Congenital Anomalies

I. UPPER URINARY TRACT

A. Abnormalities of the Kidney Position and Number

1. Simple ectopia

a) Incidence is approximately 1 per 900 (autopsy) (pelvic, 1 per 3000; solitary, 1 per 22,000; bilateral, 10%). Left side favored.

b) Associated findings include small size with persistent fetal lobations, anterior or horizontal pelvis, anomalous vasculature, contralateral agenesis, vesicoureteral reflux, Mu¨llerian anomalies in 20–60% of females; undescended testes, hypospadias, urethral duplication in 10–20% males; skeletal and cardiac anomalies in 20%.

c) Only workup, ultrasound, voiding cystourethrography.

2. Thoracic ectopia

a) Comprises less than 5% of ectopic kidneys.

b) Origin is delayed closure of diaphragmatic angle versus

“overshoot” of renal ascent.

c) Adrenal may or may not be thoracic.

3. Crossed ectopia and fusion

a) Incidence is 1 per 1000 to 1 per 2000; 90% crossed with fusion; 2:1 male, 3:1 left crossed; 24 cases solitary, five cases bilateral reported to date.

b) Origin from abnormal migration of ureteral bud or rotation of caudal end of fetus at time of bud formation

c) Associated findings include multiple or anomalous vessels arising from the ipsilateral side of the aorta and vesicoureteral reflux; with solitary crossed kidney only; genital, skeletal, and hindgut anomalies .

4. Horseshoe kidney

c) Associated findings include anomalous vessels; isthmus between or behind great vessels hindered by the inferior mesenteric artery; skeletal, cardiovascular, and central nervous system (CNS) anomalies (33%); hypospadias and cryptorchidism (4%), bicornuate uterus (7%), urinary tract infection (UTI) (13%); duplex ureters (10%), stones (17%); 20% of trisomy 18 and 60% of Turner's patients have horseshoe kidney.
d) Excluding other anomalies, survival is not affected.
e) Stones; infection may result from stasis; rarely is true obstruction present (see ureteropelvic junction obstruction [UPJO]).

a) Incidence is 1 per 400 or 0.25%; 2:1 males.

b) Origin is fusion of lower poles before or during rotation

(4½ to 6 weeks' gestation).

Bilateral renal agenesis

a) Incidence is 1 per 4800 births or 1 per 400 newborn autopsies (75% are male) and typically lethal.

b) Origin either ureteral bud failure or absence of the nephrogenic ridge.

c) Associated findings include absent renal arteries, complete ureteral atresia in 50%, bladder atresia in 50%, Potter's syndrome .Also low birth weight, oligohydramnios, pulmonary hypoplasia, bowed limbs.

Unilateral renal agenesis

a) Incidence is 1 per 1100 in autopsy series, 1 per 1500 in radiographic series, 2:1 male, left kidney is more often involved than right kidney.
b) Origin is probably ureteral bud failure; there is a familial trend.
c) Associated findings include absent ureter with hemitrigone (50%), adrenal agenesis (10%), genital anomalies (20–40% in both sexes).
i) Mu¨llerian anomalies in females include uterovaginal atresia (Mayer-Rokitansky-Ku¨ster-Hauser syndrome), uterus didelphys, and vaginal agenesis.
ii) In males, the vas and seminal vesicle are absent or atretic.
d) If the single kidney is normal, no special precautions required and survival is not affected; management of the genital abnormalities.

Supernumerary kidney

a) Incidence is unknown.

b) Origin a combined defect of ureteral bud and metanephros.

c) Associated findings are hydronephrosis (50%), common
ureter (40%), duplex ureter (40%), and ectopic ureter or one ending in the pelvis of the ipsilateral kidney (20%).

Autosomal dominant polycystic kidney disease

a) Chromosome 16 and chromosome 4.
b) Autosomal dominant transmission.
c) Adult type is the most common cystic disease in humans, with an incidence of 1 per 1250 live births and accounts for 10% of all end-stage renal disease.
d) Usually presents after between 30 and 50 years with pain, hematuria, and progressive renal insufficiency, but it is also seen in children. Rarely present in newborns.
e) Intravenous urography (IVU) reveals irregular renal enlargement with calyceal distortion; ultrasound shows multiple cysts of variable sizes.

f) Associated findings are liver cysts without functional impairment in one third of patients and berry aneurysms in 10–40%.
g) Complications include uremia, hypertension, myocardial infarction, and intracranial hemorrhage (9%).
h) Management involves control of blood pressure and urinary infection, relief of cardiac failure, and eventually dialysis or transplantation. Some of these patients encounter issues with pain typically from renal capsular stretching by the cysts.
i) Pathology: rounded or irregular cysts located in all parts of the nephron.

