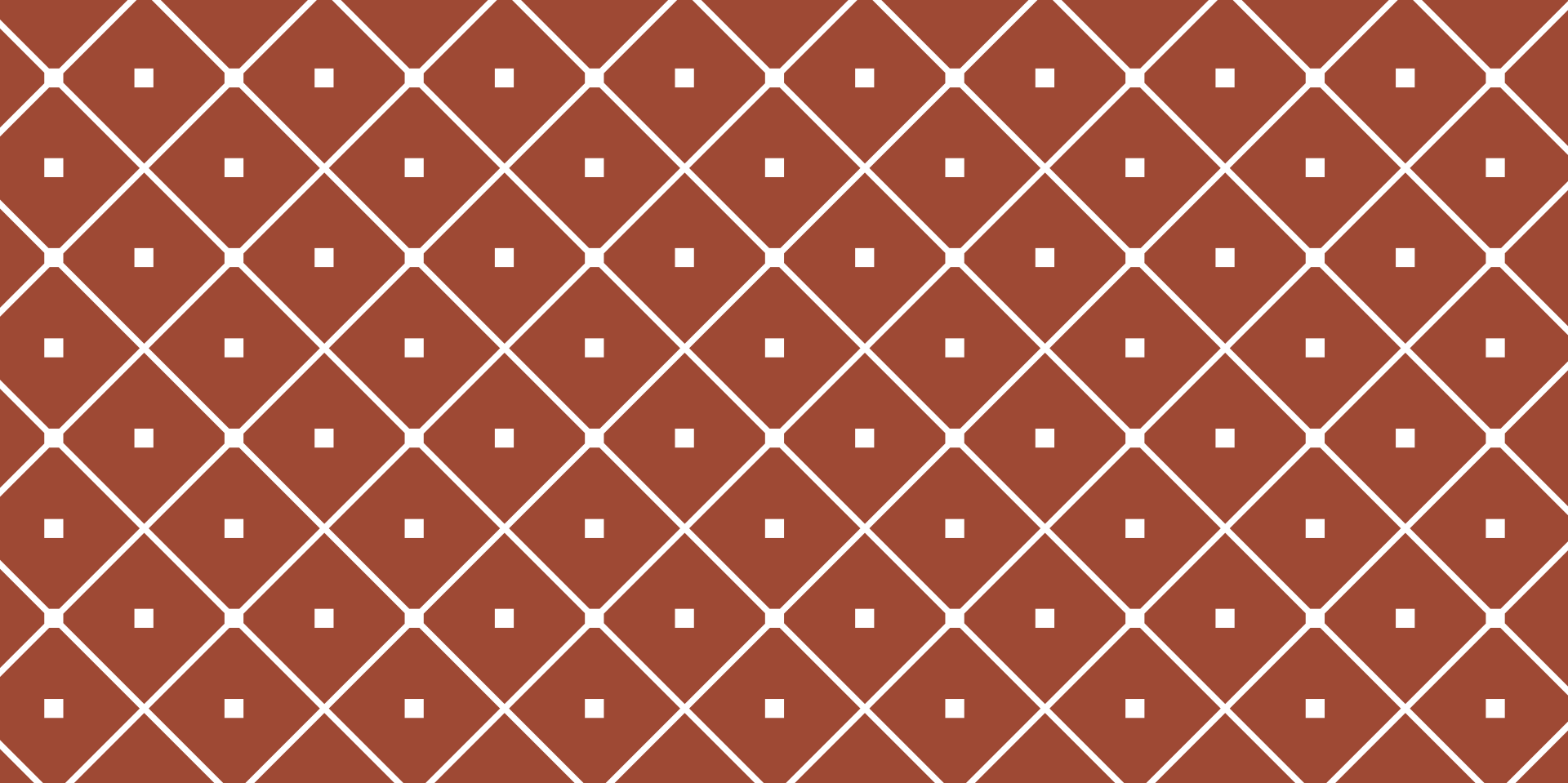




PEDIATRIC UROLOGY

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FEBU EGYPT

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MEMBER OF AUA & EAU

CONSULTANT UROLOGIST , SHIFA HOSPITAL



CONTENTS

VUR.

PUJ.

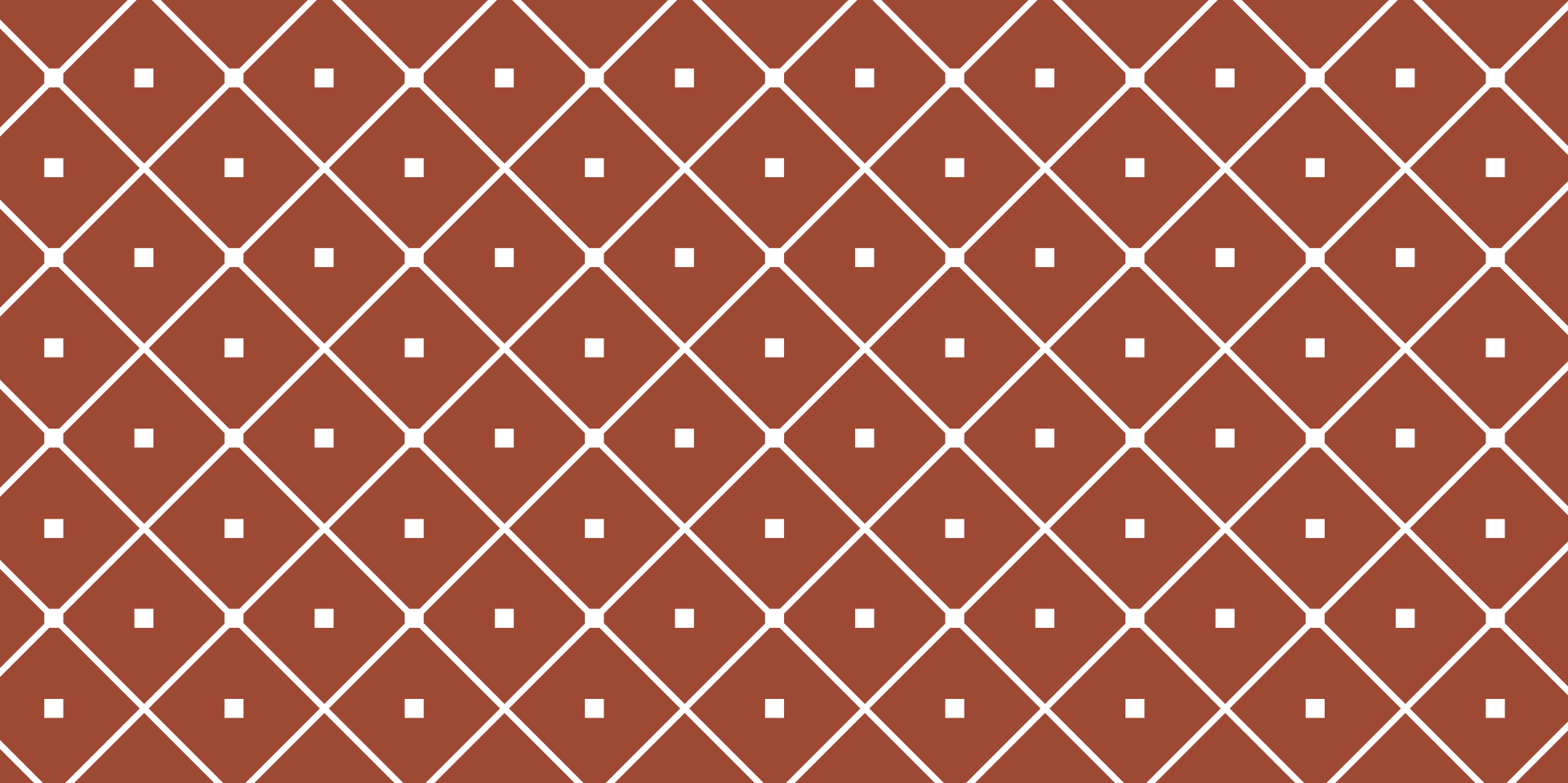
PUV.

Megaureter.

Anomalies of UUT.

Ureterocele.

Cystic Diseases of the Kidney.



VUR

VUR is the retrograde flow of urine from the bladder to the upper urinary tract

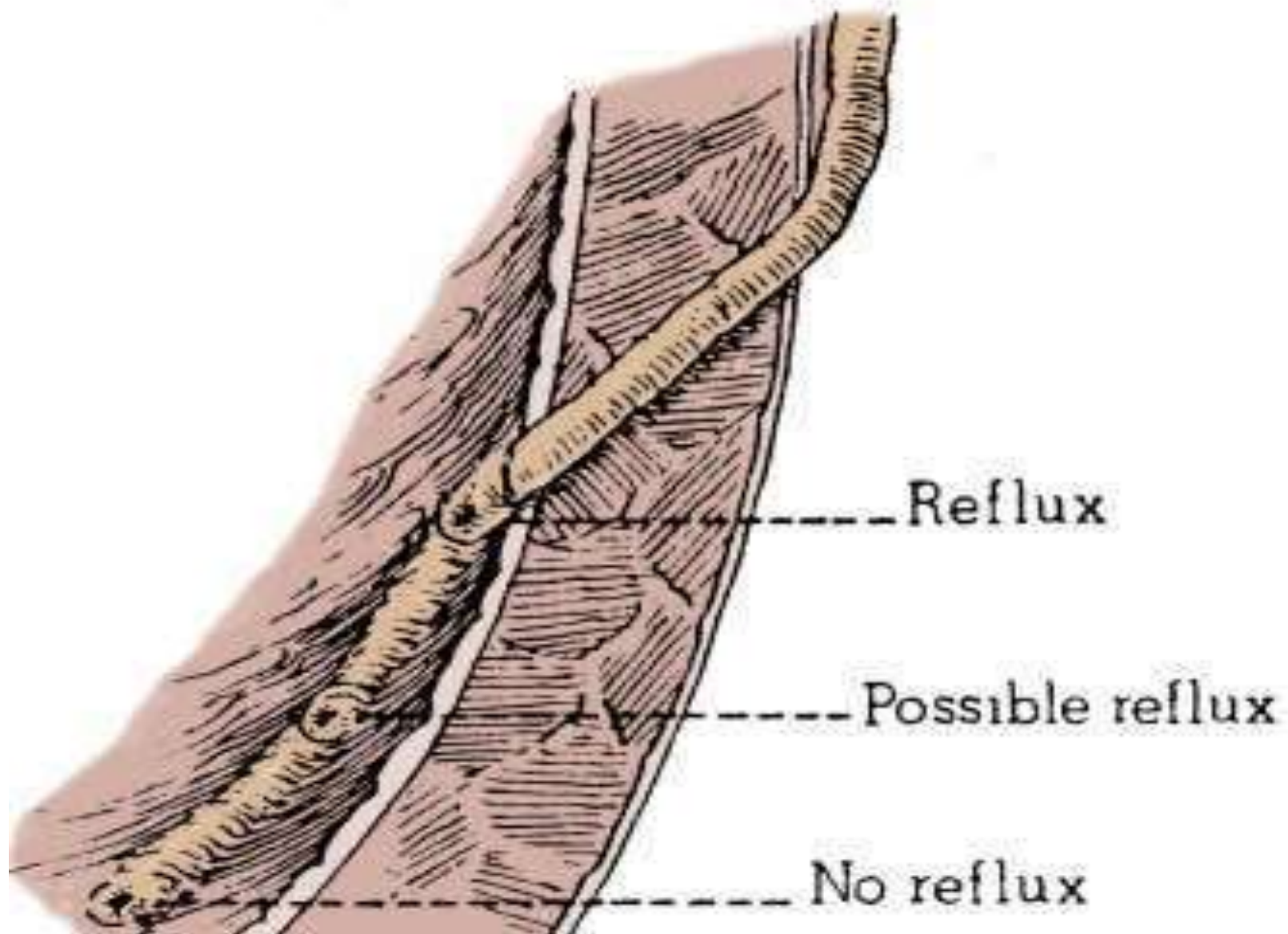
76% of refluxing infants are male even though (85%) of reflux in older children is in females.

INCIDENCE (%)	AGE (yr)
70	<1
25	4
15	12
5.2	Adults

ANTIREFLUX MECHANISM

Anatomic composition of the ureterovesical junction: children revealed an approximately **5 : 1** ratio of tunnel length to ureteral diameter in non refluxing junctions compared with a **1.4 : 1** ratio in refluxing UVJs.

Functional dynamics of the bladder.



CAUSES OF VUR

Primary Reflux:

fundamental deficiency in the function of the UVJ (length-diameter ratio is almost always less than **5:1**)

Secondary reflux:

normal function of the UVJ being overwhelmed

Anatomical causes:

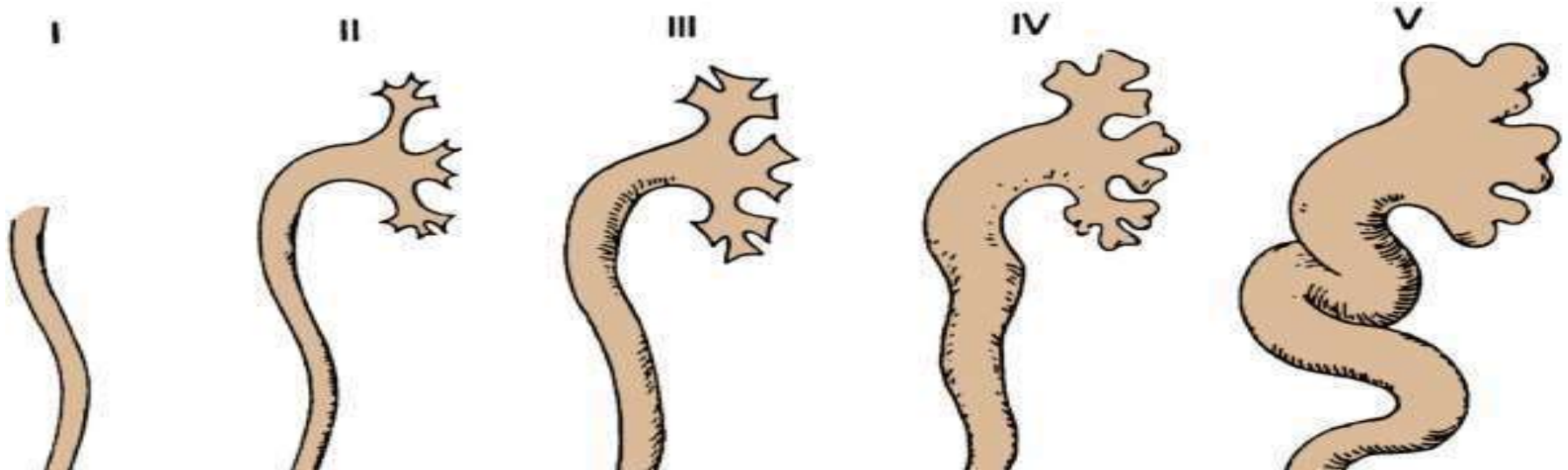
- PUV's is The most common Reflux is present in **48% to 70%** of PUV patients .
- Ureterocele
- Ureteral duplication

Neuro-functional causes:

- Neurogenic bladder – Spina bifida
- Uninhibited bladder contractions.
- Constipation

Grade Description

I	Into a nondilated ureter
II	Into the pelvis and calyces without dilatation
III	Mild to moderate dilatation of the ureter, renal pelvis, and calyces with minimal blunting of the fornices
IV	Moderate ureteral tortuosity and dilatation of the pelvis and calyces
V	Gross dilatation of the ureter, pelvis, and calyces; loss of papillary impressions; and ureteral tortuosity



CLINICAL FEATURES

Features of recurrent **UTI**:

- Fever
- Flank pain
- pyuria.

Palpable renal **mass**

Delayed growth

Weight loss

ASSESSMENT OF LOWER URINARY TRACT

The voiding cystourethrogram (**VCUG**) and radionuclide cystogram (**RNC**) are the present-day gold standard approaches to reflux detection.

VCUG Provides information on :
functional dynamics and structural anatomy

Contraindicated in active cystitis.

Static films

- bladder contour
- presence of diverticula
- ureterocele
- grade of reflux
- configuration & blunting of calyces
- bladder neck anatomy
- urethral patency.

Dynamic films:

- active reflux

Delayed or **postvoid** films:

- Crucial in documenting clearance of contrast from the upper tracts
- **Dilated PCS + Retained contrast = PUJO**



CYSTOSCOPY

Routine use is not mandated.

