newborn	M	33th 21 days	30 years	Cesarean section for oligohydramnios and intestinal perforation
6	1998	foetus	M	23th wks	29years	MA. On ultrasound absence of amniotic fluid and cystic hygroma of neck, dilatation of urinary tract and probable cardiac malformation
7	1999	foetus	M	15th wks	30 years	History of VZV infection MA Probable renal malformation for oligohydramnios and severe dilatation of urinary bladder for stenosis of urethra
8	1999	foetus	NV	17th wks	29 years	MA. On ultrasound hydrop foetalis, cystic hygroma of neck. Karyotype not valutable
9	2000	foetus	M	17th wks	35 years	Mother with congenital hydronephrosis for stenosis of pelviureteric junction MA On ultrasound absence of amniotic fluid, severe dilatation of urinary bladder Kidneys and urinary tract not valutablei
10	2000	newborn	M	38th wks few min	42 years	Intrapartum asphyxia. Oligohydramnios and exposure to NSAIAs in utero
11	2001	foetus	M	21th wks	28 years	MA. Probable renal malformation for oligohydramnios Normal karyotype
12	2001	foetus	F	23th wks	37 years	MA. On ultrasound absence of amniotic fluid and probable multiple foetal malformations Normal karyotype
13	2001	foetus	M	17th wks	29 years	MA. On ultrasound absence of amniotic fluid, severe dilatation of urinary bladder and atresia of urethra
14	2001	foetus	F	16th wks	25 years	MA. On ultrasound oligohydramnios, defect of torax and abdomen with herniation of stomac, small bowel, R lung and heart. Morphology of heart not valutable Probable fusion of lower limbs

Legend: NV=not valutable, M=male; F=female; wks=weeks; min=minutes; MA=medical abortion; R=right; L=lef, IUD= intrauterine death, VZV=varicella zoster virus; NSAIAs=non-steroidal anti-inflammatory agents

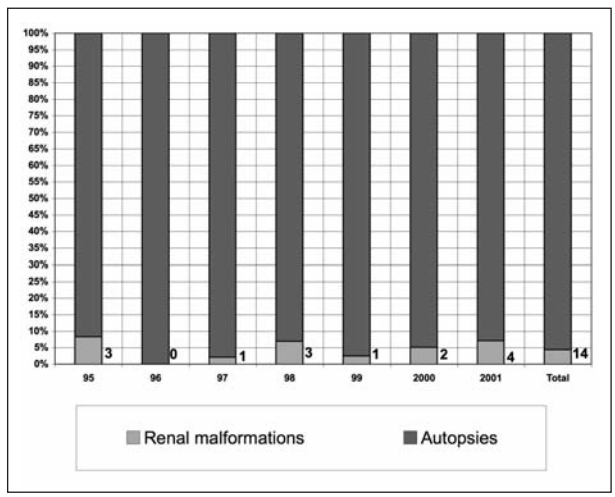
ing by amniocentesis or biopsy of villi (cases 1 and 2) (table 1)

2) presence of hydrops foetalis, with cystic hygroma of neck (case 8)
3) probable renal malformations associated with oligohydramnios, absence of amniotic fluid and abnormalities of urinary tract (cases: 4, 6, 7, 9, 11, 12, 13, 14)

Pathological findings

Pathological findings are summarized in table 2.

In all cases we evaluated external anomalies, renal anomalies and alterations of urinary tract, other organs and placenta.



Graphic 1. Distribution of renal malformations (1995-2001)

Table 2. Renal malformations. Pathological findings

Case	Sex	External abnormalities	Renal abnormalities	Abnormalities of urinary tract	Abnormalities of other organs and placenta
1	M	Abnormal face with prominent occiput, reduction of bifrontal diameter	Horseshoe Kidneys	Absence of other malformations	Absence of other malformations. Placenta with subcorial thrmbosis
2	F	Abnormal face with micrognatia, cystic hygroma of neck	Horseshoe Kidneys	Absence of other malformations	Interventricular septal defect. Placenta with histological findings due to probable chromosome aberration
3	F	Potter's face with prominrnt folds, flattened nose and low-set posteriorly rotated ears. Anus located at vestibular region	R kidney located between poas muscles. L kidney located in iliac region Presence of cystic dysplasia of its parenchymas	Small tubular urinary bladder	Abnormalies of intestinal rotation Rectal atresia. Placenta with chorioamnionitis and funisitis
4	M	Potter's face with prominernt epicanthic folds, flattened nose and low-set posteriorly rotated ears. Anus located at vestibular region	Absence of R kidney. Cystic dysplasia of L kidney	Absence of R ureter. Small, tubular urinary bladder	Absence of other malformations. Placenta with changes due to MA
5	M	Potter's face with prominernt epicanthic folds, flattened nose and low-set posteriorly rotated ears, reduction of head circumference	Absence of proximal tubules, presence of basophilic tubules sometimes with cystic dilatations, sinuous profile and immureactivity for EMA	Small tubular urinary bladder	Microcephaly Intestinal agangliosis Intestinal perforation with meconium peritonitis. Emphysema. Diffuse hepatic steatosis
6	M	Generalized soft tissue edema. Abdominal distension and talipes	Horseshoe Kidneys, with cystic dysplasia	Severe dilatation of urinary bladder and ureters	Cardiac malformations including L ventricular hypoplasial, atresia of mitral valve, interventricular septal defect,. Pulmonary hypoplasia
7	M	Potter's face with prominernt epicanthic folds, flattened nose and low-set posteriorly rotated ears	Cystic dysplasia	Severe dilatation of urinary bladder and ureters	Absence of other malformations. Placenta with changes due to MA
8	F	Abnormal face with low-set posteriorly rotated ears. Generalized soft tissue edema	Horseshoe Kidneys, with cystic dysplasia	Absence of other malformations	Foetal hydrops. Placenta with histological findings due to probable chromosome aberration
9	F	Abnormal face with low-set posteriorly rotated ears and micrognatia. Absence of external genitalia, anus vagina and urethra	Absence of R kidney, hypoplasia and cystic dysplasia of L kidney	Severe dilatation of urinary bladder Duplication of L ureter Atresia of urethra	Rectal atresia. Absence of uterus and vagina Presence of overies