Autosomal recessive polycystic kidney disease

a) Chromosome 6.
b) Infantile type. Rare (1 per 10,000 live births), usually presents with bilateral flank masses in infancy but can present in childhood with renal or hepatic insufficiency.
c) IVU shows huge (12–16 times normal) kidneys with a pronouncedly delayed nephrogram and characteristic streaked appearance ("sunburst" pattern).
d) May be distinguished from hydronephrosis, renal tumor, and renal vein thrombosis by IVU and ultrasound. (Bright echoes on ultrasound.)

e) Associated findings are congenital hepatic (periportal)
fibrosis and dilation of bile ducts with the
degree of hepatic insufficiency varying inversely with the severity of renal disease and directly with the age of presentation; cysts elsewhere are uncommon.
f) Complications are renal and hepatic failure, hypertension,
and respiratory compromise in the newborn;
patients usually die within the first 2 months of life.

Medullary cystic disease (juvenile nephronophthisis)

refers to a group of disorders with various genetic patterns characterized pathologically by bilateral small kidneys, attenuated cortex, atrophic and dilated tubules, medullary cysts, and some interstitial fibrosis.

a) Patients progress to end-stage renal disease by about age 20; juvenile form is responsible for 20% of childhood renal failure deaths.

b) Medical management of renal failure can delay need for transplant.

c) Polydipsia and polyuria in 80%, retinitis pigmentosa in 16%.

Unilateral multicystic dysplastic kidney

is the most common
cystic disease of the newborn and the second most common abdominal mass in infants after hydronephrosis, including UPJO.
a) The left kidney is more commonly involved, but there is no sex predilection or familial tendency.
b) Origin either (1) ischemic, from failure of the normal shift of vasculature as the kidney migrates, producing also the associated atretic ureter or
(2) failure of ureteral bud to stimulate metanephric blastoma.

c) Contralateral renal abnormalities are most common when the multicystic kidney is small and/or the ureteral atresia is low. Vesicoureteral reflux may be present in up to 20% of cases.

d) Ultrasound is the most diagnostic study (multiple hypoechoic areas of various sizes without connections

or dominant medial cyst and without identifiable parenchyma),

as IVU or renal scan demonstrates ipsilateral nonfunction; IVU and voiding cystourethrography (VCUG) are done to evaluate the remainder of the urinary tract.

Collecting System Abnormalities

1. Calyceal diverticulum occurs in 4.5 per 1000 urograms.
a) Origin is failure of degeneration of third- and fourthorder branches of ureteral bud, leaving a pocket lined with transitional epithelium connected to the collecting system near the calyceal fornix.
b) In approximately one third of patients, stones will form; some will become sites of persistent infection due to stasis; the rest remain asymptomatic.
c) Treatment involves removal of stones, drainage of purulence, and marsupialization to the renal surface with closure of the collecting system and cauterization of the epithelium.

2. Hydrocalycosis is a rare lesion involving vascular compression, cicatrization, or achalasia of the infundibulum;
it rarely requires any intervention.
3. Megacalycosis is a rare lesion involving all of one or both kidneys with dilated unobstructed calyces usually numbering more than 25 per kidney (normally 8–10 calyces
per kidney). May be confused radiographically with obstructive uropathy.
a) Results from combination of faulty ureteral bud division, hypoplasia of juxtamedullary glomeruli, and maldevelopment of calyceal musculature.
b) Males 6:1 over females, only in Caucasians. X linked recessive gene.
c) May be associated with stones or infection but in itself causes no deterioration of renal function.

4. Infundibulopelvic stenosis may involve part or all of one

or both kidneys.

a) The calyces become quite large but usually no progressive

functional deterioration occurs. Pain difficult

to manage when present.

b) May be associated with dysplasia and lower tract

anomalies (e.g., urethral valves).

c) Commonly associated with vesicoureteral reflux.