Role immediately prior to surgery for confirming:

- orifice position
- duplication
- proximity of diverticula to the orifice
- urethral patency

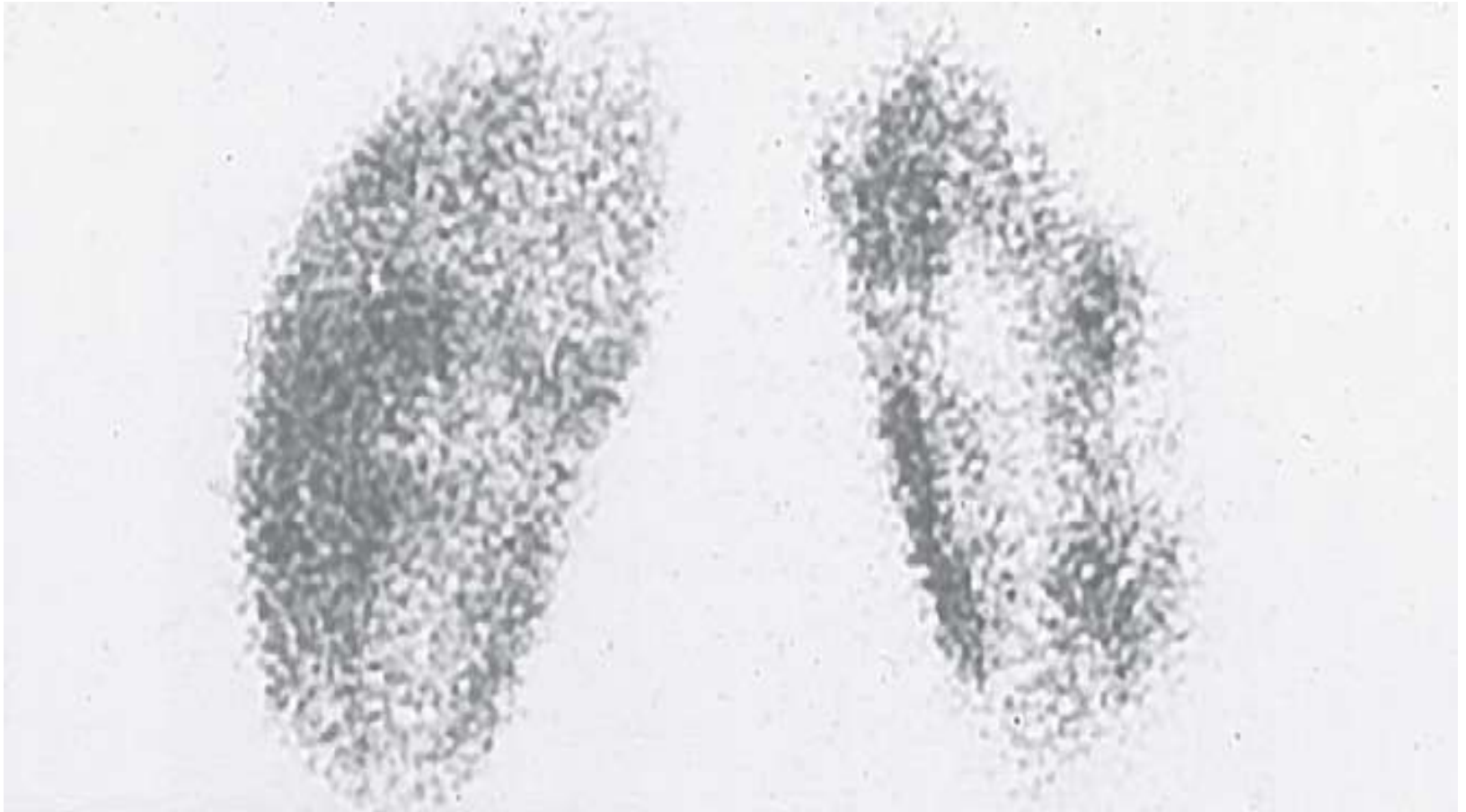
ASSESSMENT OF UPPER URINARY TRACT

USS

DMSA:

- detection of reflux-associated renal damage
- follow-up of reflux

RENAL SCINTIGRAPHY



ASSOCIATED ANOMALIES

PUJ Obstruction:

- incidence of VUR associated with PUJO is (**9%-18%**).
- the incidence of PUJO in patients with reflux is **0.75% to 3.6%**
- incidence with high-grade reflux is five times more likely than lower grades of reflux

Ureteral duplication:

- reflux occurs most commonly into the **lower pole**.
- **Weigert and Meyer rule** (the more lateral and proximal insertion of the lower pole ureter associated with a shorter intramural ureter at VUJ).

Bladder diverticulae:

Outpouching of mucosa between detrusor muscle bundles without any true muscle backing itself

Renal anomalies:

- Renal agenesis: **46%** association
- MCDK: **28%** association
- Presence mandates VCUG

Megacystis-Megaureter syndrome:

- More common in males
- Differentiation from PUV

Resolution by **grade**:

- Most cases of low-grade reflux (grade I and II) will resolve : **63-85%**
- Grade III reflux will resolve in approximately **50%** of cases
- Higher-grade reflux (grades IV and V and bilateral grade III) : **9-25%**

Resolution with **age** :

- Age has greater significance than grade
- Younger patients (**<12 months** old) resolved more quickly.
- The traditional period of observation for resolution is 5 years, probably because the greatest proportion of growth and anatomic remodeling of the UVJ is complete.

PRINCIPLES OF MANAGEMENT

Spontaneous resolution of reflux is very common.

High-grade reflux is less likely to resolve spontaneously.

Extended use of prophylactic antibiotics & “
Watchful waiting”



The success rate with surgical correction is very high.

Sterile reflux is benign.

ABSOLUTE INDICATIONS FOR SURGERY

Break through **urinary tract infections**

Failure of medical management

- patient noncompliance
- persistence of reflux with prolonged medical management.
- progressive deterioration in renal function.



Ureteral **obstruction** assoc with VUR

Refluxing ureter **opening into bladder diverticulum**

Cystoscopic observation of **golf hole orifice**

RELATIVE INDICATIONS FOR SURGERY

Presence of **massive reflux** – gr IV & V

Reflux associated with **paraureteral diverticulum**

In girls whose reflux persists after they have reached the full somatic growth potential at puberty.

Parental preference

TYPES OF SURGICAL REPAIR

According to **approach** :

- Intravesical
- Extravesical
- Combined

According to the position of the sub mucosal tunnel **in relation to the original hiatus** :

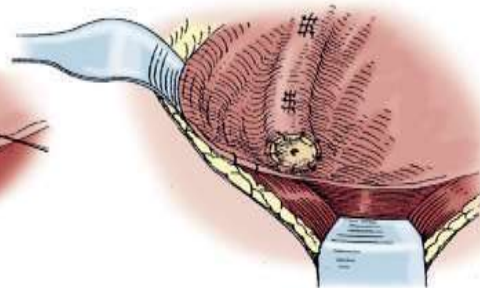
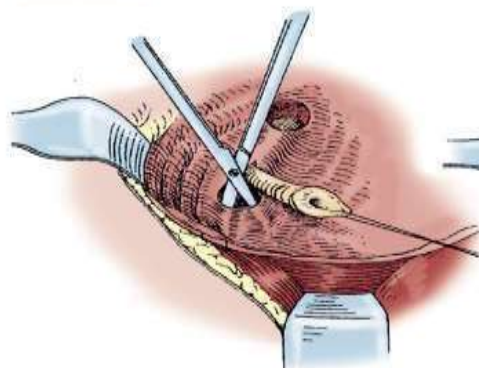
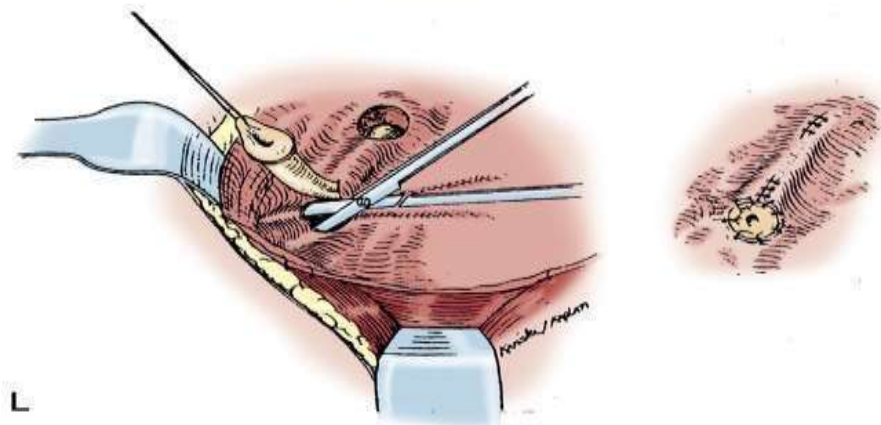
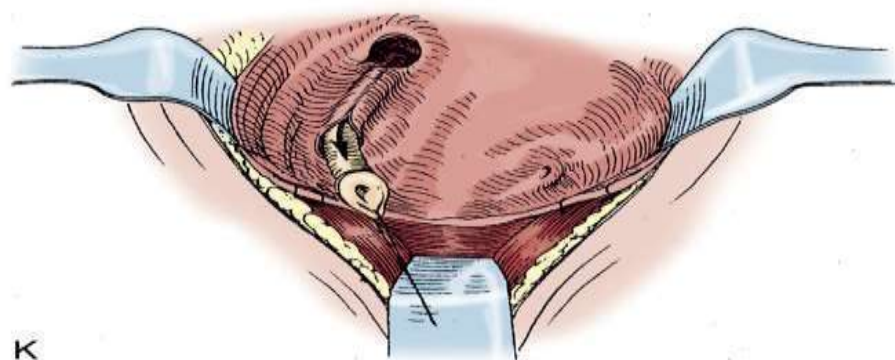
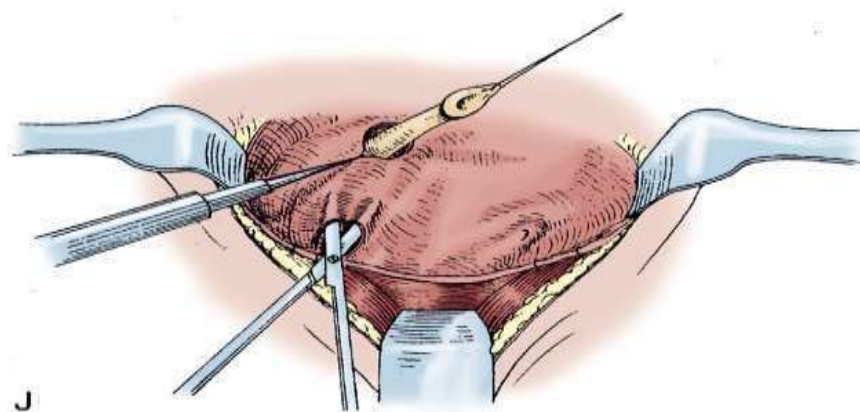
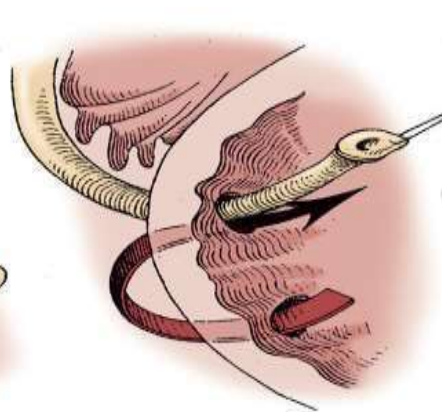
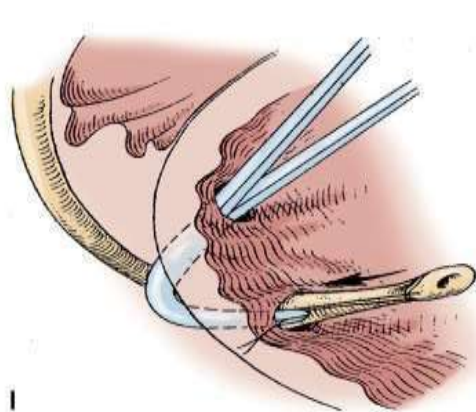
- Suprahiatal
- Infrahiatal

SUPRA HIATAL TUNNEL

Politano-Leadbetter Technique:

A submucosal tunnel is created in the direction of the trigone, medial to the original orifice.

The advantage of this technique is that a long tunnel can be created, which is valuable in the higher grades of reflux.

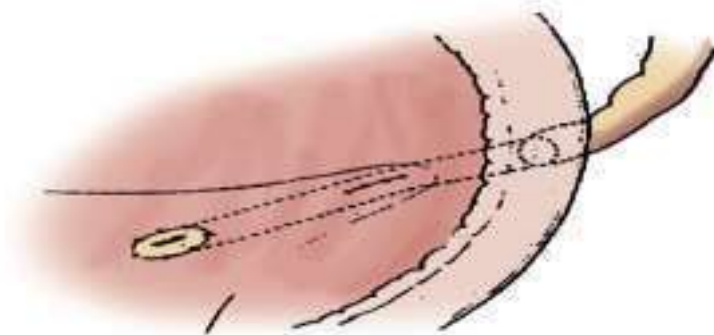
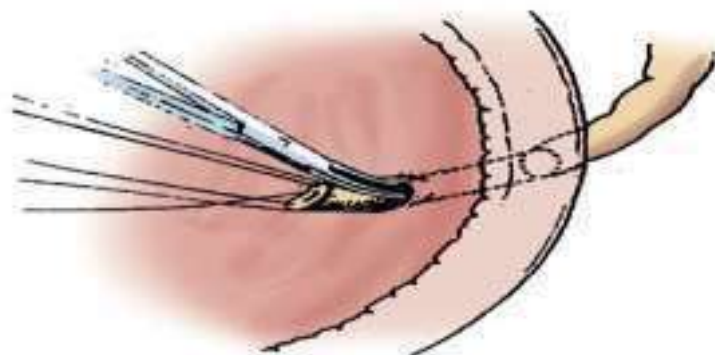
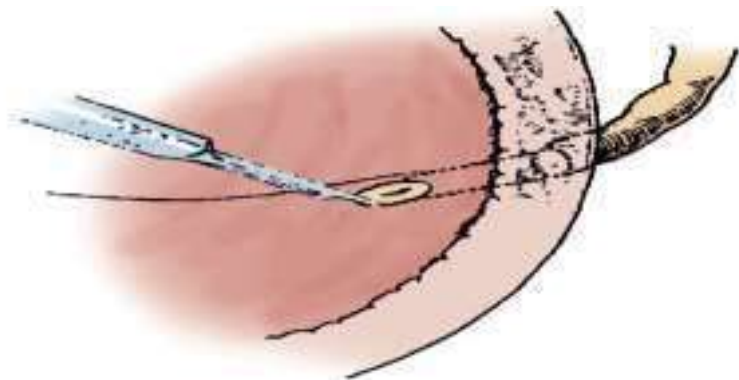


INFRAHIATAL

Glenn-Anderson Technique:

by using the same hiatus and advancing the ureter distally toward the bladder neck.

The distance from the hiatus to the bladder neck limits the length of the tunnel.

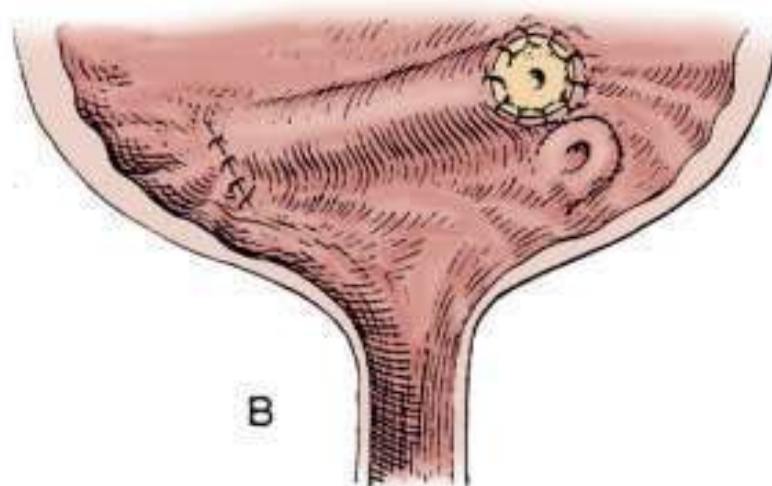
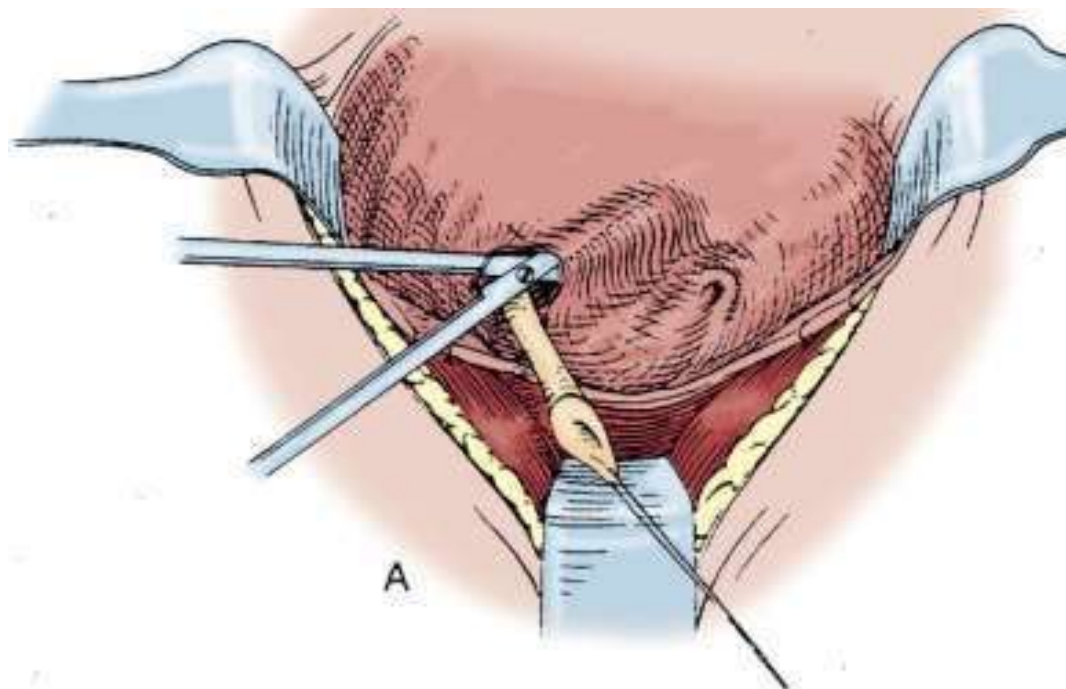




Cohens cross trigonal technique:

- Intravesical, infrahiatal procedure
- Good for small capacity bladder
- Success > 95%

Problem is Difficult retrograde catheterization of ureters





Lich –Gregoir technique:

- Extravesical procedure.
- The advantage is that the bladder is not opened; thus postoperative hematuria and bladder spasms are minimized.

LAPAROSCOPIC SURGICAL PROCEDURES

Gil-Vernet Procedure:

In this procedure the trigonal mucosa is incised vertically and then two ureters are approximated into the midline with a single submucosal suture.

Reported success rates of **60%**.



COMPLICATIONS

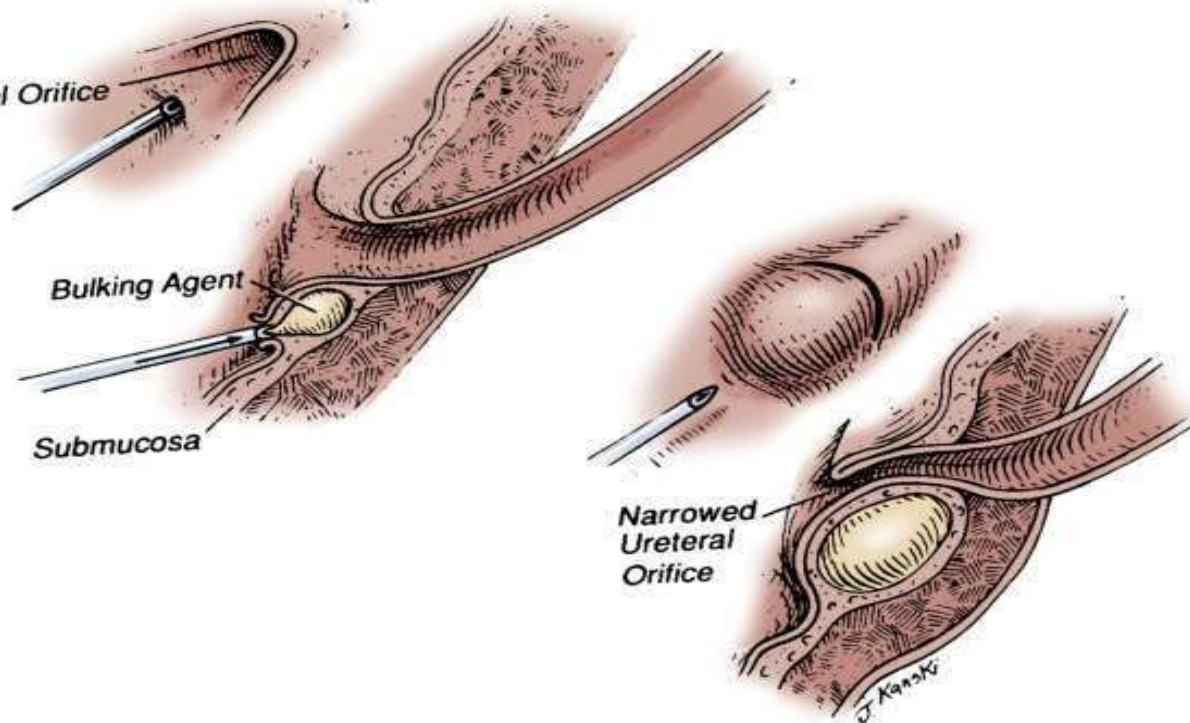
Persistent Reflux.