(continued)

Table 2 ***(continued)***. Renal malformations. Pathological findings

Case	Sex	External abnormalities	Renal abnormalities	Abnormalities of urinary tract	Abnormalities of other organs and placenta
10	M	Potter's face with prominernt epicanthic folds, flattened nose and low-set posteriorly rotated ears	Reduction of proximal tubules, presence of basophilic tubules sometimes with cystic dilatations and sinuous profile	Absence of other malformations	Pulmonary hypoplasia Emphy sema and pneumotorax
11	M	Abnormal face with prominent epicanthic folds Abdominal distension	Renal parenchyma with cystic dysplasia and occasional foci of metaplastic cartilage	Severe dilatation of urinary bladder and ureters. Stenosis of memabranous urethra	Absence of other malformations Placenta with changes ùdue to MA
12	F	Abnormal face with with prominent epicanthic folds. Absence of L thumb, anus,urethra external genitalia. Presence of single lower limb without feet	Abesence of kidneys	Absence of ureters, urinary bladder and uterus. Presences of ovaries	VACTERL complex Abnormalies of intestinal rotation, with atresia of colon. Absence of gallbadder uterus and vagina. L radial aplasia. Single umbilical artery arising from abdominal aorta. Presence of single femur, tibia and one phalanx
13	M	Abnormal face with with prominent epicanthic folds. Abdominal distension.	Cystic dysplasia	Severe dilatation of urinary bladder and ureters	Absence of other malformations. Placenta with changes due to MA
14	F	Abnormal face with with prominent epicanthic folds. Defect of torax and abdomen. Lower limbs fused. Presence rudimentary feet	Absence of Kidneys, ureters and uterus and vagina. Presence of ovaries	Absence of ureters, urinary bladder and urethra	Herniation of heart, stomac part of R lung, small bowel and liver through defect of abdominal and thorax.wall. Absence of uterus and vagina Presences of ovaries

Legend: M=male; F=female; MA= Medical abortion, R=Right, L=Left; EMA=epithelial membrane antigens

In some cases of this study the renal malformations and anomalies of other organs already were evident on macroscopic examination.

Histological examinations, which were always performed, in some cases added new data to macroscopic findings or disclosed microscopic parenchymal abnormalities.

Thus, in cases 2 and 7 histological examination of placenta confirmed or proposed diagnosis of chromosome aberrations.

In either cases, infact we observed alterations of villi, such as edema of the stroma, trophoblastic inclusions and mineralizzation of basal lamina (Fig. 1).

In a cases 5 and 10 histological examination of kidneys was essential for diagnosis because it revealed a peculiar alterations of renal tubular differentiation, named renal tubular dysgenesis.

This disorder in case 5 was characterized by absence of cortical proximal tubules and the presence of basophilic tubules, sometimes with cystic dilatations, sinuous profile (Fig. 2a) and immunohistochemical positivity for EMA (Fig. 2b).

In case 10 renal tubular dysgenesis showed severe reduction of proximal tubules and numerous basophilic tubules (Fig. 3a).positive for EMA on immunohistochemical analysis (Fig 3b).

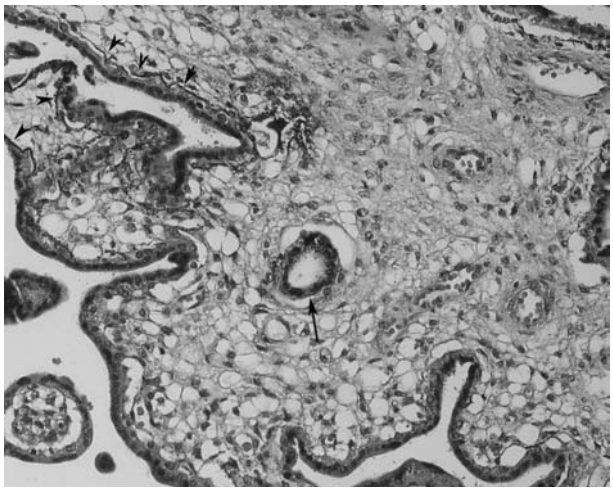


Figure 1. Histological examination of placenta in cases of chromosomal aberrations showed alterated villi with oedema of the stroma trophoblastic inclusions and mineralization of basal lamina (haematoxylin-eosin x 200, arrow: trophoblastic inclusions)

Histological examination of intestinal wall, in case 5 furthermore disclosed reduction of gangliar cells, hypertrophy of nervous fibers (Fig. 4), while macroscopic examination revealed microcephaly and large cranial fontanels.

All cases of renal malformations, characterized by cystic aspect of parenchyma, with or without alterations of urinary tract, by histological examination were classified as cases of renal cystic dysplasia.

