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Major anomalies of the genitourinary tract in the neonate

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Thirty-two neonates were investigated or operated upon for major anomalies of the genitourinary tract during the ten-year period up to June 1981. The infants ranged in age from 1 to 31 days. Obstructive uropathies and conditions which appear mainly in later infancy or childhood were excluded.

Diagnostic and therapeutic problems of renal dysplasia (multicystic kidney), infantile polycystic kidney disease, renal venous thrombosis, tumours, exstrophy of the bladder, combination of paediatric surgical and urological anomalies, and sex assignment in infants are discussed with some case reports.

Data are scarce concerning the surgical management of malformations of the genitourinary tract in the neonate [5]. Analysis of the records of all neonates in whom a malformation of the genitourinary tract required investigation and/or surgery has allowed to draw conclusions as to the diagnostic manoeuvres and management of neonatal genitourinary surgical diseases.

MATERIAL

During the ten-year period up to June 1981, 32 neonates were investigated or operated upon for major anomalies of the genitourinary tract. The age of the infants ranged from 1 to 31 days.

Obstructive uropathies, which comprise nearly half of the urologic diseases in the newborn and present the most challenging problem to those responsible for the care of these patients, were excluded from the study. Furthermore, conditions which appear mainly in later infancy or childhood and usually do not require emergency surgery were also neglected, thus undescended testicles, testicular torsion, inguinal hernia, hydrocele, phimosis, hydrocolpos, vulvar synechia, hymenal polyp, vaginal and paraurethral cysts.

Renal dysplasia (multicystic kidney)

Although surgically managable cystic diseases of the kidney are uncommon in early infancy, the possibility should be taken into account, especially in the case of a palpable abdominal mass.

Of the group of renal dysplasias multicystic kidney is the most frequent abdominal mass palpable in newborn infants and the most common cystic lesion of the neonatal kidney. In this condition the kidney

s composed of cysts of variable size and is usually larger than normal (Fig. 1). During excretory urography contrast medium could not be visualized on the affected side, while the unaffected side had a normal pyelographic appearance. In view of the risk of injury, hypertension or infection and the possibility of a neoplasm, the mass is usually removed surgically.

All of our 6 neonates with multicystic kidneys were treated by nephrectomy and all of them are thriving.

Polycystic kidney

The term "polycystic kidney disease" includes more than one syndrome. Any classification of these diseases is only an arbitrary frame to work with and evaluate data. Most workers distinguish between an infantile and an adult type of polycystic kidney. The two entities differ greatly in pathology, genetics, clinical picture, X-ray appearance and prognosis.

Infantile polycystic kidney disease

This condition is considered to have a single, autosomal, recessive mode of transmission. Blyth and Ockenden divided it into four groups: — perinatal, neonatal, infantile and juvenile — according to the patient's age at the clinical presentation [3]. The kidneys in infantile polycystic disease are huge or enlarged and may almost fill the abdomen. Both cortex and medulla are filled with radially oriented small cysts. Death, often shortly after birth, is usually the result of respiratory distress, pulmonary hypoplasia, lung rupture, etc. Increasing renal insufficiency, pyuria, haematuria and hypertension develop in the infants who survive the neonatal period. The X-ray findings are striking. A plain film of the abdomen shows the massively enlarged kidneys as soft tissue masses. In the excretory urogram there is often a delay of several minutes or hours before the appearance of the contrast medium in the kidneys. The renal pelvis and calyces are distorted. In several cases

FIG. 1. Multicystic kidney

the dye never concentrates in the calyces or pelvis. Only a purely symptomatic treatment is a reality in the infantile and juvenile forms (increasing chronic renal insufficiency, hypertension, anaemia, respiratory problems, etc.). Diagnosis, treatment, genetic counselling and prognosis are mainly determined by the type of the disease.

In one of our neonates with polycystic kidney disease, genetic counselling and prognosis met with difficulties. The disease, infantile polycystic kidney, is known to be hereditary, and analysis of the family pedigree showed a dominant trait (adult type polycystic kidney). The boy was born of the 3rd pregnancy of the mother. The abdomen was protuberant, both kidneys were enlarged to 8×4 cm, with lobated contours (Fig. 2a). The urogram showed poor dye excretion. Angiographic studies via the umbilical artery confirmed a neonatal type polycystic kidney (Fig. 2b).

The maternal grandmother died of renal failure at 43 years of age. Post mortem revealed huge kidneys with numerous cysts of variable size, containing blood-tinged or clear fluid. The mother has elevated blood pressure, her left kidney is enlarged, the calyces are distended but clinically she is healthy. The patient's sister died at the age of 2 hours. Each of her kidneys was as large as a female fist, and they displayed countless small cysts in both cortex and medulla. The infant's hypertensive sibling, a 4-year old boy, is also affected; his left kidney is enlarged.