Contralateral reflux

Obstruction

ENDOSCOPIC MANAGEMENT

Injection of a bio-compatible bulking agent beneath intravesical portion of ureter in sub



Advantages:

- OPD based treatment
- less morbidity, no mortality
- No surgical scar
- Success rate almost equivalent to open surgery for primary reflux.

Disadvantages:

- Cost
- Lower success rate compared to surgery for high grade reflux, up to 90%.

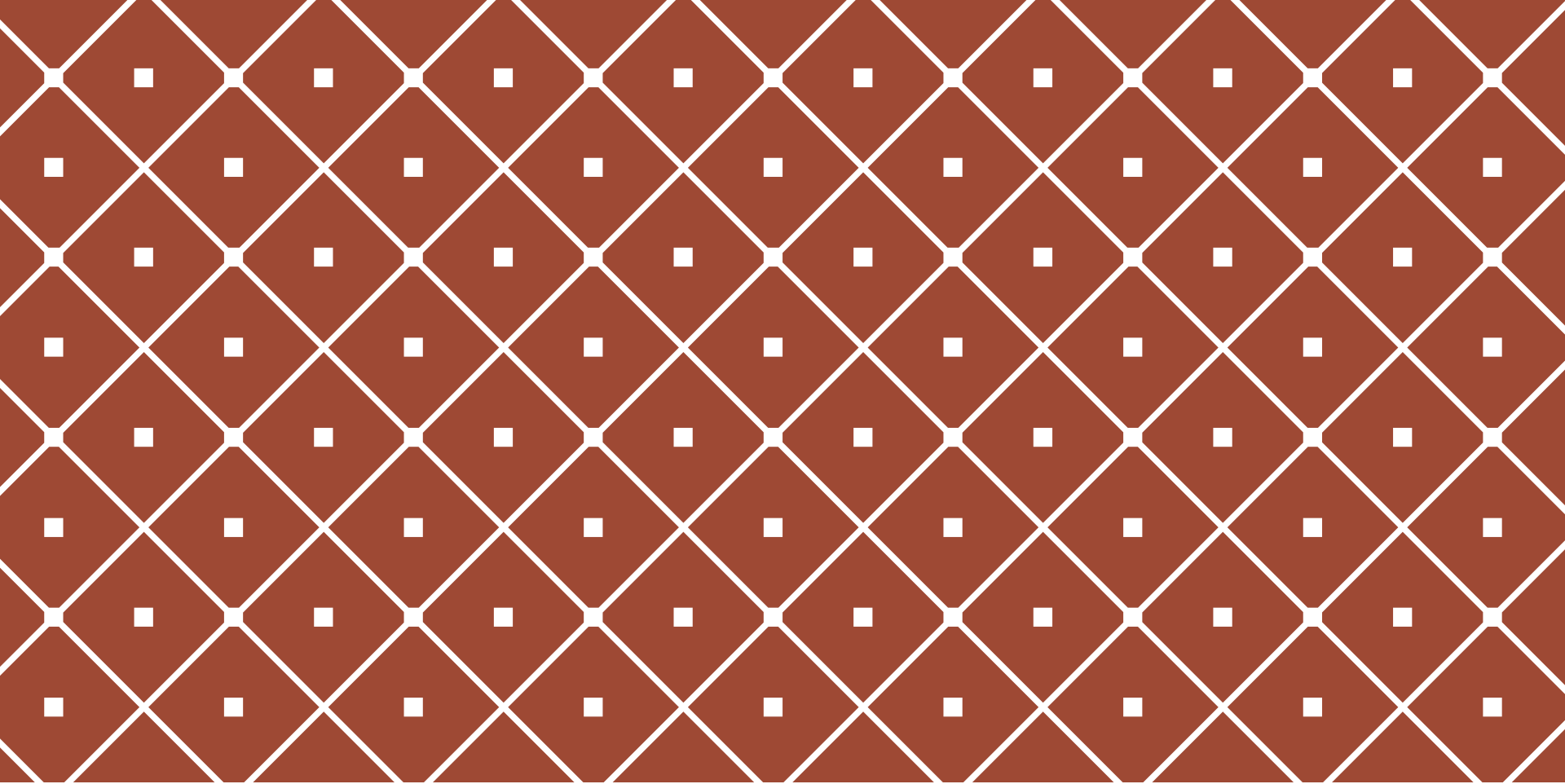
AGENTS FOR INJECTION

Nonautologous Materials:

- Polytetrafluoroethylene (PTFE)
- Cross-linked bovine collagen
- Polydimethylsiloxane
- Dextranomer hyaluronic copolymer (Deflux)
- Coaptite

Autologous Materials:

- Chondrocytes
- Fat
- Collagen
- Muscle



PUJ OBSTRUCTION



It's impairment of urinary transport from the renal pelvis to the ureter.

Most cases are **congenital**.

ETIOLOGY

Acquired condition:

- Stone disease.
- Postoperative.
- Inflammatory stricture.
- Urothelial neoplasm.

Extrinsic obstruction:

- High insertion of ureter at renal pelvis.
- Crossing vessels near UPJ (50% of older children)
- Abnormal rotation of the kidney.

CLINICAL PRESENTATION

Can be manifest clinically **any time** of life.

Most common presentation was finding of palpable flank **mass**.

Asymptomatic newborns being diagnosed with hydronephrosis which caused by UBJO during antenatal follow with US .



Evaluation of azotemia, that result from bilateral obstruction in functionally or anatomically solitary kidney.

Older children or adult:

- Intermittent abdominal or flank pain.
- Nausea or vomiting.
- Hematuria either spontaneous or associated with relatively minor trauma.

DIAGNOSIS

Laboratory finding:

- Microhematuria
- Pyuria
- Frank urinary tract infection.

Imagings: The goal is determining both anatomic site & functional significance of an apparen obstruction.

DIURETIC UROGRAPHY

Commonly used study for diagnosis both UPJ & ureteral obstruction

Provide quantitative data regarding differential renal function and obstruction , even in hydronephrotic renal units.

Used to follow patients for functional loss.



Effective in predicting recovery of function in case in which intravenous urography has revealed nonvisualization.

Allows quantification of the degree of obstruction & can help differentiate the level of obstruction.

It is useful in children to rule out concomitant UPJO with associated high grade reflux.

CT SCAN & US

Obtained for any patient with acute flank pain.

Provide detailed anatomic & functional information.

Both US & CT have role in differentiating acquired causes of obstruction such as radiolucent calculi or Urothelial tumors.



In neonate & infants the diagnosis usually suggested either by routine performance of maternal US, or by finding of flank mass.

Differentiate UPJO from multicystic kidney & determine the level of obstruction.

RETROGRADE PYELOGRAPHY

Confirm the diagnosis & demonstrate the exact site & nature of obstruction before repair.

Indicated emergently when UPJO requires acute decompression in the setting of infection or compromised renal function.

✓ If cystoscopic retrograde manipulation unsuccessful or may be hazardous, particularly in neonates or infants, placement of **percutaneous nephrostomy** is preferred, which allows the performance of antegrade studies that will help define the nature and exact anatomic site of obstruction.

✓ **Diuretic renography is the first line of differentiating between UPJO & reflux.**

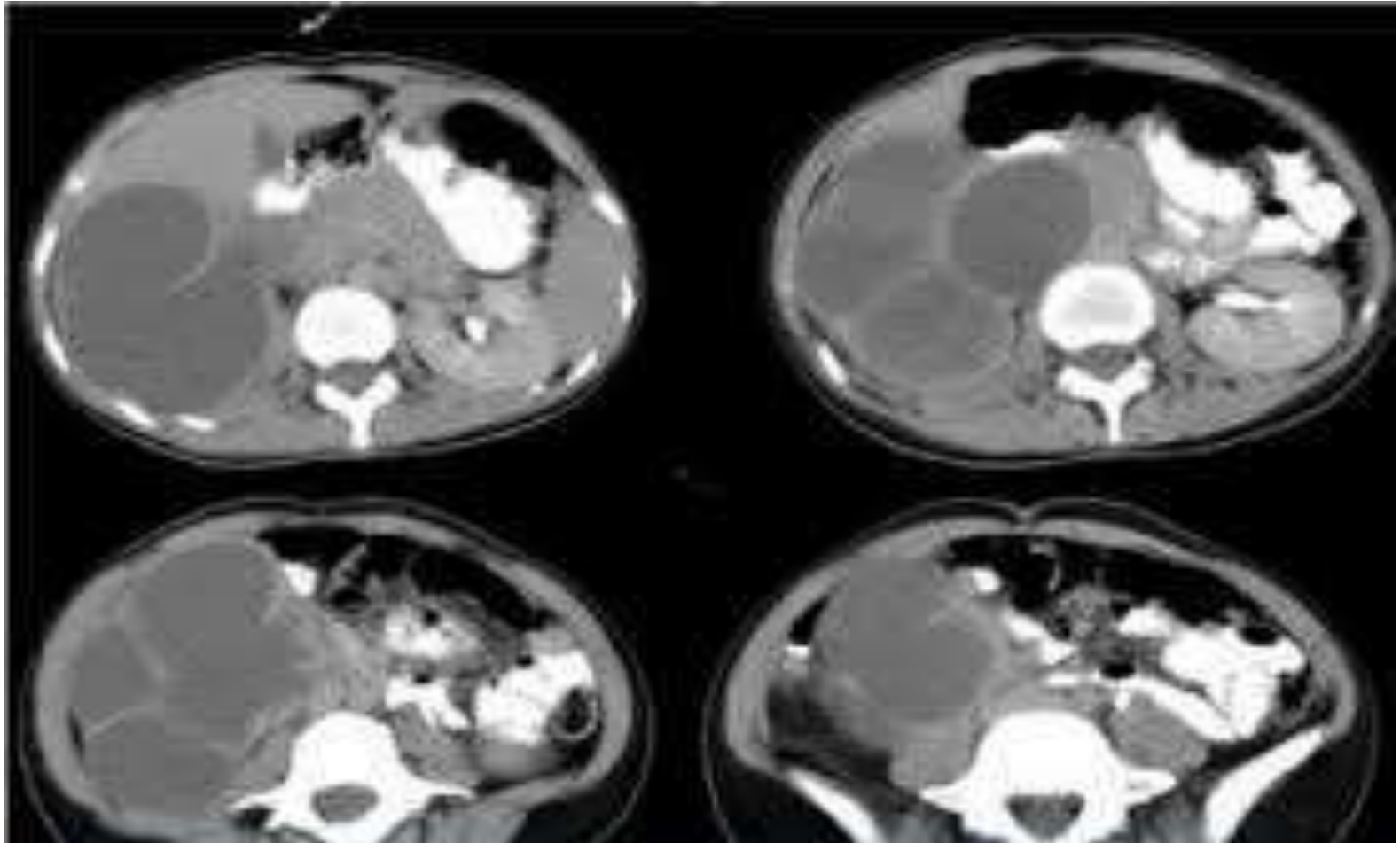
✓ “we must differentiate between vesicouretral reflux in children which lead to upper tract dilatation with subsequent elongation, tortuosity, & kinking of the ureter that mimic the radiographic finding of true UPJO.”

✓ **The diagnosis of UPJO basis on the clinical presentation & the result of any one or more of the imaging studies.**

PUJO FROM ACCESSORY RENAL ARTERY



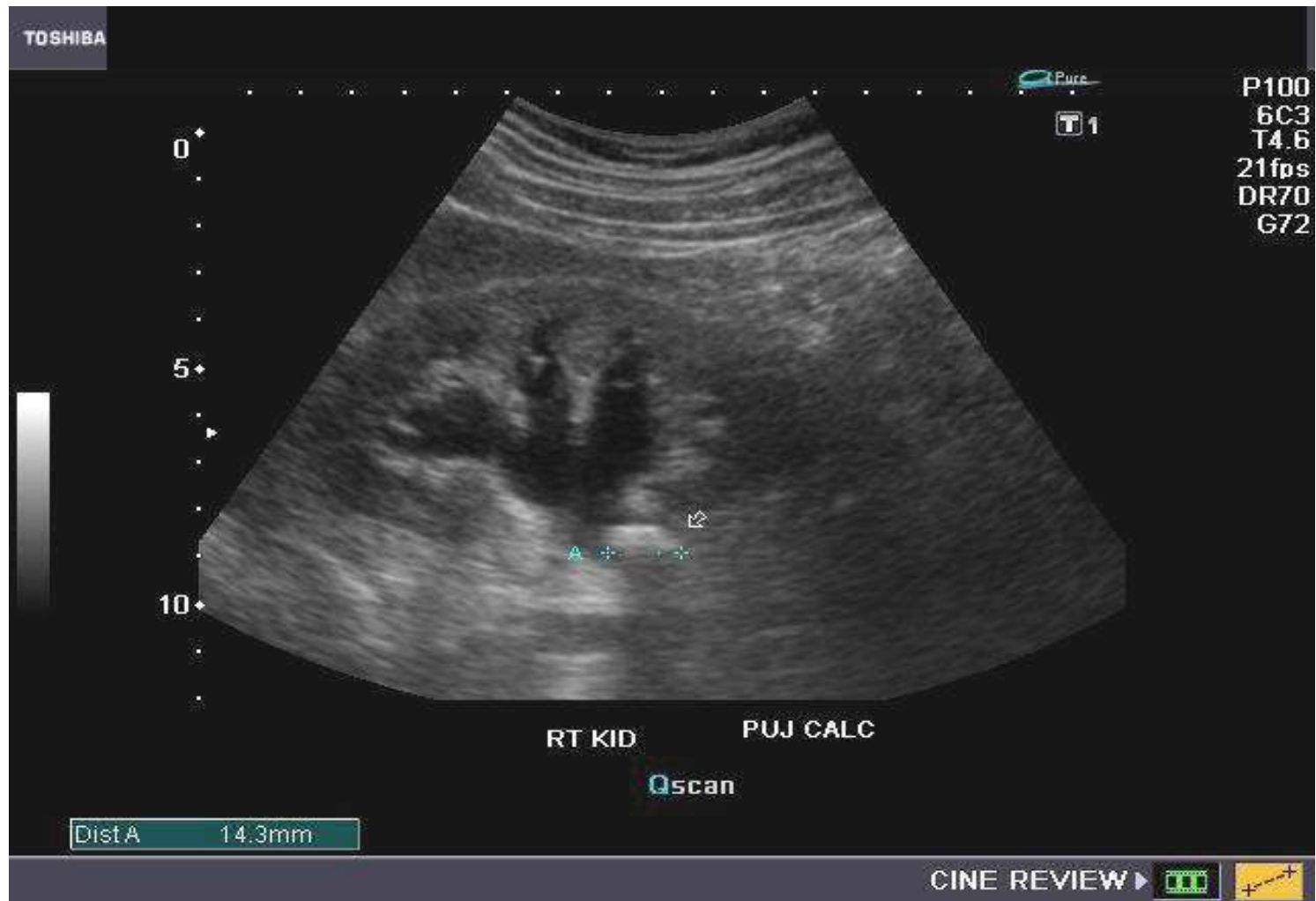
UPJO WITH ABDOMINAL MASS



PUJO IN 18 MONTHS OLD CHILD



PUJO CALCULUS US



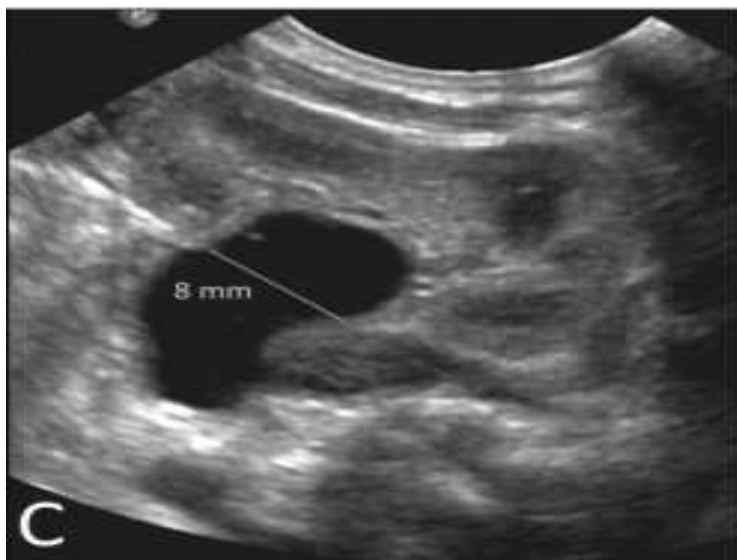
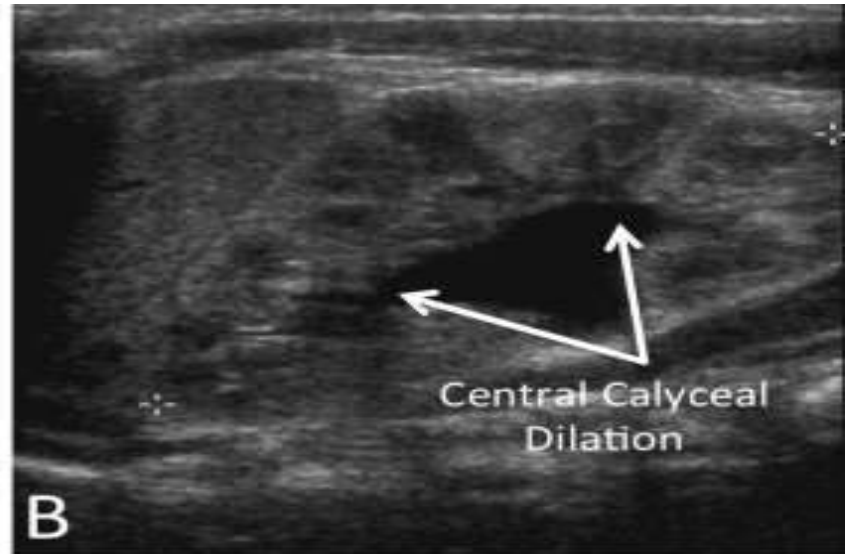
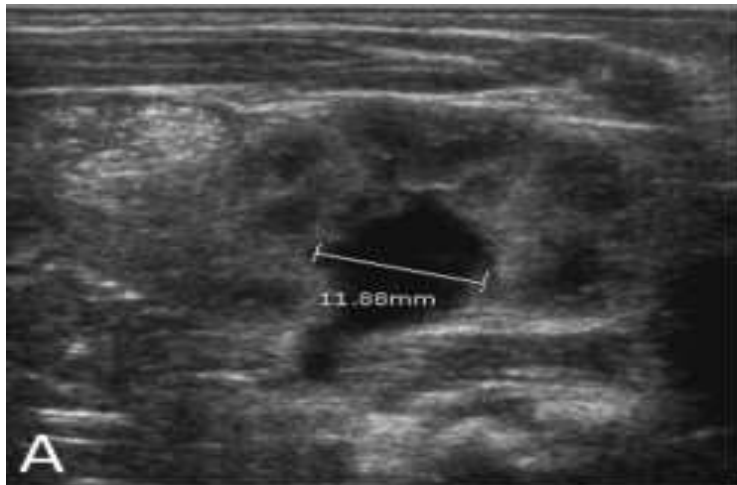
PUJO WITH CROSSING ABERRANT VEIN