Sometimes cysts enlarged kidney and replaced completely renal parenchyma (Fig. 5).

This malformation histologically was recognized by disorganization of renal parenchyma, which showed cyst formation and presence of immature nephronic and ductal structures.

Primitive ducts characteristically were lined by columnar epithelium and surrounded by collar of mesenchymal cells in whorl-like pattern (Fig. 6a), containing foci of extramedullary hemopoietic cells, nervous fibers or bars of metaplastic cartilage (Fig. 6b).

Histologic serial sections of urethra in case N 11 disclosed presence of stenosis (Fig. 7).

In cases 4 and 9 because cystic dysplasia of one kidney was associated with agenesis of other, absence of uterus and vagina, we formulated diagnosis of renal adysplasia.

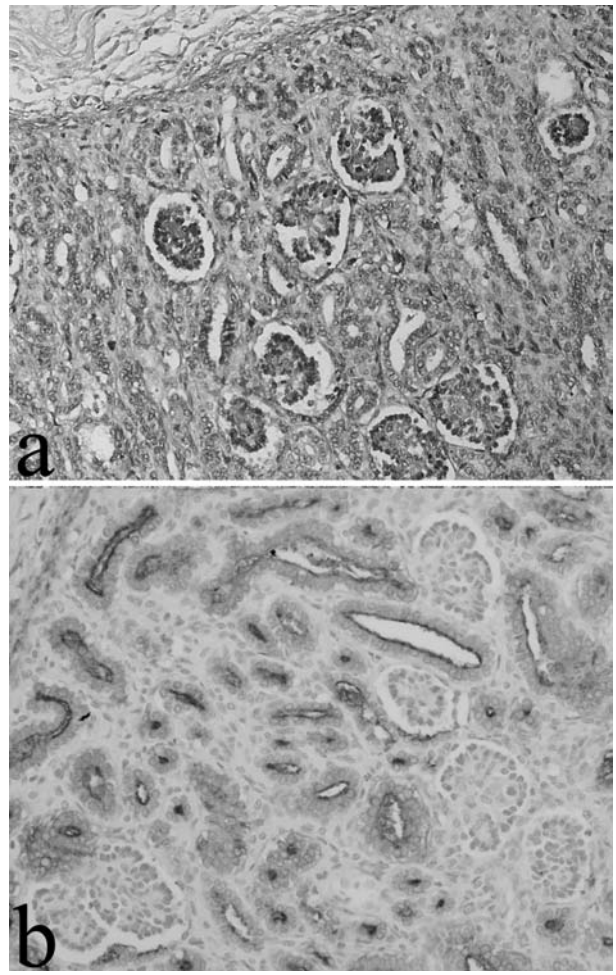


Figure 2. Histological section of kidney in renal tubular dysgenesis (case 5) revealed presence of basophilic tubules , sometimes with cystic dilatations, sinuos profile (a: haematoxylin-eosin x100) immunoistochemically positive for EMA (b: x 100)

In cases 6 and 8 cystic dysplasia instead was combined with horseshoe kidneys.

In cases 12 and 14 macroscopic examination was essential for diagnosis.

External anomalies in these cases were severe and evident. Case 12 were characterized by abnormal face, absence of left thumb, anus urethra and external genitalia and presence of single lower limb without feet (Fig. 8).

In case 14 lower limbs were fused, feet were rudimentary (Fig. 9a), thoracic and abdomen wall presented a defect with herniation of heart, stomac, part of right lung, small bowel and liver (Fig. 9b).

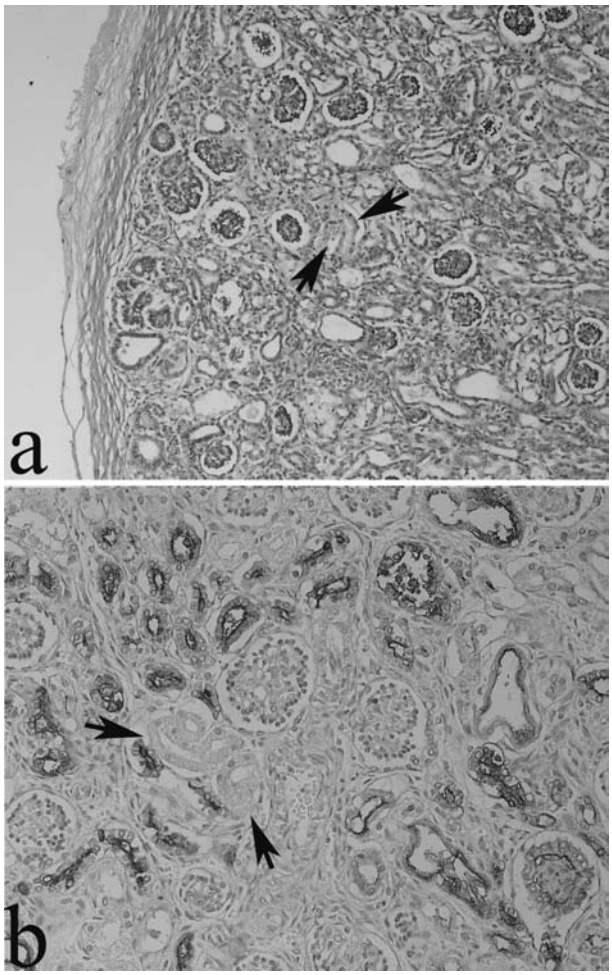


Figure 3. Histological examination of kidney in renal tubular dysgenesis (case 10) revealed severe reduction of proximal tubules and numerous basophilic tubules (a: haematoxylin-eosin x100; arrow: rare proximal tubules). All basophilic tubules in renal tubular dysgenesis (case 10) were positive for EMA, while proximal tubules were negative (b: x 200; arrow: negative proximal tubules)

Either cases were evaluated by radiological study which disclosed in the first case presence of single femur, single tibia, single lower phalanx (Fig. 10a) and absence of radius (Fig.10b); while in the second fusion of lower limbs.