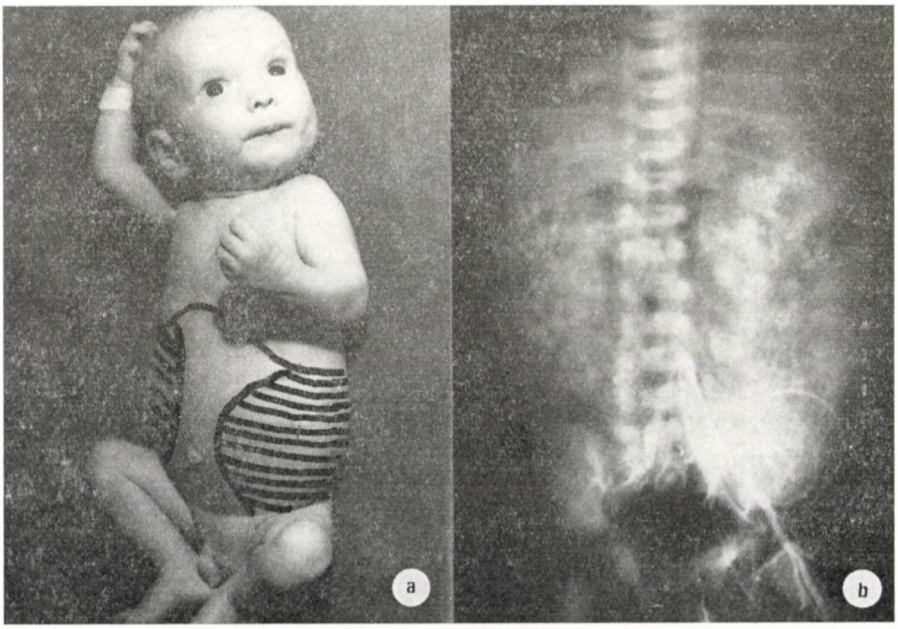

FIG. 2. Infantile polycystic kidney: *a)* hatched areas show the enlarged kidney *b)* angiography via umbilical artery

Renal venous thrombosis

Renal venous thrombosis is suspected in a neonate when haematuria and an enlarged kidney are observed shortly after a period of asphyxia, shock, dehydration or sepsis. The suggestion has been made that renal venous thrombosis is a bilateral disease and only the less involved kidney is overlooked [1].

Nephrectomy used to be advocated to prevent infection and the infrequent hypertension which usually means a serious problem after healing, and renal atrophy. Now it would seem that immediate nephrectomy, unless infection or some other untoward development occurs, is no longer the operation of choice for unilateral thrombosis since without thrombectomy some recovery of function may be achieved as a result of an incomplete or segmental involvement or subsequent recanalization [7]. Bilateral thrombosis may be treated by thrombectomy. During the ten-year period analysed, we observed 3 neonates with this condition. In one of them a non-surgical approach was chosen; this patient survived. In one of the nephrectomized infants the infection and thrombosis progressed, the thrombosis spread to both renal veins and inferior venae cavae. This patient died 8 days postoperatively.

TABLE I

Major genitourinary anomalies requiring surgical consideration in the neonate*

Anomalies	No. of cases	Total
Renal dysplasia (multicystic kidney)	6	
total		5
segmental		1
Polycystic renal disease	5	
infantile type		4
adult type		1
Renal vein thrombosis	3	
Tumour	5	
Wilms'		1
cong. mesoblast. nephroma		1
retroperit. neuroblast.		1
pelvic neuroblastoma		2
Exstrophy of bladder	1	
Combination of paediatric surgical and urological anomalies	7	
compatible with life		2
incompatible with life		5
Severe cases of ambiguous external genitalia	5	
Total	32	

* Obstructive uropathy, undescended testes, testicular torsion, inguinal hernia, hydrocele, phimosis, hydrocolpos, synechia vulvae, hymenal polyp, vaginal and paraurethral cysts, etc., have been excluded.

Tumours

A review of our records revealed an interesting fact: from 1960 to 1975 we had no patient with a malignant tumour at birth or within the first months of life, whereas in the past 5 years we have recorded eight such patients, five of them requiring urological management.

Two of the tumours arose from the kidneys. One of them was a Wilms tumour, the other a congenital mesoblastic nephroma. Physical examination and X-ray studies at birth detected in a 1650 g preterm infant a left renal tumour (Fig. 3a). The removed kidney with the tumour weighed 170 g (Fig. 3b). On the cut surface we saw a nearly complete replacement of the renal parenchyma by a bulging firm mass, presenting a yellowish silk pattern (Fig. 3c). Microscopic diagnosis was congenital mesoblastic nephroma. The infant is now 13 months old, weighs 11 kg and thrives well.