PUJ OBSTRUCTION BY USS



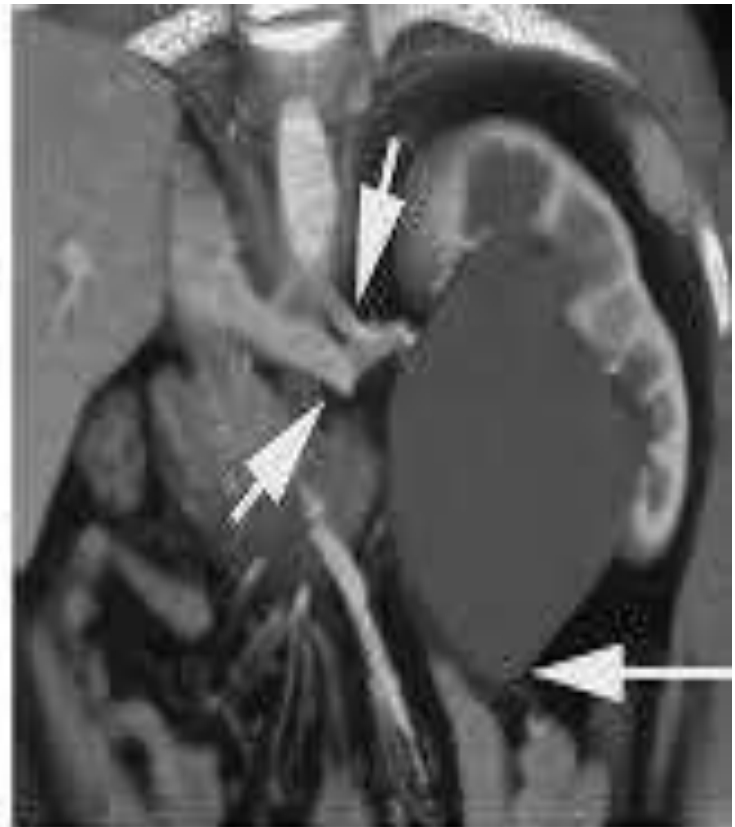
POST NATAL US



CT OF ADULT UPJO

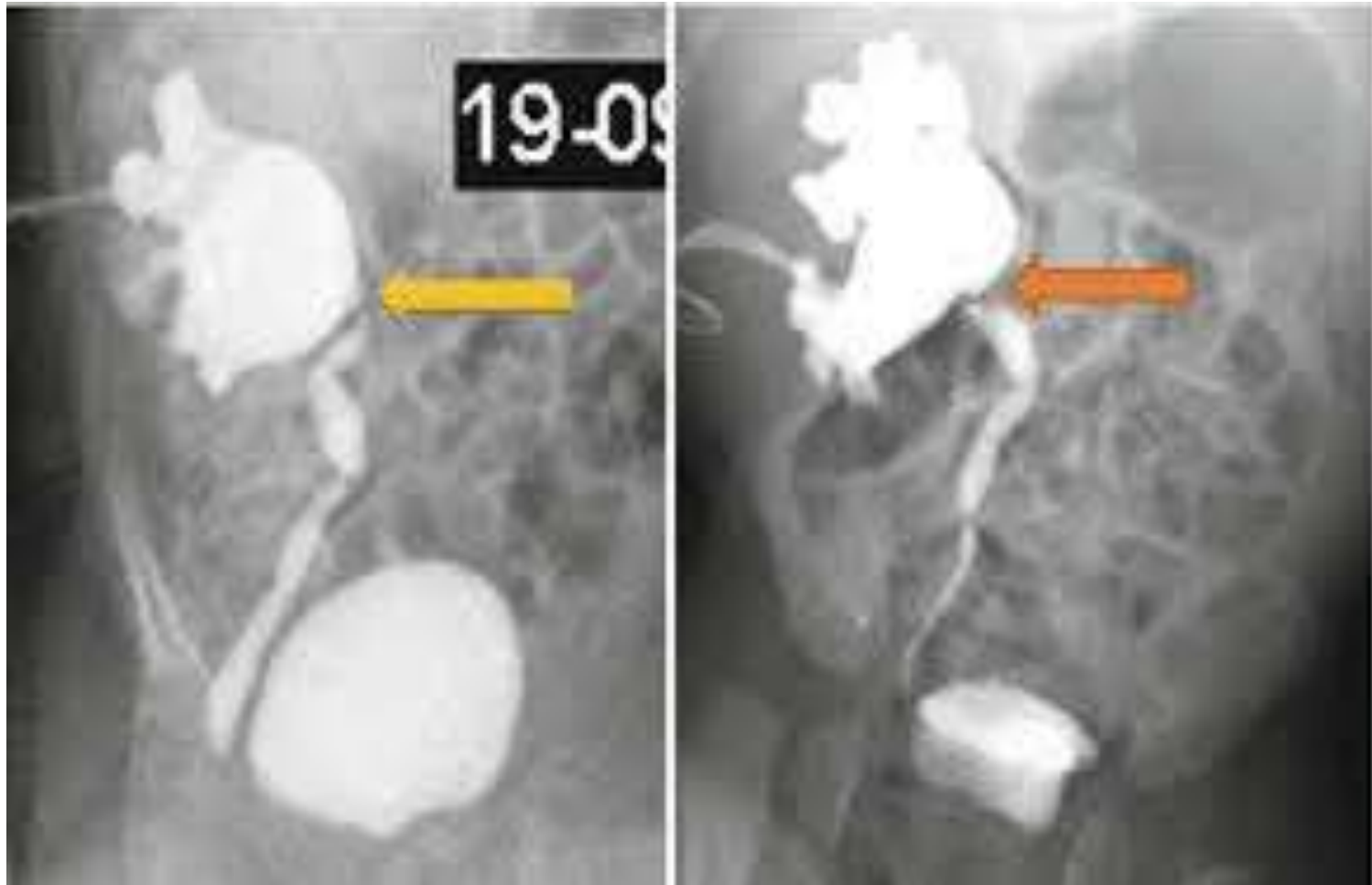


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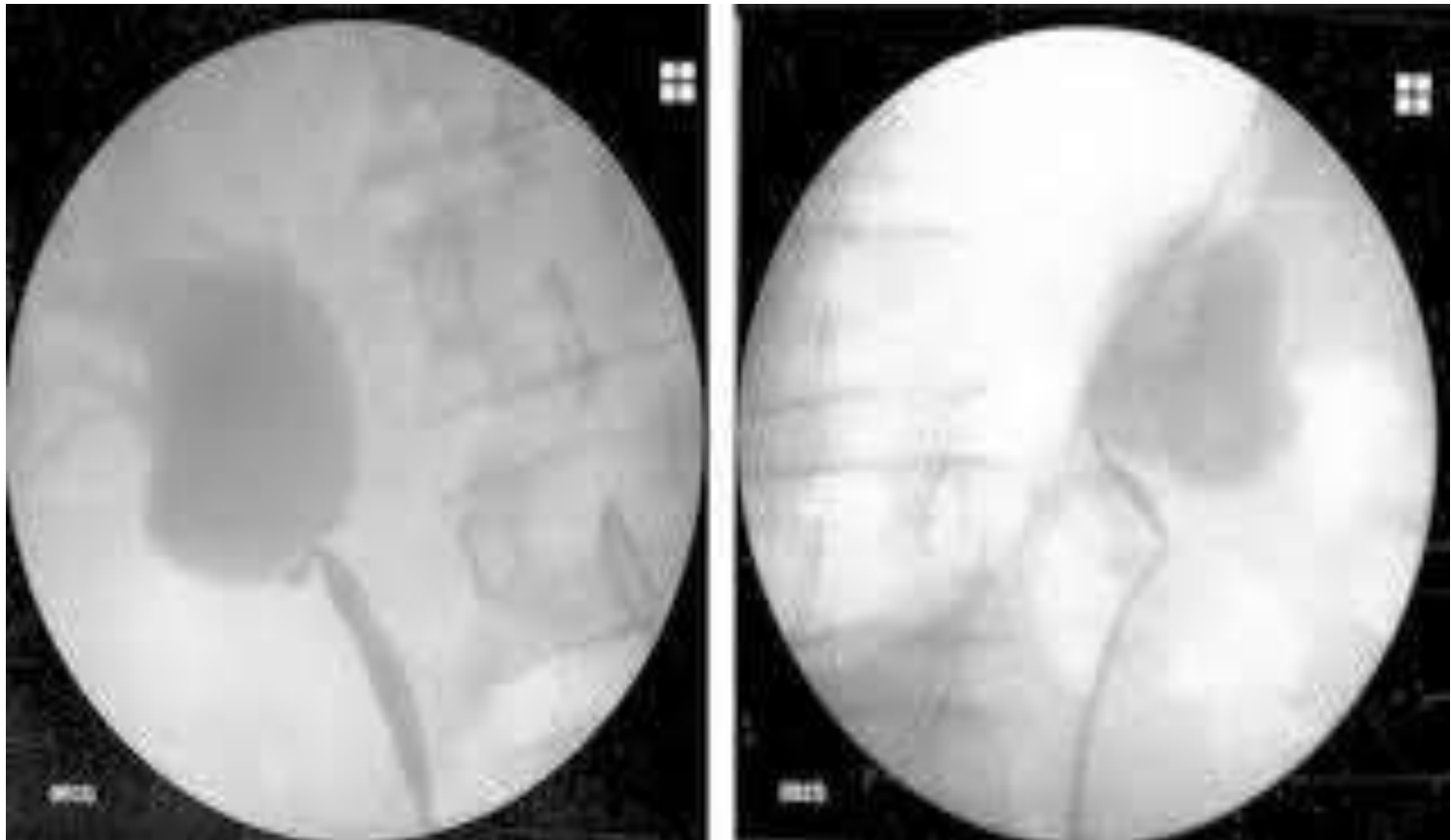


b.

PUJO PYELOGRAPHY



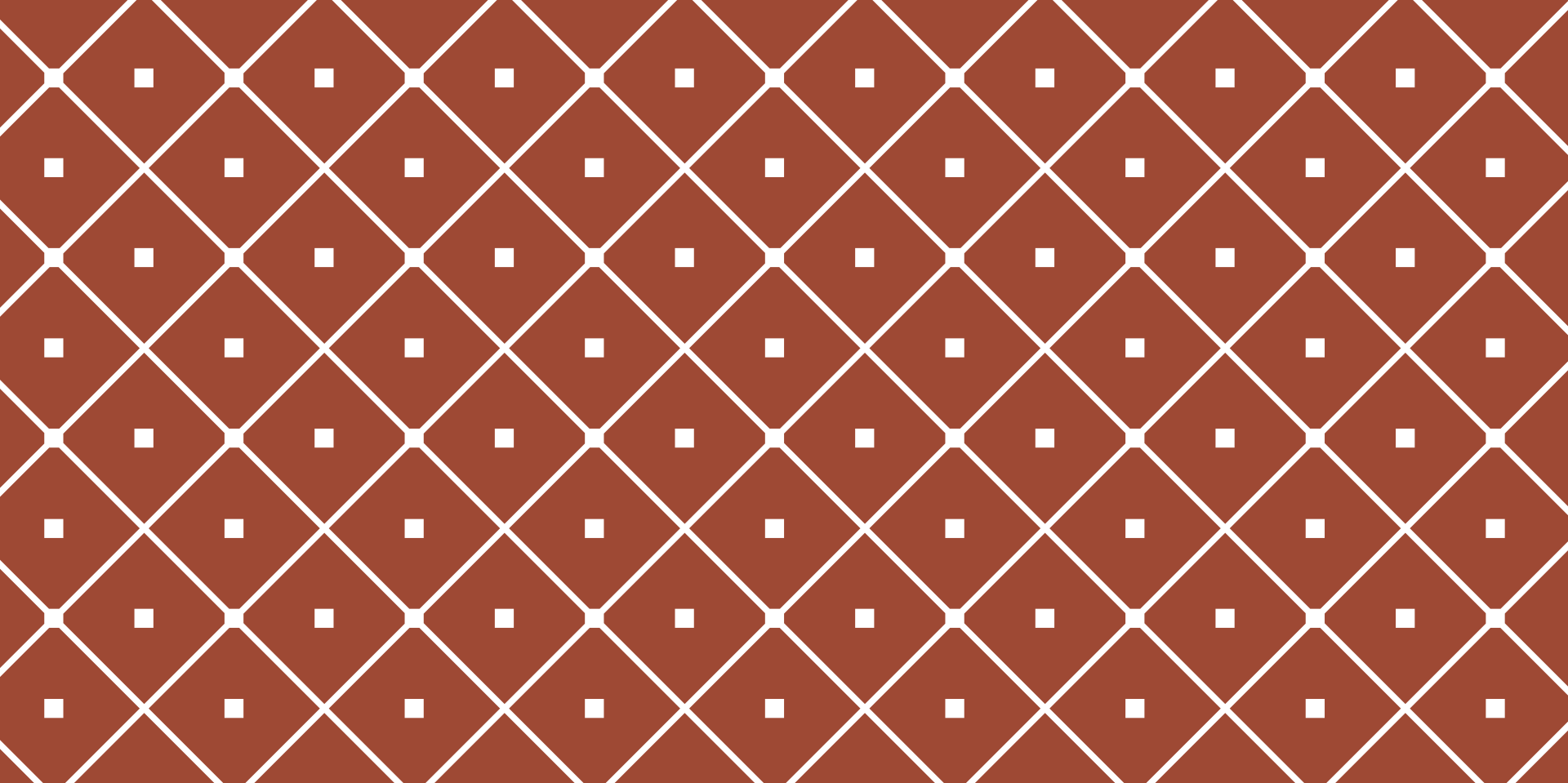
PUJO BY RETROGRADE PYELOGRAPHY



A) Intravenous pyelogram

B) CT show impacting of stone with sever hydronephrosis





PUV



It's a congenital membranes that cause obstruction to antegrade urinary flow in baby boys.

It's the most common cause of sever obstructive uropathy in male infants.

50% of patients has VUR.

CLASSIFICATION

Type 1:

- folds going inferiorly from verumontanum to membranous urethra.
- it's the most common type.

Type 2: folds radiating from verumontanum to bladder neck.

Type 3: valves are concentric diaphragms within the prostatic urethra.

CLINICAL FEATURES

Prenatally: Bilateral Hydronephrosis, full UB, Oligohydraminous.

Neonates: palpable and distended UB, weak stream of urine.

Infants: failure to thrive.

Children: incontinence, UTI.

DIAGNOSIS

US: hydronephrosis, full UB, Oligohydramnios.

VCUG:

- it's the best imaging technique for the diagnosis of PUV.
- also, for the diagnosis of associated VUR.

KEY HOLE SIGN



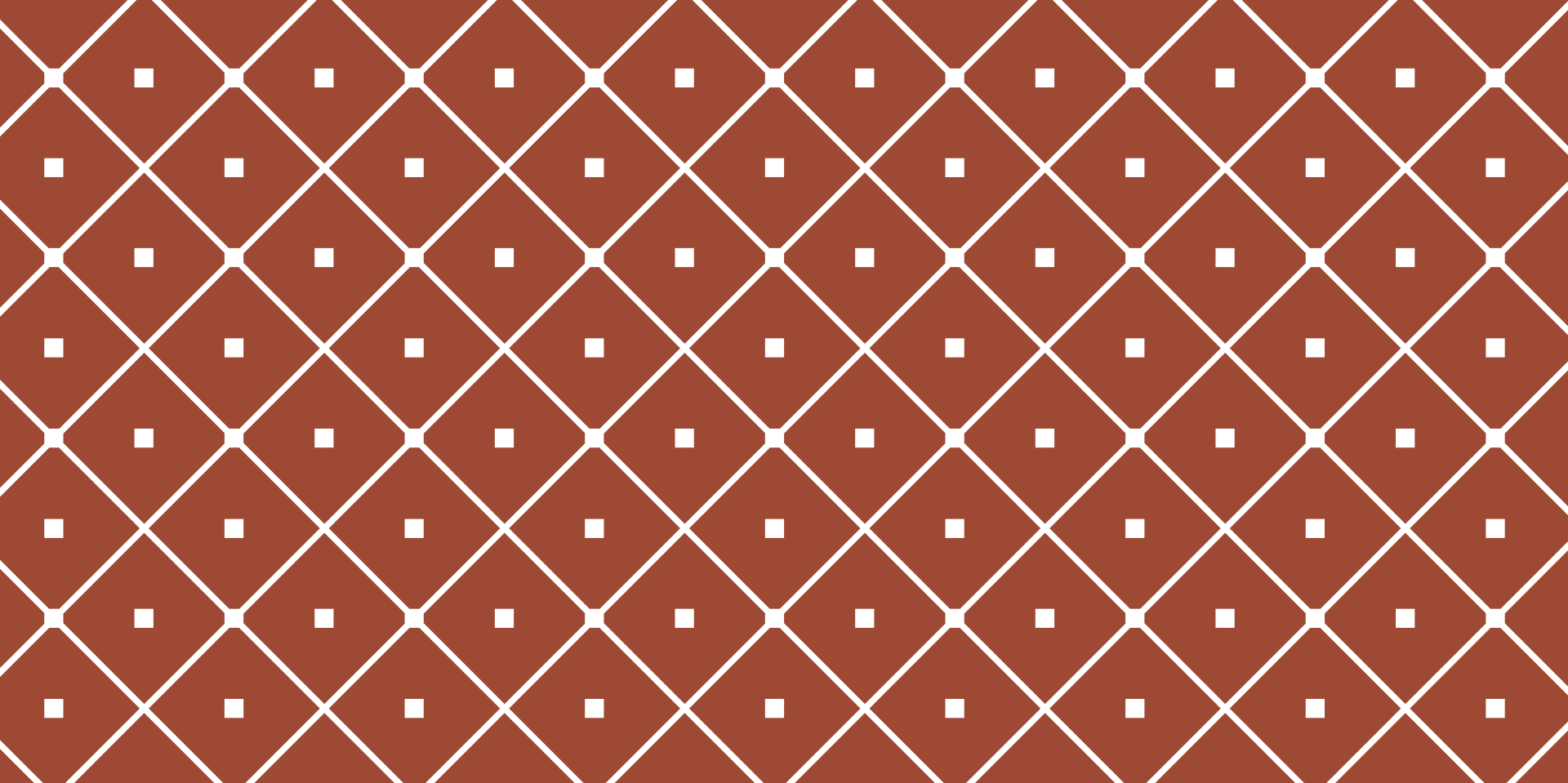
MANAGEMENT

Small feeding tube is inserted in the UB and left for several days. Then Transurethral ablation of the valve.