During evisceration in case 12 it was observed absence of kidneys, abnormalies of intestinal rotation (Fig. 11a), VACTERL complex, with absence of gallbladder and interventricular septal defect (Fig. 11b), atresia of rectum, absence of uterus and vagina and single umbilical artery arising from abdominal aorta.

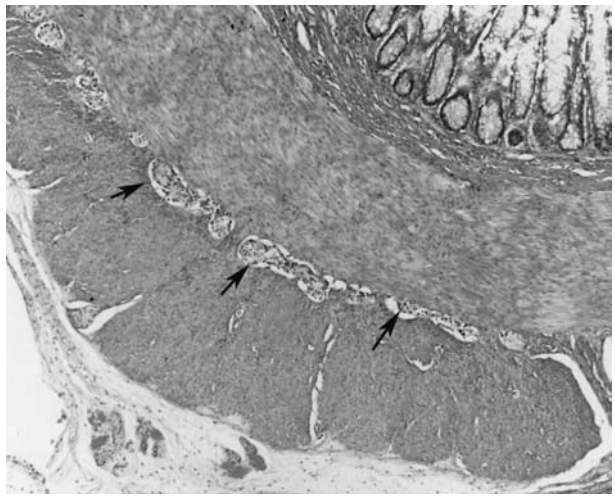


Figure 4. Histological section of intestinal wall (case 5) disclosed reduction of gangliar cells and hypertrophy of nervous fibers (haematoxylin-eosin x 40; arrows: hypertrophy of nervous fibers)

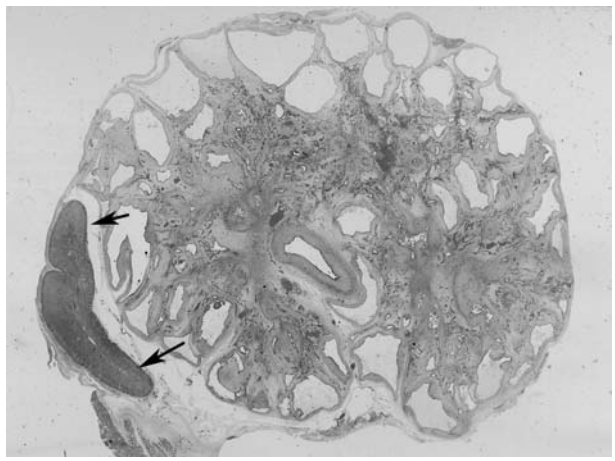


Figure 5. Histological section of kidney with cystic dysplasia at low magnification; note the complete replacement of renal parenchyma with cysts and atrophy of adrenal gland (haematoxylin-eosin; arrows: atrophic of adrenal gland)

In case 14 examination of internal organs revealed absence of kidneys and excluded cardiac malformations.

Thus, we might formulate in these cases the diagnosis of sirenomelia and bilateral renal agenesis

associated with radial aplasia, VACTERL complex in cases 12 and defect of thoracic and abdominal wall with herniation of some organs in case 14.

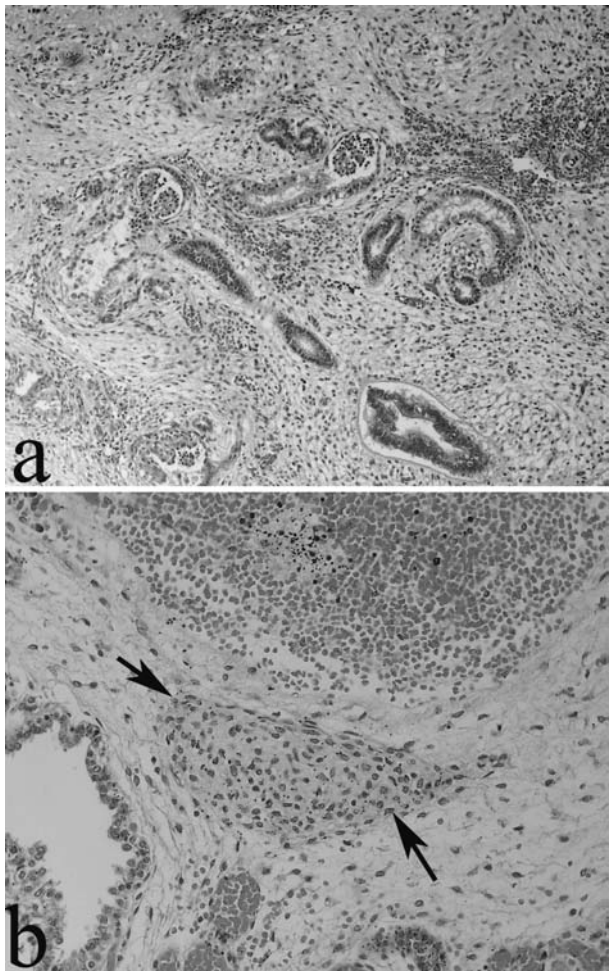


Figure 6. On histological examination kidney with cystic dysplasia showed presence of primitive ducts lined by columnar epithelium, surrounded by collars of mesenchymal cells (a: haematoxylin-eosin x 100), containing sometimes bar of metaplastic cartilage (b: haematoxylin-eosin x 200 arrows metaplastic cartilage)