The third tumour also mimicked a kidney neoplasm on the intravenous urogram, but surgery proved it to be a retroperitoneal tumour located in front of the intact but displaced and compressed kidney. Histologically it was a neuroblastoma.

Two patients presented with urinary retention due to pelvic and presacral tumours, which had dislocated the rectum and obstructed the blad-

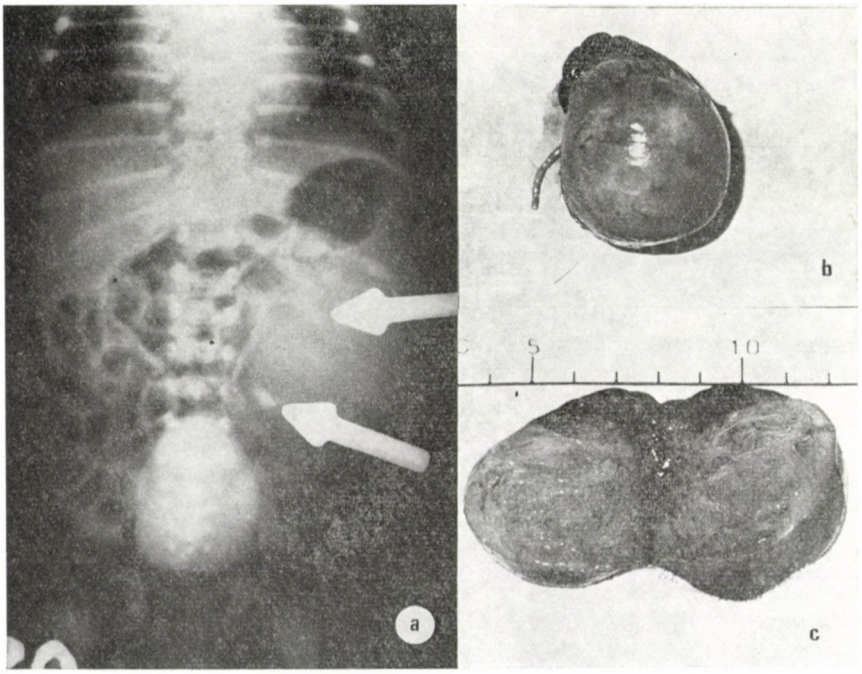

FIG. 3. Congenital mesoblastic nephroma: *a*) intravenous urogram: the distorted left renal pelvis is displaced medially and compressed (arrows), *b*) removed kidney with tumour below *c*) cut surface of tumour and kidney

der outlet. Histologically both tumours proved to be capsulated differentiated neuroblastomas.

In view of the age of these patients and the radical excision of the tumours, neither radiotherapy nor cytostatic drugs were given, but all patients were and still are followed regularly. Nowadays it is generally accepted that in the neonate and young infant aggressive multimodal therapy may be more dangerous than the disease itself [4].

Exstrophy of the bladder

Exstrophic conditions form a broad spectrum of developmental abnormalities: the lowest degree is represented by epispadias, while the highest is exstrophy of the cloaca or vesico-intestinal fissure.

The only treatment of bladder exstrophy applicable in newborns is vesical reconstruction. Several authors are of the opinion that such surgery should be carried out at the age of 2 to 3 years. However, according to a considerable number of other workers, the operation is more easily and effectively performed in the neonatal period when the wall of the bladder is still thin and pliable. Our attempts at such early reconstruction of the bladder have failed.

Combination of surgical and urological anomalies

Malformations compatible with life. We had only two neonates in whom both the paediatric and urological malformations required surgical correction within the first month of life. In one of them rectal agenesis and ureterovesical obstruction necessitated a pull-through operation and uretero-nephrectomy (Figs 4a and b).

Malformations incompatible with life. In four patients with multiple grave anomalies palliative surgical interventions were only feasible, after which

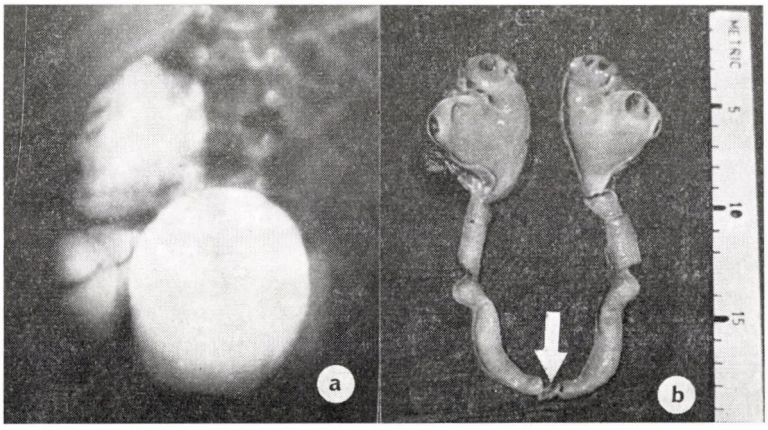

FIG. 4. Ureterovesical obstruction (↑) associated with rectal agenesis: *a*) cystogram: severe dilatation of the right upper urinary tract, *b*) removed kidney and ureter: hardly any renal parenchyma is visible