You have to rule out other associated obstruction if KFTs still high or increasing.

Vesicostomy:

- if the urethra is too small or KFTs remain high or increase despite drainage.
- it's closed after valve ablation.



MEGAURETER



Is descriptive one denoting dilatation of the ureter irrespective of cause.

Wide ureter & hydro ureter & in general refer to ureter that has diameter of **7mm** or greater.

CAUSES

Primary:

- representing condition intrinsic to the ureter itself.

Secondary:

- neurogenic bladder dysfunction.
- bladder outlet obstruction.
- and/or infection.



CLASSIFICATION

Obstructed.

Refluxing.

Non obstructed non refluxing.

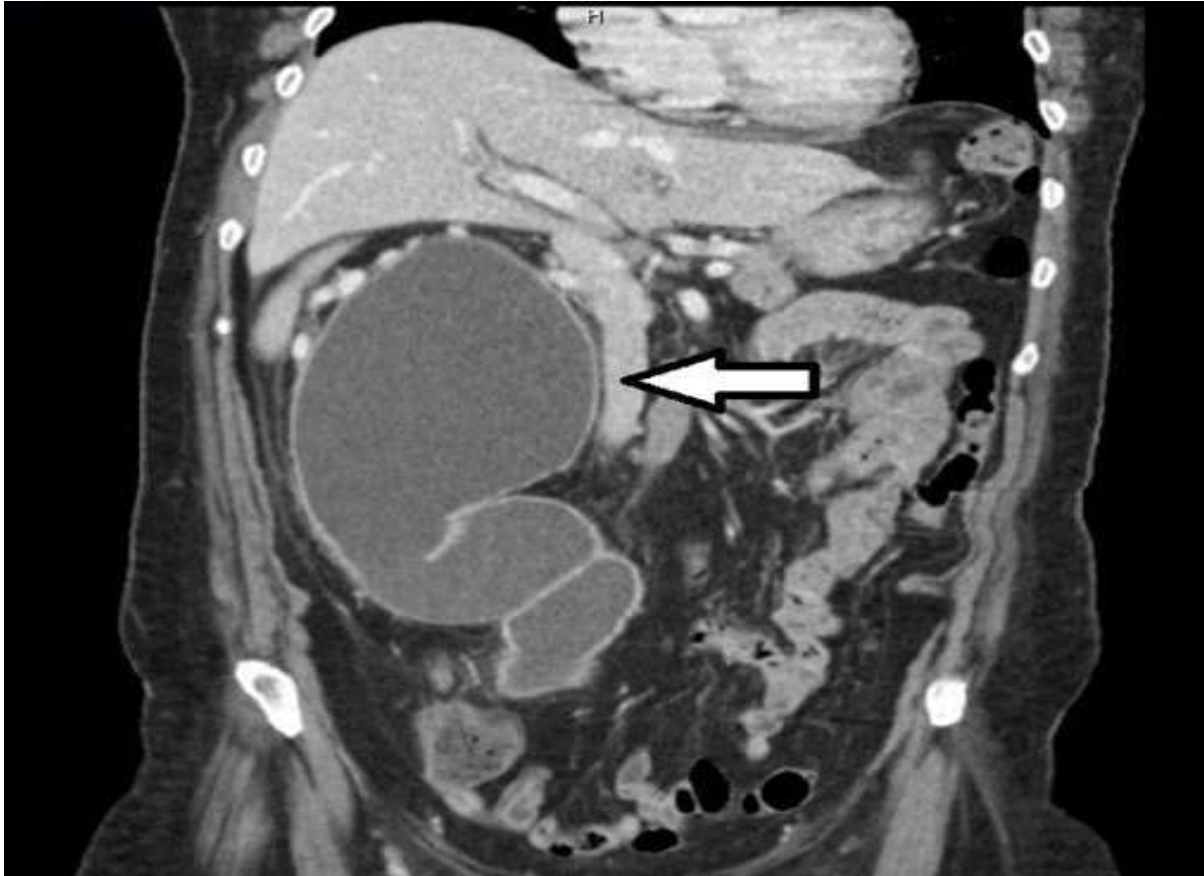
Refluxing with obstruction.

class	primary	secondary
Refluxing	Reflux is abnormality	Associated with bladder outlet obstruction or neurogenic bladder
Obstructing	Intrinsic ureteric obstruction, adynamic segment	Infravesical lesion, neurogenic bladder, extrinsic obstruction
Non-refluxing , non obstructed	Idiopathic ureteric dilatation	Polyuria ,infection
Refluxing , obstructing	Refluxing ureter without peristalsis in the distal part	Ectopic insertion at bladder neck (refluxes when relax & obstructs when tightened)

PRIMARY NON OBSTRUCTIVE MEGAURETER



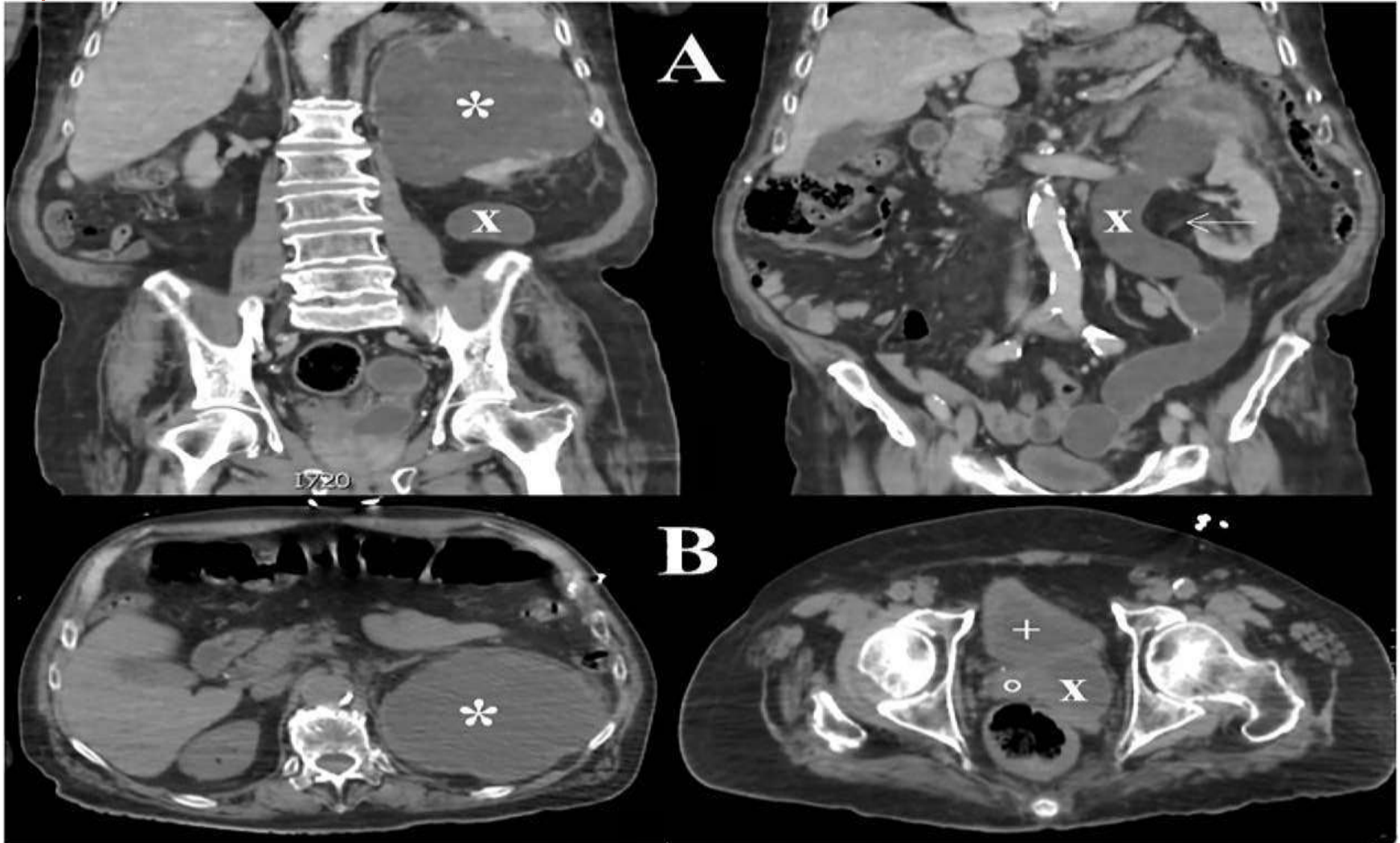
SECONDARY OBSTRUCTIVE GIANT MEGAURETER



PRIMARY NON REFLUXING MEGAURETER



ACUTE PYELONEPHRITIS REVEALING INTRAPROSTATIC OBSTRUCTIVE MEGAURETER



INCIDENCE

Anomaly has predilection for **boys & left side.**

It is bilateral in **25%** of cases & is associated with contralateral dysplasia or obstruction in **10% to 15%** of cases .

CLINICAL PRESENTATION

Mostly are detected by **antenatal** ultrasound screening.

The majority are **asymptomatic**.

Only **8%** of patients presenting symptomatically & found to have upper urinary tract dilatation were diagnosed with POM.



Symptoms include:

- **UTI**
- Abdominal **pain**.
- Microscopic **hematuria** even in absence of infection.
- Undiagnosed patients can present with abdominal **mass**.
- Signs & symptoms of **renal impairment**.

TREATMENT

The majority are benign in course & resolve **spontaneously** within the first few years of life while maintained on **antibiotic prophylaxis**.

Complete resolution & time to resolution were inversely related to ureteral diameter.



Surgery in the case of POM when:

- patients are symptomatic or have recurring UTI.
- progressive un remitting dilatation on U/S
- differential renal function less than 40%.

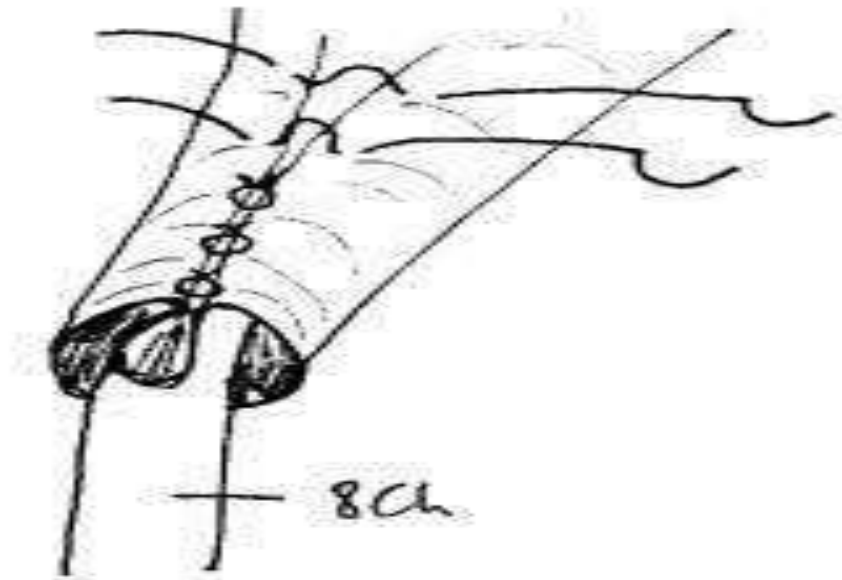
SURGERY

In summery, the stenotic distal part of the ureter is excised , the megaureter is straightened, then tapered to facilitate reimplantation in non refluxing fashion with adequate length to diameter ratio of **5:1** so as to improve coapation of the ureteral lumen whereby effective peristalsis & urine transportation are achieved.

Tailoring of the ureter can be achieved by one of two basic ways: plication or excisional tapering . With plication the megaureter is imbricated or folded around an **8-or 10-Fr** catheter , depending on patient age.

STARR PLICATION

Ureteral plication is performed over the appropriate catheter , with interrupted 5-0 monofilament resorbable sutures placed in Lembert clamp.



KALICINSKI FOLDING

The blood supply to the ureter is preserved which decrease the risk of ischemia & stenosis.

Suitable only for the moderately dilated ureter (< **1.75** cm in diameter).

Good results have been reported with folding techniques with success rates of **90% to 95%**.

EXCISIONAL TAPERING

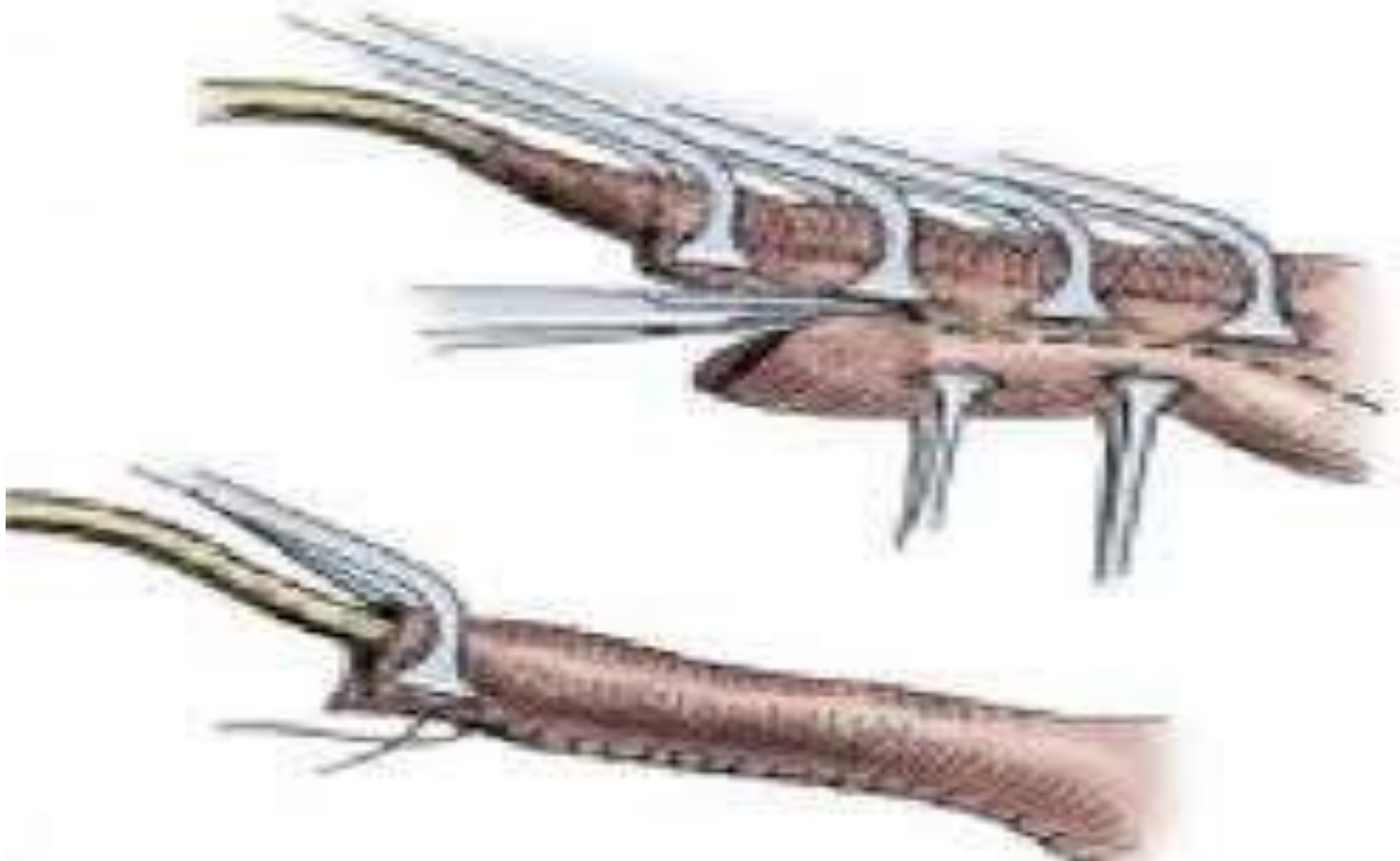
The forerunner for the later modifications of folding techniques, because it is thought & has been shown that excisional tapering may jeopardize ureteral vasculature with all its attendant complications.



Procedure has advantages especially when dealing with massively dilated ureter that are not amenable to folding , & when performed with care the risk of vascular compromise should be minimal.

Success rate excellent.