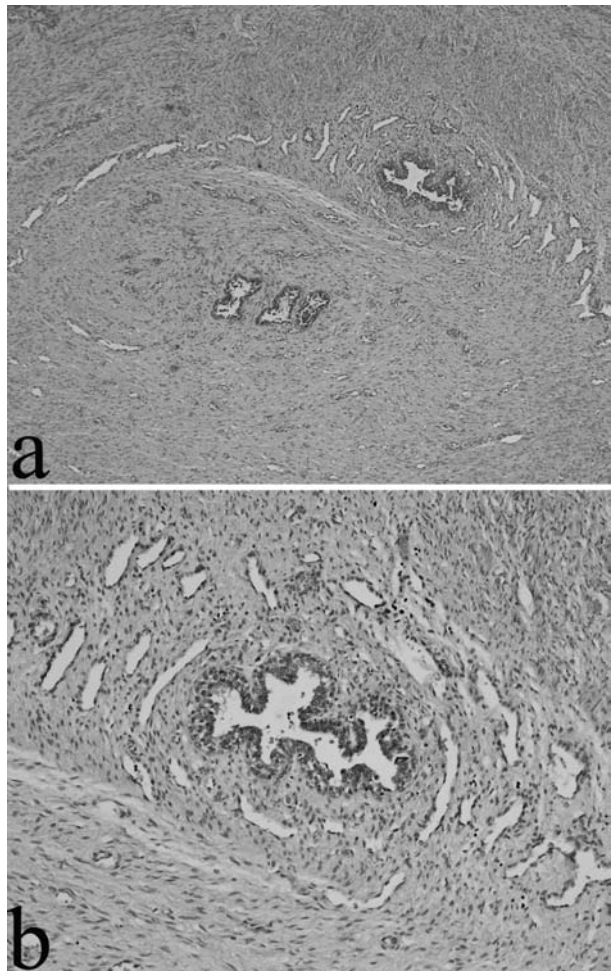


Figure 7. On serial histological examination urethra showed an abnormal urethral configuration, characterized by two lumens (a: haematoxylin-eosin x 40) and then one chanel (b: haematoxylin-eosin x 100)

In conclusion, by macroscopic and microscopic examination we identified in this series the following renal malformation:

- bilateral renal agenesis: 2 cases (cases: 12 and 14) (table 2)
- renal tubular dysgenesis: 2 cases (cases: 5 and 10) (table 2)
- abnormalities of renal shape: 2 cases (cases: 1 and 2) (table 2)
- renal cystic dysplasia: 4 cases (cases: 3, 7, 11, 13) (table 2)
- renal adysplasia: 2 cases (cases 4 and 9) (table 2)
- abnormalities of renal shape with cystic dysplasia: 2 cases (cases: 6 and 8) (table 2).

The distribution of these subtypes of renal malformations are illustrated in graphic 2, where it is evident that all subtypes had same frequency except for cystic dysplasia, which represents the most frequent anomaly (Graphic 2)

The comparison of clinical and pathological diagnosis, summarized in table 3, revealed that autoptic examination:

- confirmed clinical diagnosis of renal malformation in 1 case (case 7) (table 1 and 2).

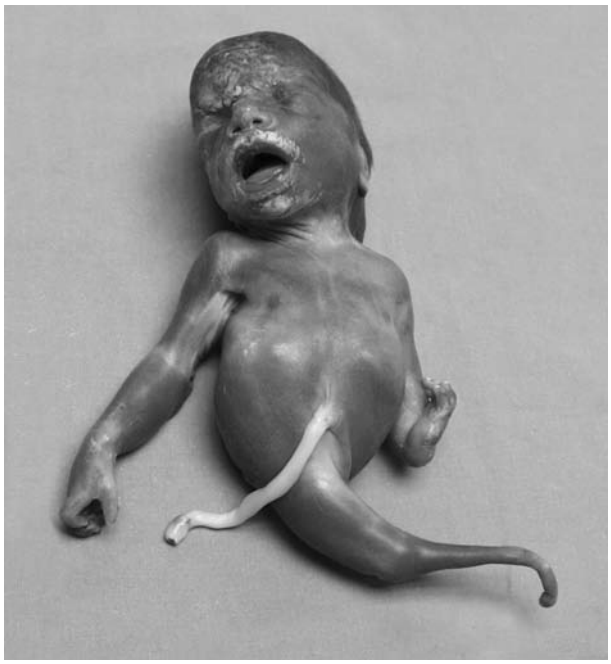


Figure 8. External examination in case 12 showed abnormal face, absence of left thumb, external gentalia and presence of single lower limb without feet

- Provided extensive additional informations in 7 cases (cases: 4, 6, 9, 11, 12, 13, 14) (table 1 and 2)
- Disclosed renal malformations escaped on prenatal studies in 6 cases (cases: 1, 2, 3, 5, 8, 10).