they died within a few days. In one of them the post mortem revealed partial agenesis of the colon, rectal agenesis, colo-vesical fistula, bipartite uterus, urogenital sinus, obstruction of the left pelviureteric junction and the uretero-vesical junction, with Meckel diverticulum and vertebral anomalies.

Another neonate had a megacystis-microcolon-hypoperistalsis syndrome, a condition described by Berdon et al in 1976, as a new cause of neonatal intestinal obstruction [2], a uniformly fatal combination of urological and intestinal abnormalities. The patient was a girl born at term; she was admitted with abdominal distension and bilious vomiting. The stomach was displaced upwards by the distended urinary bladder. Gastrographin enema demonstrated a microcolon. Meconium ileus or intestinal obstruction was suspected. At surgery the enlarged bladder was filled with urine and reached the umbilicus. The small intestine measured only 60 cm from the pylorus to the ileocoecal valve. Both kidneys were enlarged. A double-barrel ileostomy was prepared but satisfactory intestinal function never ensued and the infant died on the 28th postoperative day.

Sex assignment in the newborn with ambiguous external genitalia

Prompt and appropriate assignment of sex in the newborn infant with ambiguous genitalia is an important task. When confronted with such conditions the attending physicians including the paediatric surgeon should be aware of the anxiety imposed on the family.

The first task is to examine the external genitalia. One should appraise the anatomic abnormalities, noting the size of the phallus, the location of the urethral orifice, the degree of labioscrotal fold fusion, the presence or absence and the size and location of palpable gonads, and the presence of other abnormalities. The findings are usually confusing even for the experienced specialist. The continuum of changes between purely female and purely male external genitalia have been divided into five stages by Prader [6]. It should, however, be stressed that the findings obtained on examination of the external genitalia never allow a diagnosis to be made, since disorders with an identical external appearance may have different causes (Figs 5a and b).

Evaluation of the internal genitalia is more difficult. It should be known whether or not gonads are present and whether they are dysgenetic or histologically normal testes or ovaries. In addition, it is important to know whether or not an uterus and Fallopian tubes are present.

The urogenital sinus can be explored under general anaesthesia, and a catheter and an endoscope of 10 Ch may be helpful in finding the vaginal opening. Radiological examination of the urogenital sinus, using a viscid contrast medium, is often informative. In favourable cases the vagina and the narrow cavity of the uterus will be outlined.

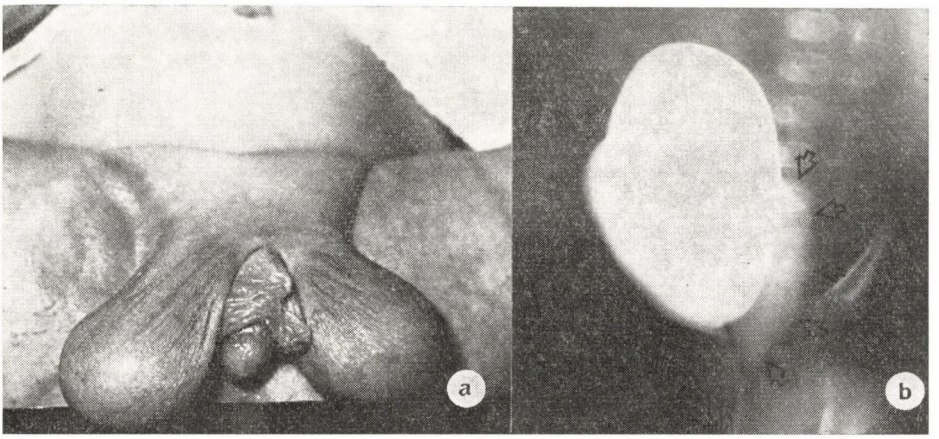

FIG. 5. Neonate with ambiguous genitalia: *a*) external appearance, *b*) genitography: uterus outlined by arrows (chromosomal analysis: 46 XY)

In order to ascertain the absence or presence of internal genitalia and to obtain biopsies from the gonads, a laparotomy or laparoscopy [8] has to be carried out even in small neonates. Nowadays, we give preference to the latter method.

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