Fig. 10.10.1. Intraoperative view of the right



Tapering of megaureter



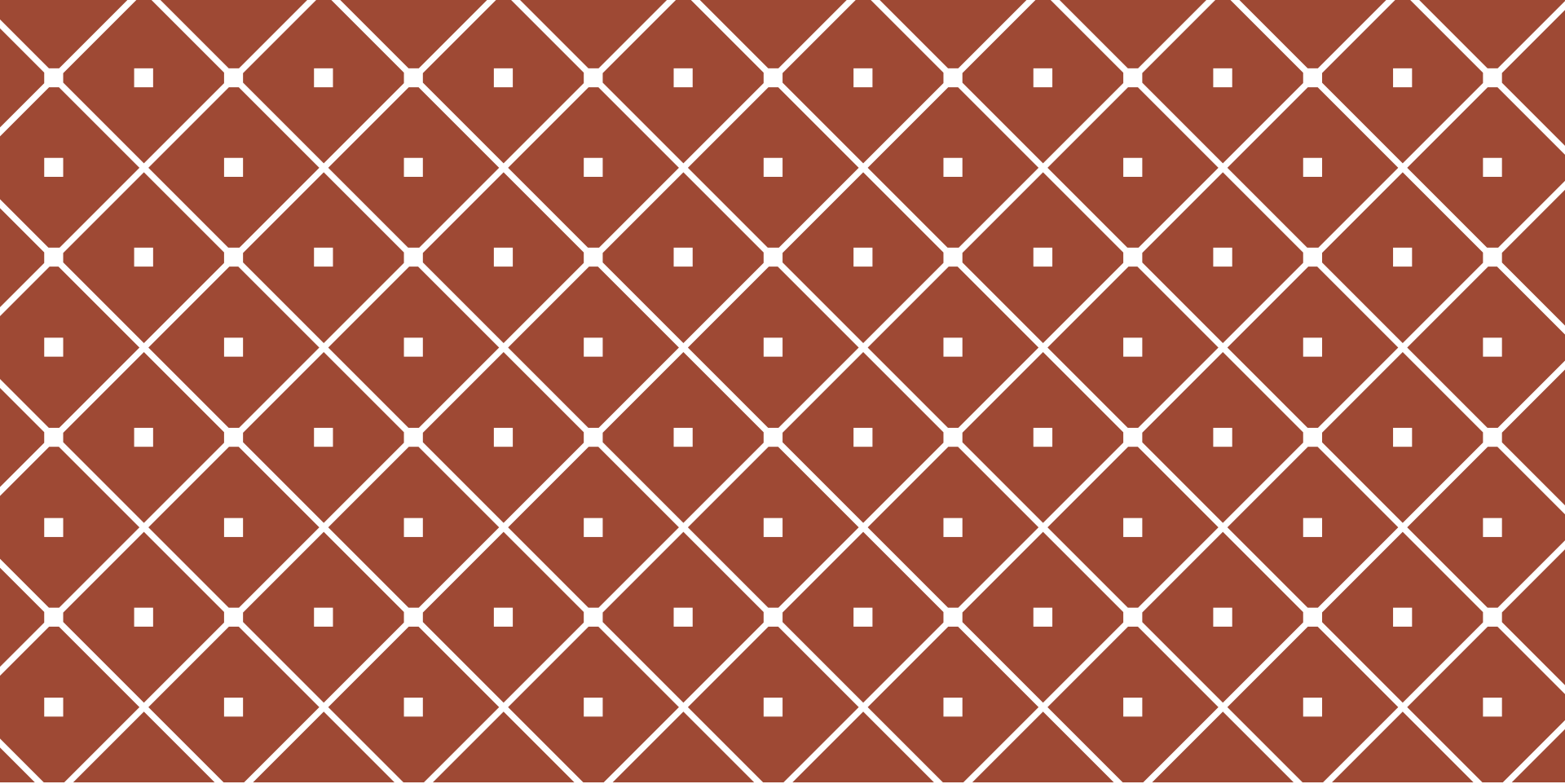
COMPLICATIONS

Obstruction which managed by stenting.

VUR, conservative management.

Persistent dilatation.

Ischemic stricturing especially after excisional tapering.



ANOMALIES OF THE UPPER URINARY TRACT



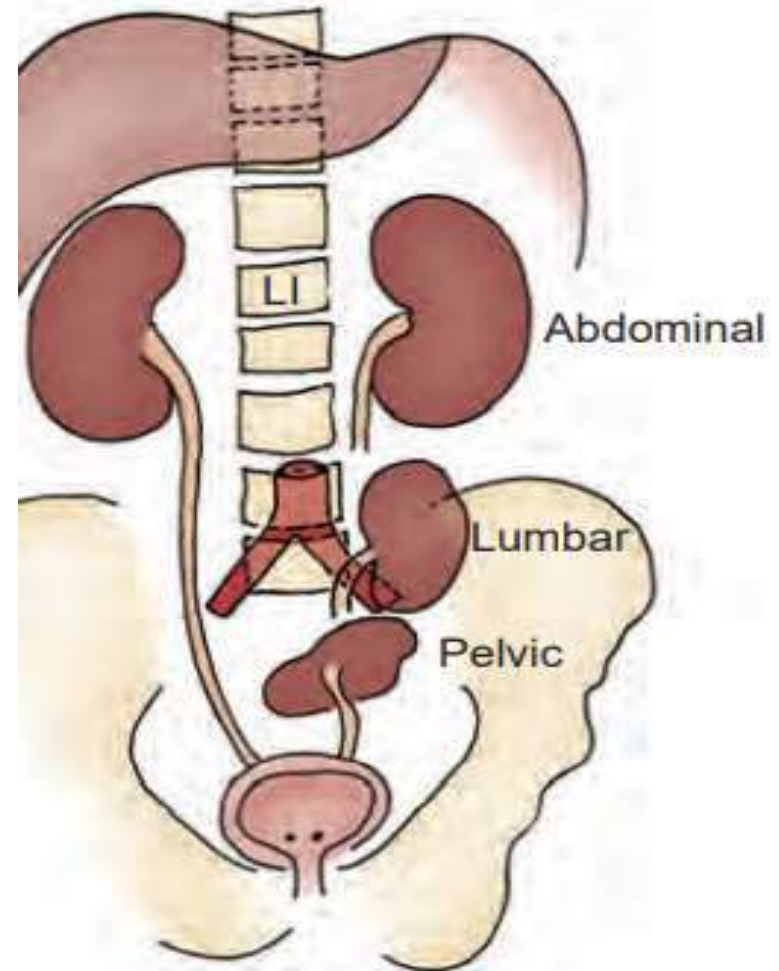
SIMPLE RENAL ECTOPIA

An ectopic kidney can be found in one of the following positions: pelvic, iliac, abdominal, thoracic, and contralateral or crossed.

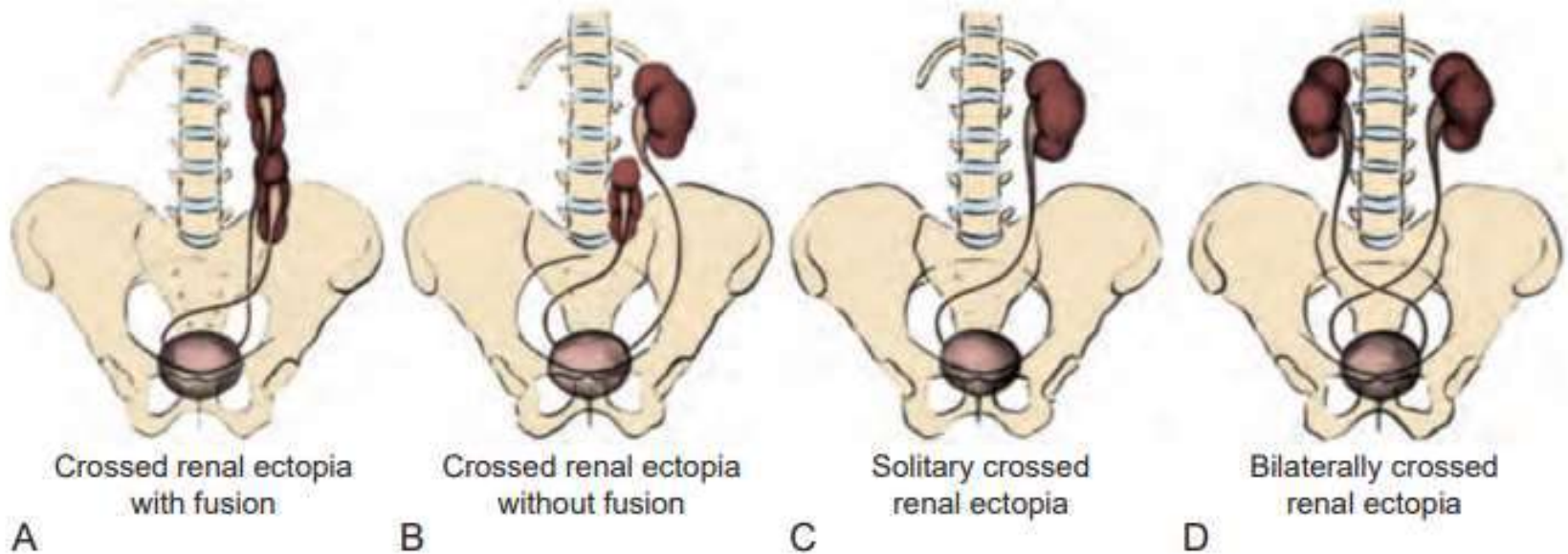
Average occurrence of about 1 in 900 with no significant difference between the sexes.

The most striking feature of renal ectopia is the association of **genital anomalies**. The incidence varies from **15% to 450%**

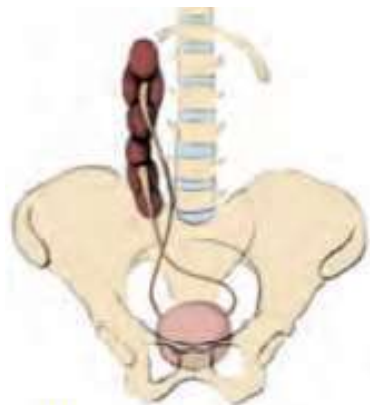
Most ectopic kidneys are **asymptomatic**.



CROSSED RENAL ECTOPIA WITH AND WITHOUT FUSION



SIX FORMS OF CROSSED RENAL ECTOPIA WITH FUSION.



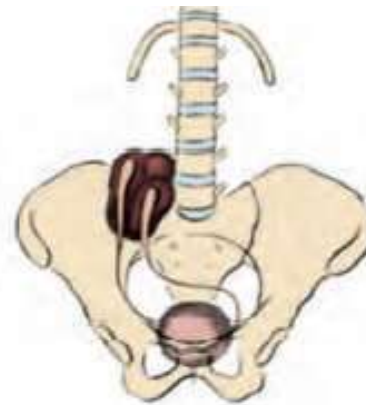
Unilateral fused kidney
(inferior ectopia)

A



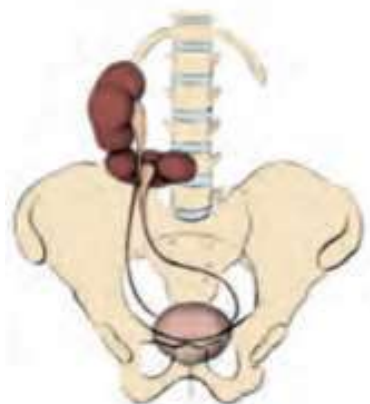
Sigmoid or
S-shaped kidney

B



Lump kidney

C



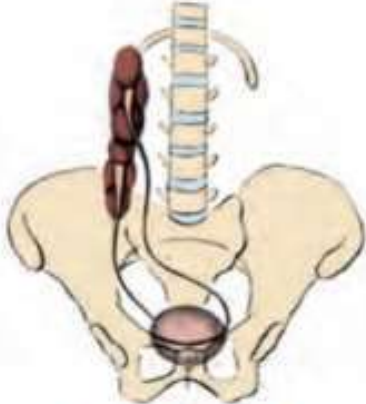
L-shaped kidney

D



Disc kidney

E



Unilateral fused kidney
(superior ectopia)

F



DUPLEX RENAL DISEASE

Partial or total:

- Bilateral **40%**
- **0.1%** are complete **F>M**

The majority are **asymptomatic**.

Single ureteric bud >>> incomplete duplication

2 ureteric buds >>> complete duplication

- the upper moiety inserts distal in bladder or can be in urethra.
- the lower moiety inserts supero-lateral to trigone.

The **lower moiety reflux**.

The **upper moiety obstructs** with hydronephrosis and often dysplastic kidney and a **ureterocele** in bladder.

CLINICAL PRESENTATION

May be seen on **neonatal** scans.

May present with recurrent **pyelonephritis**.

May present with **incontinence**.

Investigations include: **USS, MCUG, CT**.

HORSESHOE KIDNEY

The horseshoe kidney is the most common of all renal fusion anomalies.

Horseshoe kidney occurs in **0.25%** of the population, or about **1 in 400** persons.

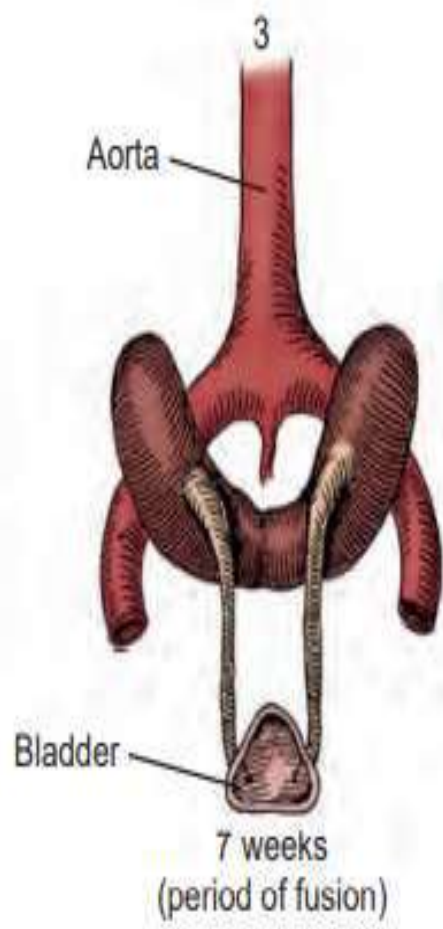
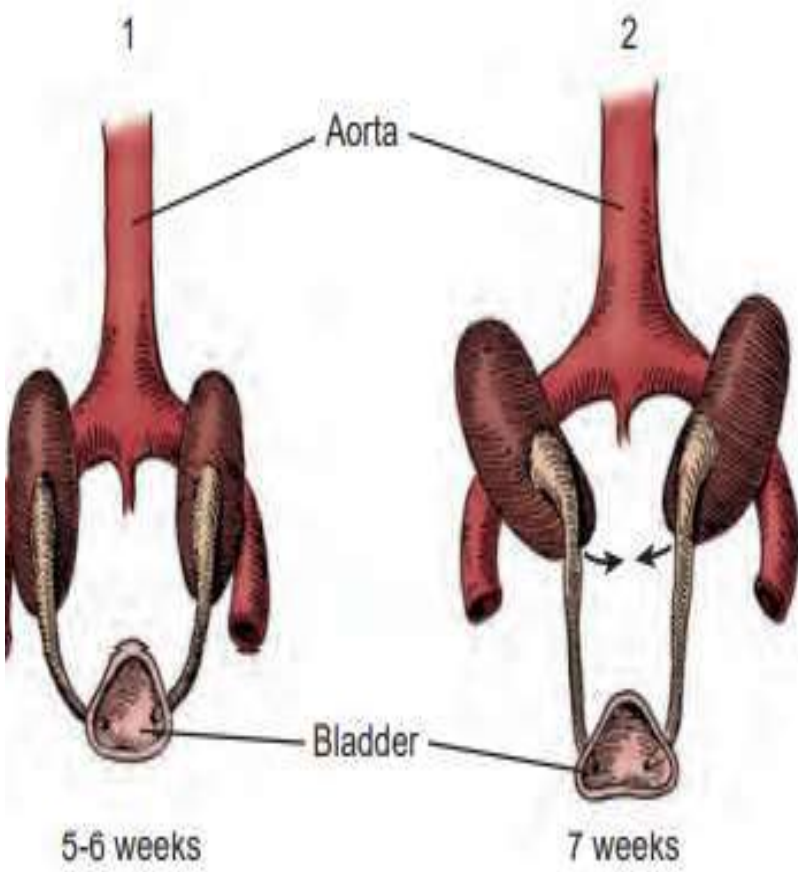
In **95%** of cases, the kidneys join at the lower pole.

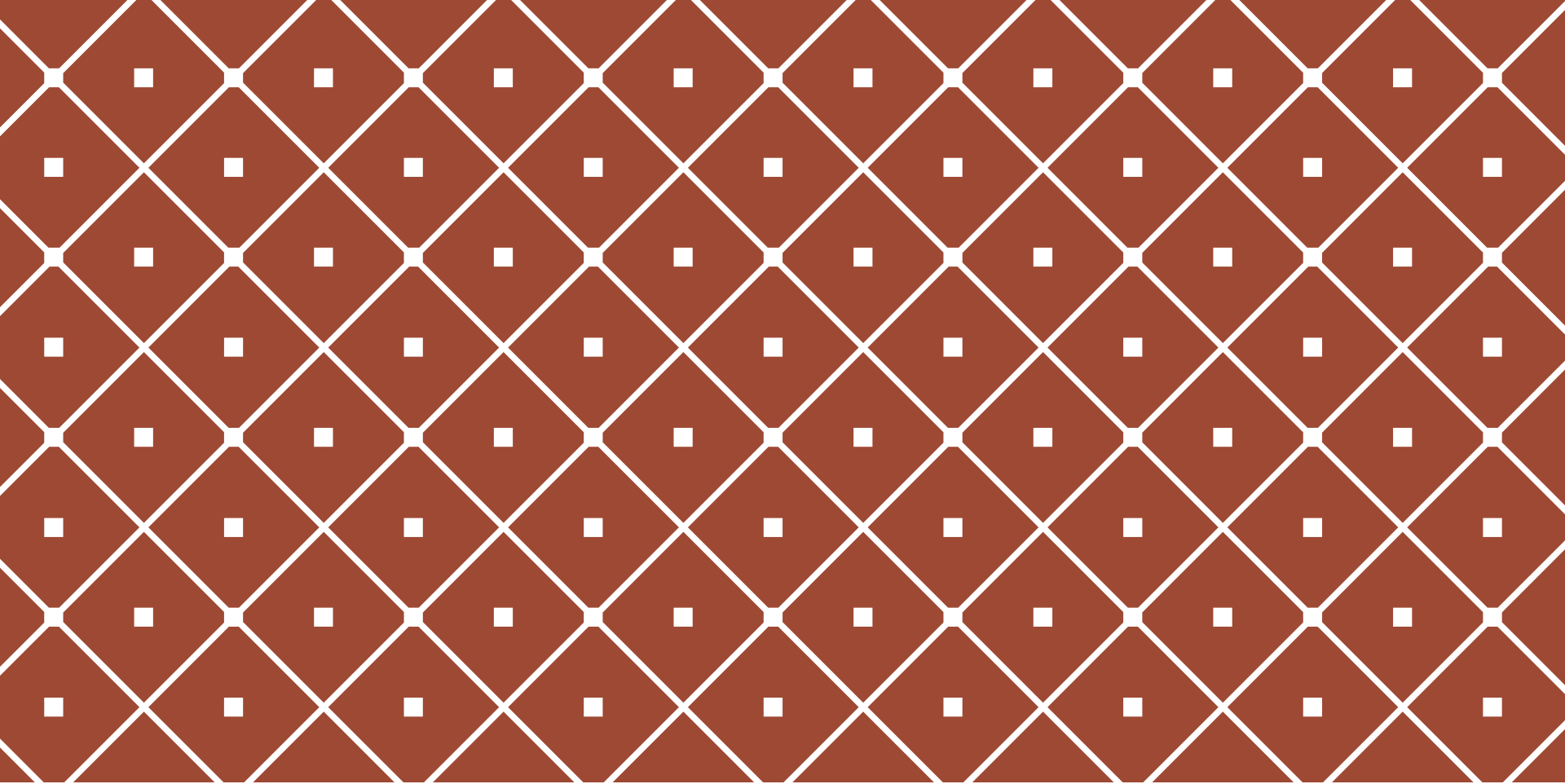
Duplication of the ureter occurs in **10%** of cases.