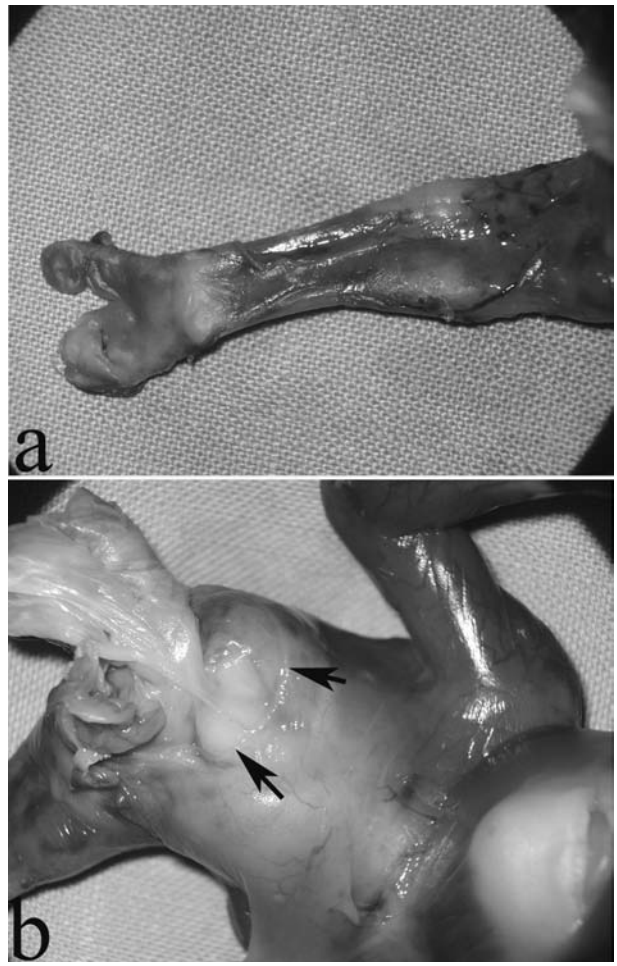


Figure 9.External examination in case 14 revealed fusion of lower limbs and rudimentary feet (a) and defect of thoracic and abdominal wall with herniation of part of heart right lung, small bowel and liver (b, arrow: defect of thoracic and abdominal wall

Conclusions

Malformations of kidney result from alterations of normal process which follow one another, during normal development of the kidney and the urinary tract.

Traditionally malformations of kidneys are classified as abnormalities of position, number, shape or parenchymal differentiation.

Often, two or more of these abnormalities coexist and may be associated with anomalies of other systems to form recognizable syndromes.

Malformations of kidney also may be classified as malformations compatible with life and malformations incompatible with life.

Malformations incompatible with life are due alterations of parenchymal differentiation and its are characterized by reduction or absence of renal function; these only after birth cause death because during intrauterine period, renal functions are absolved by placenta.

Characteristically renal malformations with alterations of parenchymal differentiation during intrauterine life are associated with absence or reduction of amniotic fluid (oligohydramnios).

The lack or reduction of amniotic fluid, arising as decreased foetal urine production and the resultant pressure of the uterine wall on the developing foetus,

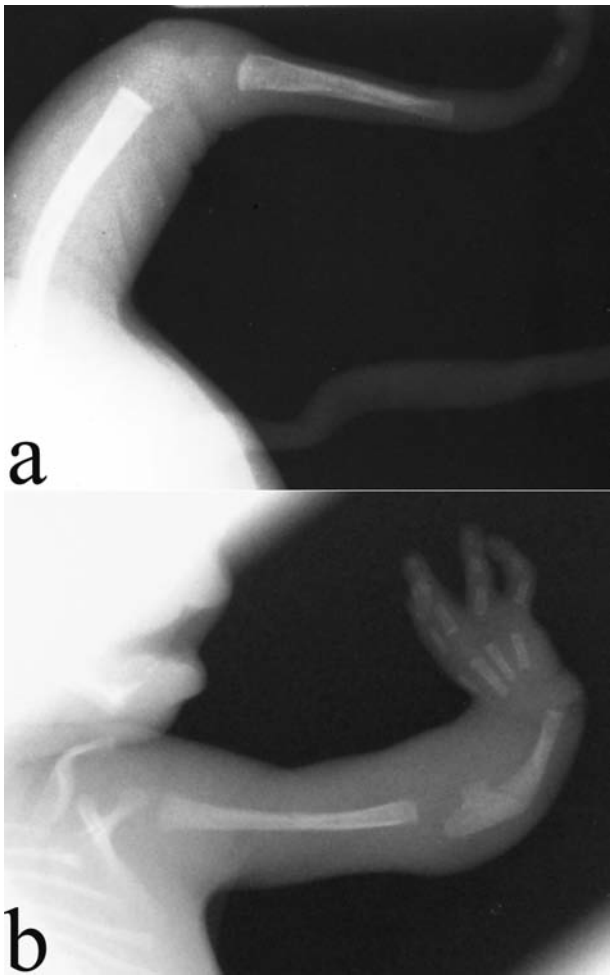


Figure 10. Radiological study of single lower limb in case 12 disclosed single femur, tibia and single phalanx (a) and absence of radius, first metacarpus and phalanxes of thumb in upper left limb

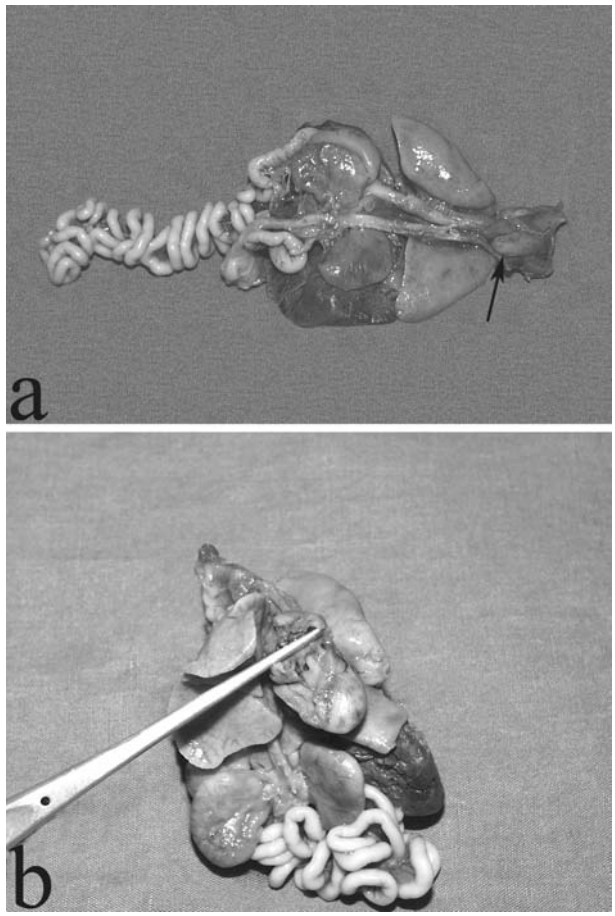


Figure 11. Internal examination in case 12 revealed oesophageal atresia abnormal intestinal rotation with apple-peel deformity (a: arrow oesophageal atresia) and interventricular septal defect (b).

contribute to the accompanying abnormalities in severe renal malformations, termed Potter sequence.

This includes abnormal face with prominent epicanthic folds, flattened nose and low set posteriorly rotated ears, bowing of the legs and inward rotation of the feet, spadelike hands and reduced chest circumference.

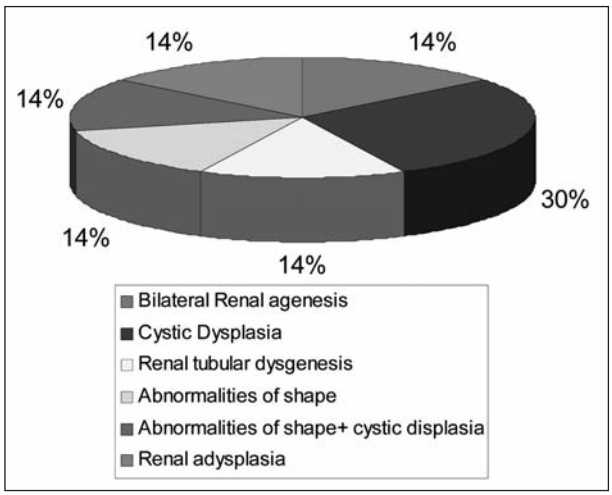
Potter's phenotype is also characterized by pulmonary hypoplasia, which results from alterations of physical factors that permit the lungs to grow and to mature such as adequate intrathoracic space, adequate amount of amniotic fluid.

Oligohydramnios and absence of amniotic fluid possibly retard lung growth for lung compression, affecting foetal breathing, the volume of fluid within the airspaces and stretching of the pulmonary tissue (2).

Prenatal ultrasound examination of renal malformations are essential for early diagnosis.

However, prenatal diagnosis of renal malformations is yet more difficult than other malformations, because they are associated with absence of amniotic fluid or oligohydramnios.

Ultrasound examination of the foetus in these conditions is severely hampered, because the normal acoustic window and foetal-fluid interfaces which aid imaging are lost and the foetus lies in a flexed position with crowding of the small parts making examination difficult.



Graphic 2. Renal malformations: distribution of subtypes

Table 3. Comparison of clinical and pathological diagnosis

Cases in which post-mortem examination confirmed clinical diagnosis	1 case	7,14%
Cases in which post-mortem examination confirmed clinical diagnosis and provided extensive additional informations	7 cases	50%
Cases in which only post-mortem examination indivituated renal malformations escape on prenatal studies	6 cases	42,85%

The injection of warmed isotonic fluid into the amniotic cavity, a procedure known as amnioinfusion may be helpful to became foetal image more clear (3, 4).

Accurate echographic diagnosis of renal malformations is also correlated with other factors, including experience of operator, the resolution of echographer, foetal position during ultrasound examination, the thickness of maternal abdominal wall and type of malformation (5, 6).

Peculiar renal malformations remain particularly difficult to discover on ultrasound examination.

The ultrasonic diagnosis of renal agenesis is difficult since it is harder to diagnose the absence of a structure than the presence of an abnormal structure.

Hypertrophied adrenal glands filling the renal fossae in renal agenesis may give the false impression of kidneys.

Similarly renal malformations characterized by parenchymal microscopic alterations escape to ultrasound diagnosis.

In our series there are two examples of renal malformation whose diagnosis was made only in neonatal period by histological examination. These cases are a rare lethal renal malformation, known as renal tubular dysgenesis. This abnormality may be an autosomal recessive disorder and it may be recognized by only histological examination of kidneys, which reveals parenchymal alterations including reduction or absence of proximal tubules (7, 8).

The cardinal clinical manifestations are oligohydramnios, Potter phenotype and neonatal renal and respiratory failure.