Vesicoureteral **reflux** has been noted in more than **half** .

UPJ obstruction occurs in one **third**.

Stone formation has been commonly seen in horseshoe kidney.





URETEROCELE



DEFINITION

A ureterocele is a cystic dilatation of the distal ureter, as it drains into the bladder.

INCIDENCE

One in **5000–12000** clinical paediatric admissions.

The ♀:♂ ratio is **4:1**.

predominantly affecting **Caucasians**.

10% of ureterocele are **bilateral**.

CLASSIFICATION

Ureteroceleles form due to a failure of normal perforation of **Chawalla's membrane**, which separates the ureter and bladder.

80% are associated with the **upper moiety** of a duplex kidney.

They are further classified into **intravesical** or **extravesical** ureteroceleles.



Intravesical (20%):

the ureterocele is completely confined within the bladder.

These tend to be associated with single systems and are commoner in ♂.

Subtypes include: **Stenotic** and **Non-obstructed**.



Stenotic:

small, stenotic ureteric orifice associated with obstruction.

Non-obstructed:

large ureteric orifice that tends to balloon open
when filled by peristalsis of urine.



Extravesical (or ectopic) (80%):

when the ureterocele extends to the bladder neck or urethra and tend to occur with duplex systems most commonly in ♀.

Subtypes include: **Sphincteric, Sphincterostenotic, Cecoureterocele, Blind ectopic.**



Sphincteric:

ureterocele extends into the bladder neck and urethra.

The orifice is wide and usually opens proximal to the external sphincter.

Sphincterostenotic:

similar to a sphincteric ureterocele, but the ureteric orifice is stenosed.



Cecoureterocele:

ureterocele prolapses posterior to the urethra and anterior to the vagina, but the orifice is within the bladder (affects girls only). Can cause urethral obstruction.

Blind ectopic:

similar to sphincteric, but no ureteric orifice.

PRESENTATION

Most present with **antenatal hydronephrosis**.

Later presentation in infants may be with symptoms of **UTI**, an abdominal **mass**, or **pain**.

Association with ureteric duplication increases the risk of reflux and reflux nephropathy.



Extravesical ureterocele can also cause **BOO** and bilateral hydroureteronephrosis (urological emergency) or ureteric obstruction and unilateral hydroureteronephrosis, which require urgent assessment and intervention.

A prolapsing ureterocele can present as a congested **purple mass** at the introitus in girls.

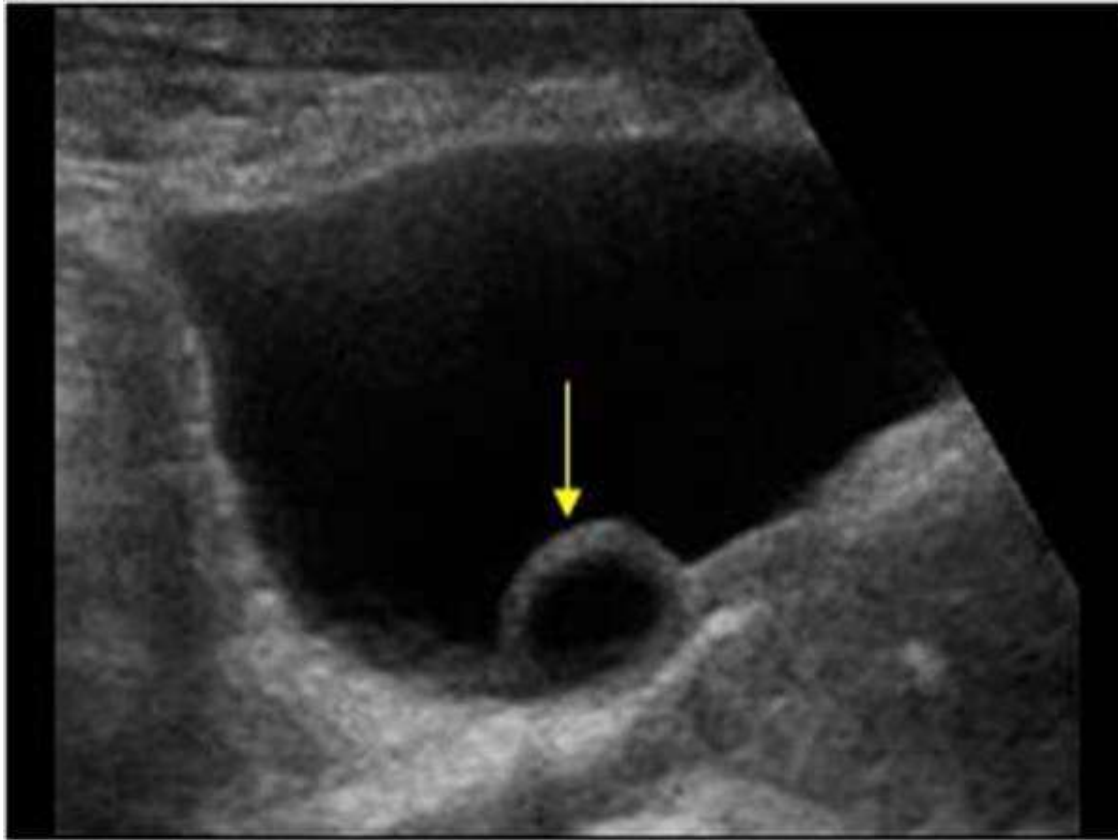


INVESTIGATION

USS :

shows a **thin-walled cyst** in the bladder, often associated with a **duplex system** and an **ectopic (dilated) ureter**.

If there are concerns about obstruction, USS should be performed immediately after birth, with a view to urgent surgical treatment.

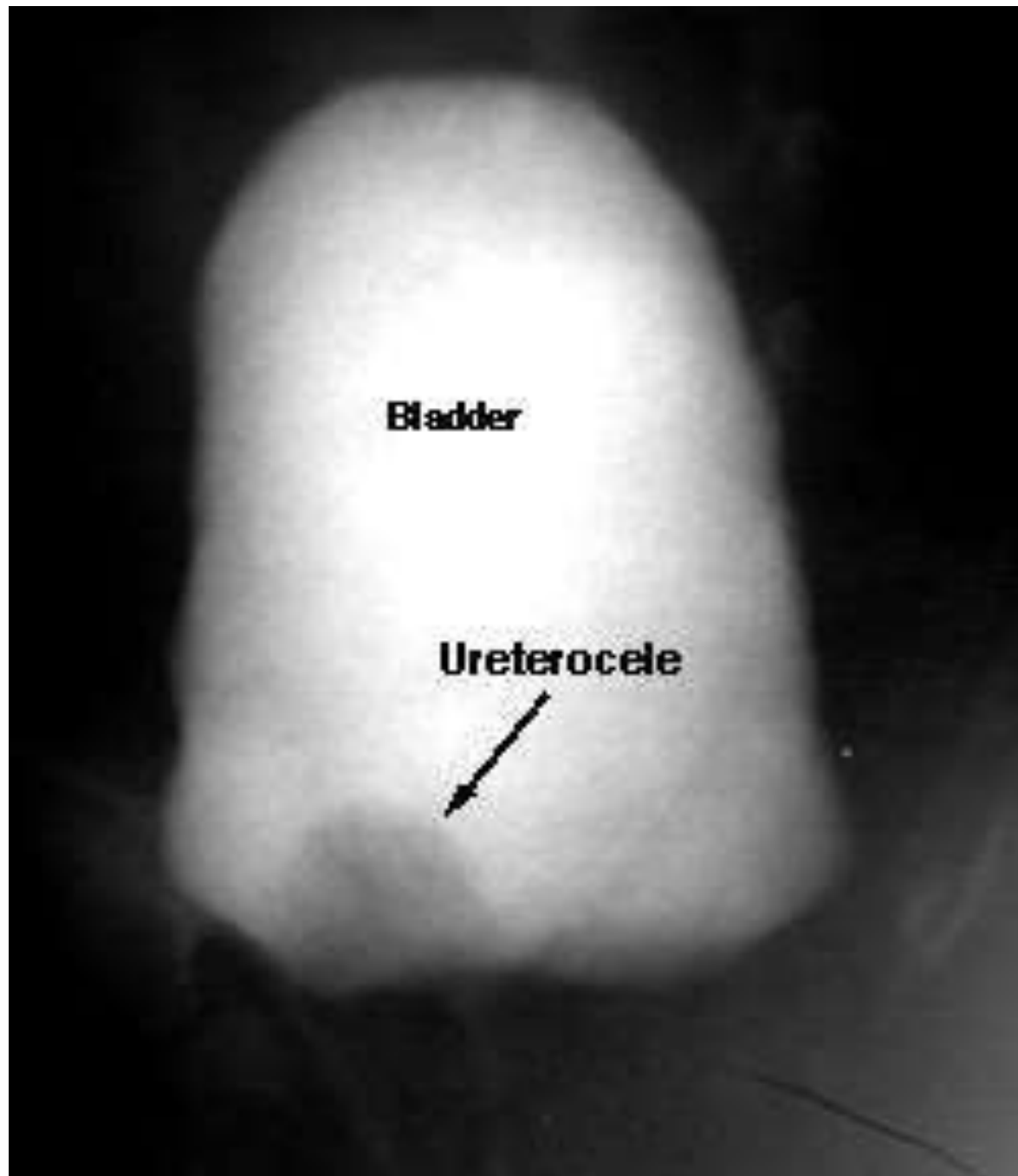


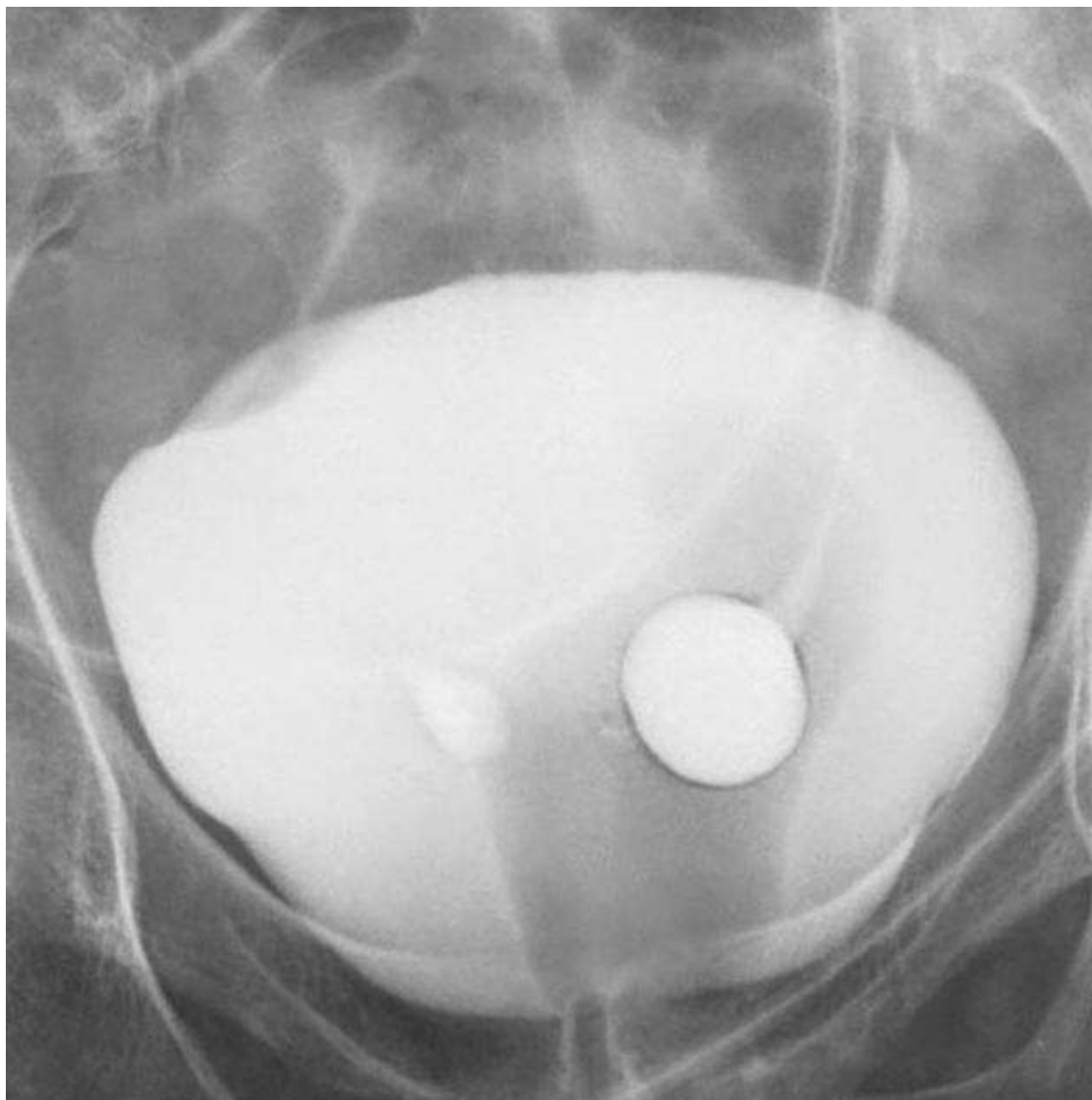


MCUG:

can identify ureterocele **location** and **size** and associated **VUR** (reflux into the lower moiety of an associated duplex kidney is seen in **50%**).

This should be performed early in the postnatal period if there is evidence of BOO; otherwise, defer 3–6 months.







MAG3 renogram:

is used to exclude **obstruction**.

DMSA renogram:

is used to assess renal moiety **function** and demonstrate renal cortical abnormalities in the presence of reflux.





Cystoscopy:

can be used for diagnosis and endoscopic treatment.



TREATMENT

Commence **prophylactic antibiotics** at birth (**trimethoprim 2mg/kg daily**).

Urgent surgical intervention is required for obstruction.



Endoscopic incision/puncture:

emergency treatment for infected or obstructed ureterocele.

Puncture is also indicated for elective management of intravesical ureteroceles with normal renal function.

rarely, these may require further surgery to preserve renal function and prevent reflux.



Uretero-ureterostomy or uretero-pyelostomy (from upper to lower pole moiety):

option for ectopic ureteroceleles associated with a duplex system, with good function in the upper moiety and no reflux in the lower moiety.



Upper pole heminephrectomy:

option for ectopic ureterocele associated with a duplex system with poor function in the upper moiety and no reflux in the lower moiety.



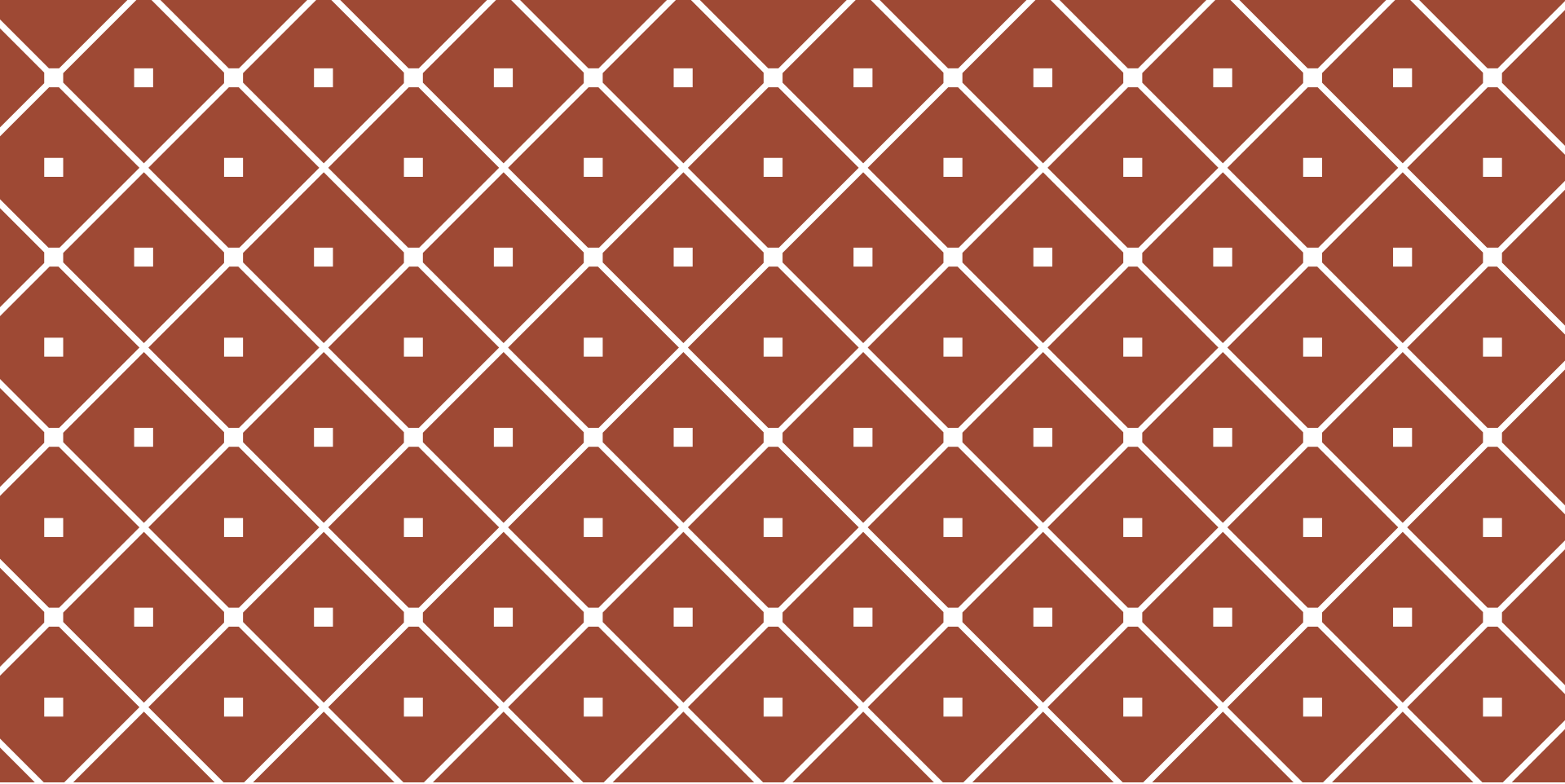
Upper pole heminephrectomy, ureterocele excision, and ureteric reimplantation:

option for ectopic ureterocele associated with a duplex system with poor function in the upper moiety and reflux in the lower moiety.



Nephroureterectomy:

indicated for significant lower moiety reflux with poor function in both renal moieties or for poor renal function in a single system.



CYSTIC DISEASES OF THE KIDNEY



The kidney is one of the most common sites in the body for cyst formation.

broad spectrum of sporadic and genetically determined congenital or acquired conditions that have the presence of cysts in one or both kidneys.



Renal cysts are cavities derived primarily from tubules and are composed of a layer of partially dedifferentiated epithelial cells enclosing a cavity filled with urine-like liquid or semisolid material.



Multicystic dysplasia arises before formation of the nephron, from abnormal induction of metanephric development, or from obstruction occurring early in renal development.

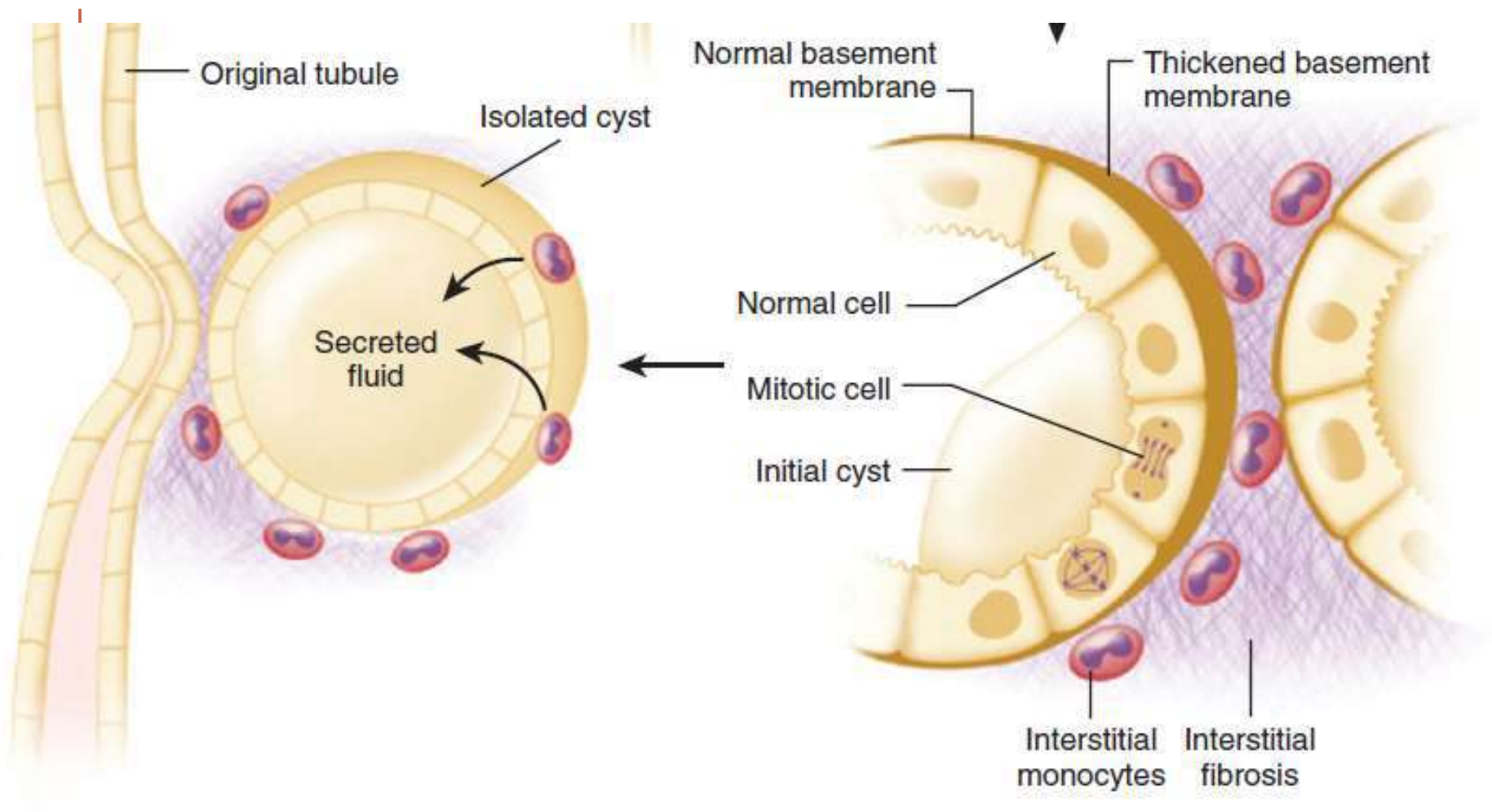
Multilocular cyst, represents a neoplastic growth.

Proliferation of epithelial cells in segments of renal tubule



Accumulation of fluid within the
expanding tubule segment,

Disturbed organization and metabolism
of the **extracellular matrix**



CLASSIFICATION

Multicystic typically refers to a dysplastic kidney resulting from aberrant renal development.

Polycystic refers to renal units that developed in a normal fashion

Inheritable

Autosomal recessive (infantile) polycystic kidney disease

Autosomal dominant (adult) polycystic kidney disease

Juvenile nephronophthisis/medullary cystic disease complex

Juvenile nephronophthisis (autosomal recessive)

Medullary cystic disease (autosomal dominant)

Congenital nephrosis (familial nephrotic syndrome) (autosomal recessive)

Familial hypoplastic glomerulocystic disease (autosomal dominant)

Multiple malformation syndromes with renal cysts (e.g., tuberous sclerosis, von Hippel-Lindau disease)



Noninheritable

Multicystic kidney (multicystic dysplastic kidney)

Benign multilocular cyst (cystic nephroma)

Simple cysts

Medullary sponge kidney

Sporadic glomerulocystic kidney disease

Acquired renal cystic disease

Calyceal diverticulum (pyelogenic cyst)

INFANTILE POLYCYSTIC KIDNEY DISEASE

The most severe forms appearing earliest in life.

Up to age **13** years or, rarely, up to age **20** years.

The reported incidence of ARPKD varies from **1 in 10,000 to 50,000** live births.

Of those infants who survive the neonatal period, approximately **50%** are alive at 10 years of age.

Rapid, symmetrical, and bilateral enlargement of the kidneys in **infants**.

It is invariably associated with some degree of congenital **hepatic fibrosis**.

The disease can present in adolescents

GENETICS

Because the disease is transmitted as an **autosomal recessive** trait, siblings of either sex have a **1 in 4** chance of being affected.

Mutations of a single gene, named **PKHD1** and located on chromosome 6 (**6p12**).

The gene produces a protein called

CLINICAL FEATURES

In utero: enlarged, echogenic kidneys. Oligohydramnios is common.

In infant: Potter faces and deformities of the limbs and may have respiratory distress due to pulmonary hypoplasia.









In newborn: usually has enormous, kidney-shaped, nonbosselated flank masses that are hard and do not transilluminate.

Thirty to 50 percent die shortly after birth as a result of uremia or respiratory failure

All patients with ARPKD have congenital hepatic fibrosis and vary in the degree of **biliary ectasia** and **periportal fibrosis**.

The earlier the age at which the disease is identified, the more severe the disease.

Hypertension and renal insufficiency are the major manifestations.

liver disease becoming more prevalent in older patients.

They develop renal failure and hypertension more slowly due to liver

hepatic fibrosis leading to **portal hypertension, esophageal varices, and hepatosplenomegaly**

Hepatocellular function is rarely abnormal.

The kidneys are often normal in appearance.

HISTOPATHOLOGY

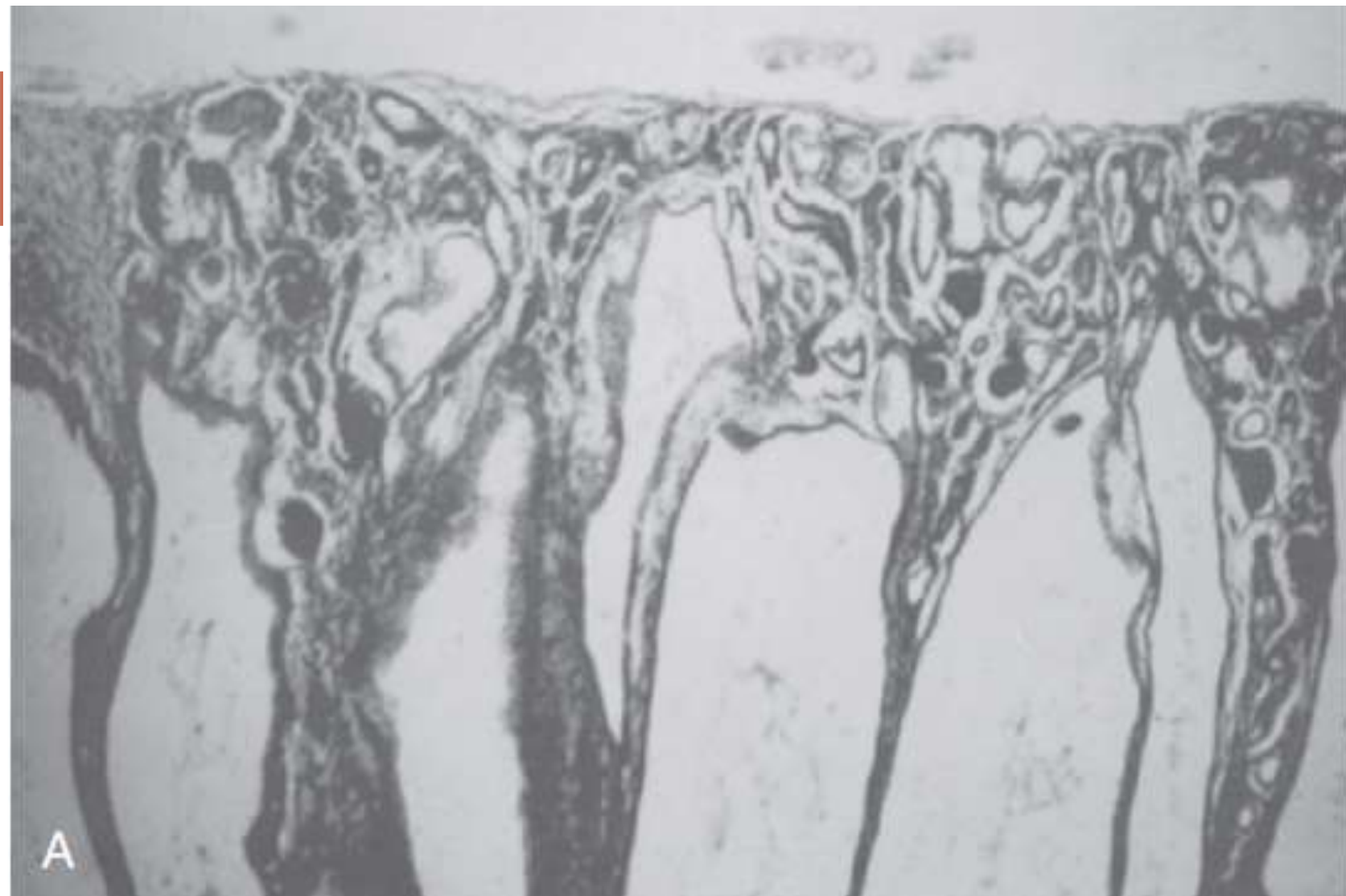
The kidneys are symmetrically enlarged (up to **20 times** their normal size).

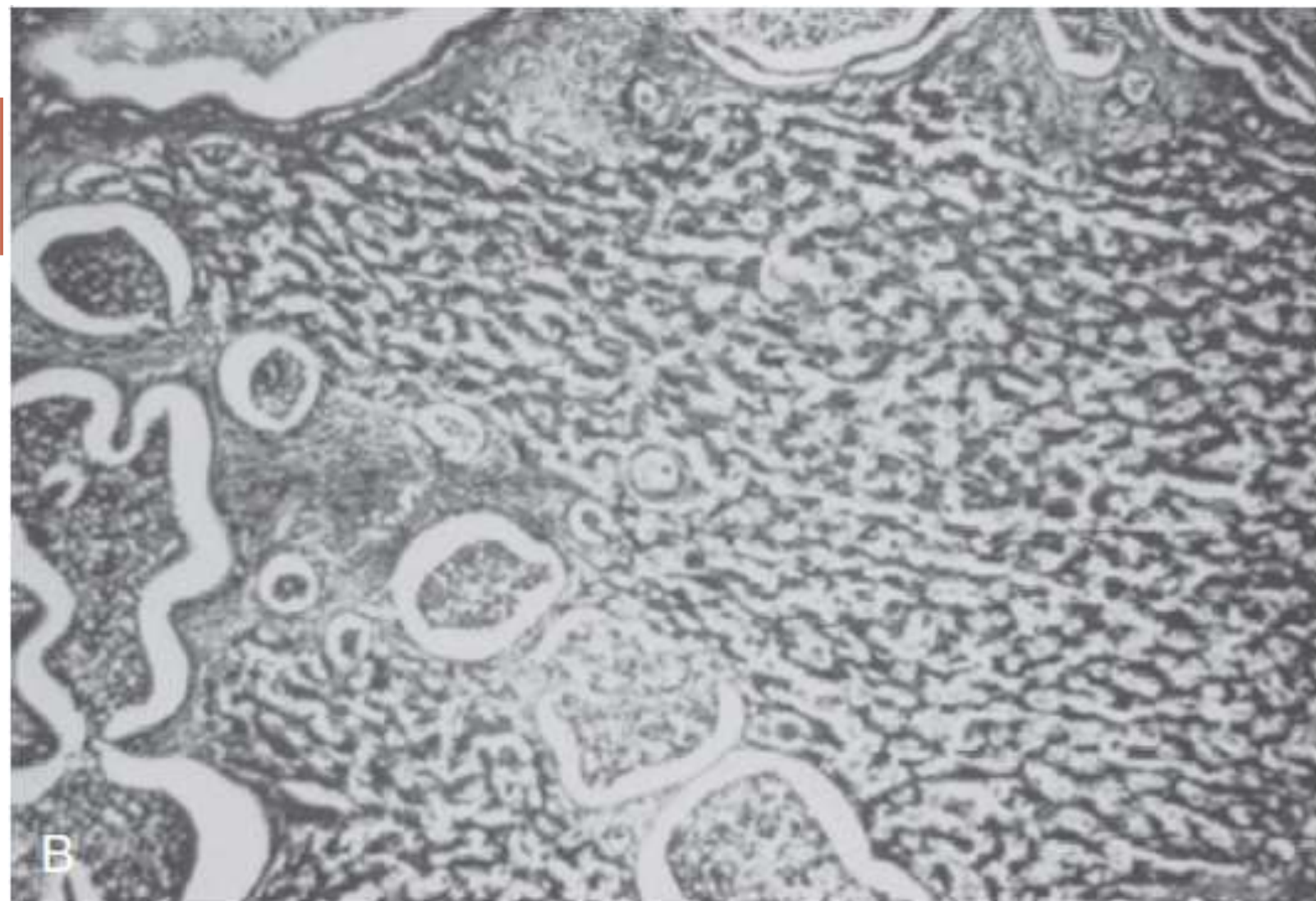
Small subcapsular cysts (**fusiform**) radiating from the medulla to the cortex.

Almost **100%** of collecting ducts are affected in the most severe cases.

All children with ARPKD have lesions in the **periportal** areas of the liver.







EVALUATION

Ultrasonography identifies bilateral, very enlarged, diffusely echogenic kidneys.

The kidneys typically have a homogeneous appearance

PA 332.5mm
SA 39S0j >97.0%

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FR 35Hz
RS

M3

2D
51%
C 58
P Low
HGen

✧ Dist 7.53 cm
✧ Dist 3.74 cm

RK

12



DDX

ADPKD: the cysts, if apparent in newborns, are usually diffuse and large

Severe bilateral hydronephrosis: (the kidneys are enlarged with hypoechogenic calyces).

Multicystic kidney: (hypoechogenic cysts lie within a nonreniform mass that has very little parenchyma)







A 2981

LI





bilateral mesoblastic nephroma

Wilms tumor

bilateral renal vein thrombosis

FR 22Hz
RS

2D
81%
C 50
P Low
Res

P

X

Left Trans

14



PAED ABDO

C9-2

40Hz

RS

Z 1.1

2D

57%

Dyn R 55

P Low

HRes

TIS0.3 MI 1.3

M3

Dist 1 8.21 cm
Dist 2 7.44 cm
✦ Dist 3 8.23 cm
Volume 263 ml

RUQ Trans

10cm



LEFT KIDNEY



If the diagnosis is in doubt, **CT** is valuable because it is more sensitive to inhomogeneity.

A newborn with **severe ADPKD** can also have enlarged, homogeneously hyperechogenic kidneys.

Age at onset can overlap, and typically, when ADPKD manifests at birth, cysts are apparent on the sonographic image.

Macrocyysts are rare in newborns with ARPKD but do increase in frequency as the child gets older.

Hepatic fibrosis can rarely occur in patients with ADPKD, as can **brain aneurysms** with ARPKD.

The correct diagnosis of ARPKD depends on the overall clinical data:

- **positive family history** with a recessive mode of inheritance
- **positive liver biopsy**,
- the **lack of the extrarenal** malformations.

TREATMENT

No cure has been found for ARPKD.

Respiratory care can ease or extend the child's life.

Unilateral or bilateral nephrectomy, because of respiratory and nutritional compromise.

treatment for **hypertension, congestive**

Portal hypertension may be dealt with by decompressive procedures such as a splenorenal shunt.

Esophageal varices may be managed, at least temporarily, by gastric section and reanastomosis.

Endoscopic sclerotherapy is widely employed in pediatric and adults with bleeding varices.



THANK YOU