Renal tubular dysgenesis also may be an acquired disorder due to conditions with decreased foetal-placental blood flow and oxygen/nutrient to the foetus, such as twin-twin transfusion syndrome (9, 10) and exposure to ACE inhibitors or non-steroidal anti-inflammatory agents in utero (11).

In this series the first case of renal tubular dysgenesis probably is an hereditary disorder, because during pregnancy there is not exposure to teratogenic agents.

This case similarly to cases reported by other Authors is associated with microcephaly and large cranial fontanels (12), but it very interesting for the presence of agangliosis of large bowel, responsible of dilatation and rupture of bowel.

The second case of renal tubular dysgenesis, diagnosed in post-natal period, probably is an acquired disease for history of exposure to non-steroidal anti-inflammatory agents during pregnancy.

Similarly with other studies, the most common renal malformation observed in foetuses in our series is renal cystic dysplasia (13, 14).

This anomaly were largely studied by Osathanondh and Potter (14).

Kidneys with cystic dysplasia do not have a uniform appearance and range from small to large. The condition may be bilateral, unilateral or segmental, involving part of kidney

Cystic dysplasia encompasses renal lesions which fall into: Potter type II, Potter type IV and Potter type III.

In cystic dysplasia type II the kidney may be small masses or may be large, associated with thread-like-ureter and a narrow tubular bladder.

In cystic dysplasia type III normal and abnormal nephrons are intermingled, both kidneys are affected

In cystic dysplasia type IV renal cysts are associated with incomplete urinary obstruction, such as valvular obstruction in posterior urethra and there is usually hydronephrosis, hydroureter and dilatation of urinary bladder (14).

It is obscure the varying significance of the differences between type of cystic dysplasia.

According to Osathanondh and Potter cystic dysplasia type II and type IV may result from a similar defect (i.e obstruction to urine outflow) and differ each other only in the stage of development at which the defect arises (14).

Type III kidneys instead are the result of a mechanism or a number of mechanisms that affect some nephrons while leaving others intact (14).

In present study almost all cases of cystic dysplasia was identified by ultrasound examination, but sometimes post mortem examination provided extensive additional informations such as agenesis of other kidney, malformations of genital tract and alteration of kidney's shape.

Thus, among cases of renal cystic dysplasia we observed two cases of renal adysplasia.

This disorder is an uncommon condition in which agenesis of one kidney is combined with cystic dysplasia in other.

Renal adysplasia can be a sporadic disorder, but according studies of some Authors it can have an autosomal dominant mode of inheritance and it has wide variability of expression (15, 16).

Other affected members of a family, infact, may show renal unilateral agenesis, renal multicystic dysplasia or alterations of genital tract (15, 16).

Thus, ultrasound study of kidney or accurate genital tract examination of parents, sibs and other relatives are recommended in all families in which a case of renal adysplasia is observed (17).

Renal malformations with abnormalities of shape in our study, detected in a case of trisomy 18 and monosomy X, are horseshoe kidneys in which the right and left kidney are linked at one end by a band of tissue as a result of fusion of the corresponding poles.

In these cases despite to the chromosomal aberrations which was clinically observed the renal malformation was undiagnosed.

Thus, post-mortem examination may confirm diagnosis of chromosomal alteration detecting alterations of phenotype and histologic alterations of placenta and it also reveals a renal malformation (18).

In another case of horseshoe kidney we may express the diagnosis a chromosome aberration observing same histological alterations of placenta (case 8).

The cases of bilateral renal agenesis in our study are observed in two examples of severe malformative complex, incompatible with life, termed sirenomelia.

This disorder is a rare condition, the reported incidence ranging from 1:60000 (19) and 1: 1000000 births (20).

Sirenomelia sequence is characterized by fusion of the lower extremities or the presence of a single lower limb, which may have two feet, one foot or no recognizable foot. It known as "caudal regression syndrome" (21).

The sirenomelia often represents the most common malformation in monozygotic twins .

Malformations reported in sirenomelia include renal agenesis, associated with genitourinary, anorectal and vertebral abnormalities (23, 24).

However, more recently some cases with normal kidney are described, too (22-25).

The etiopathogenesis of sirenomelia is unclear.

Some cases are correlated with maternal diabetes (26), but in other cases the etiologia is known.

With regard to the pathogenesis it is controversial.

According some Authors the sirenomelia is the consequence of vascular anomaly, because this disorder often is associated with presence of a single umbilical artery, which, arising from the abdominal aorta, causes deviation of blood and nutritive substances from more caudal structures of embryo to placenta (27, 28).

Other Authors suggested that sirenomelia is a consequence of a defect of more caudal part of mesoderm (29).

In our series prenatal diagnosis of sirenomelia is suspected in two cases.

Autoptic study in addiction to data obtained by ultrasound revealed other abnormalities such as VACTERL complex absence of kidneys, gallbladder, rectal atresia, radial aplasia .

The presence of Vacterl complex and thoracic and abdominal defect in our cases of sirenomelia supports the hypothesis that some malformative associations, characterized by severe and numerous defects, can be polytopic field defects, which are correlated, affect different structures in different parts of the body and develop early during blastogenesis (30-32).

In conclusion, the results of this study confirmed the need of pathological examination for foetuses and newborns, after medical abortion and neonatal death.

Furthermore, the pathologist's contribution to the multidisciplinary management of prenatal or postnatal malformations is fundamental in particular for further genetic counseling.

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Accepted: December 2nd 2011
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