5. UPJO is the usual cause of the most common abdominal mass in children (hydronephrosis).

a) There is a 2:1 male predominance in children and left-sided predominance in all ages.

b) Several possible causes, including segmental muscular attenuation or malorientation, true stenosis, angulation, and extrinsic compression. Crossing lower pole vessels are present in approximately 20–30% of cases, but an intrinsic lesion (either noncompliant or nonconducting) is common.

c) Associated findings include reflux (5–10%), contralateral agenesis (5%), and contralateral UPJO (10%); rarely dysplasia, multicystic kidney, or other urologic anomaly.

d) Symptoms and signs include episodic flank pain and/or mass, hematuria, infection, nausea and vomiting, and sometimes uremia. In infants, the flank mass may be the only sign, whereas the older child will exhibit any of the others; very often gastrointestinal distress and poorly localized upper abdominal pain are the only symptoms.

e) Radiologic findings are delayed excretion on the affected side with variable dilation of pelvis and calyces or even no visualization on IVU; on ultrasound, multiple interconnected hypoechoic areas with dominant medial hypoechogenicity and identifiable cortical rim. There is usually some measurable function on renal scan. When function is good, the drainage is delayed even in the face of furosemide (Lasix) administration beyond 20 minutes.

f) Prompt surgical repair by excision of the narrow segment and a spatulated anastomosis of the ureter to the tailored renal pelvis for symptomatic cases (Anderson Hynes pyeloplasty) .Most are diagnosed antenatally and can be followed with serial renal scan and ultrasound

g) Follow-up consists of ultrasound at 1 month and renal scan at 3 months and ultrasound at 1 year postoperatively in most cases.

Ureteral Anomalies

1. Duplication of ureter occurs in 1 per 125 autopsies; 1.6:1 female, 85% unilateral.
a) Autosomal dominant with incomplete penetrance.
b) Seems to arise from two ureteral buds meeting the metanephros—in most cases, but may also be caused by a bud that bifurcates immediately after arising, before meeting the metanephros.
c) Associated with reflux (42%), renal scarring and dilation (29%), ectopic insertion (3%), large kidneys with excess calyces, dysplasia/hypoplasia, infection, and ureteroceles.
d) Duplication itself is of no clinical significance, but the associated anomalies may require intervention

2. Atresia is usually associated with a multicystic dysplastic kidney; distal segment atresia is often associated with contralateral hydronephrosis or dysplasia (50%).

3. Megaureter has a 3:1 male and 3:1 left-sided predominance; the term is used loosely to describe any dilated ureter, but there are three distinct types.

a) Refluxing megaureter originates because of the reflux, although some cases have an abnormal distal segment and some element of obstruction.

b) A widened ureteral bud gives rise to a ureter dilated down to the orifice, which is in the normal position, and there is no obstruction (nonreflux, nonobstructed type).

c) The primary obstructed type is the most common and results from a stenotic or aperistaltic distal short segment; the orifice is in the normal position.
d) The refluxing type, with its laterally ectopic orifice, may be associated with a dysplastic kidney, one scarred by infection, or both; the other types drain normal or hydronephrotic kidneys.
e) The ultrasound will show moderate to severe hydronephrosis and proportionately greater ureteral dilation; VCUG will diagnose the reflux type; a Lasix renogram would distinguish obstructed from nonobstructed types.
f) There are mild primary obstructed megaureters with only a spindle-shape dilation of the distal ureter and normal (sharp) calyces; these require no treatment.

g) Surgical correction is needed for some obstructed and refluxing megaureters. Refluxing megaureters more commonly require tailoring than obstructed ones, Congenital Anomalies 899
which tend to decrease in caliber after excision of the aperistaltic distal segment.
h) Follow-up includes ultrasound at 1 month and renal scan at 3 months. An ultrasound is done 1 year postoperatively

4. Vesicoureteral reflux (VUR) occurs in approximately 1 per 1000 in the general population but is 8 to 40 times more frequent in affected families; it can be found in 50% of infants and 30% of children with a UTI.

a) It may occur because the ureteral bud arises ectopically

leading to a laterally placed orifice and short submucosal tunnel or because the development of the intrinsic smooth muscle of the distal ureteral segment is delayed or incomplete. High intravesical pressures may cause a marginally competent ureterovesical junction to reflux, and evidence is growing that voiding dysfunction in the child may cause or exacerbate reflux.

b) Duplicated ureters and renal hypodysplasia may be associated with refluxing ureters with laterally ectopic orifices. Infection and renal scarring are prominent findings with all types of refluxing ureters regardless of grade. Voiding dysfunction and urethral obstruction by valves are associated with an acquired form of reflux.

c) Reflux is best graded I to V by the International Reflux

Study system depending on the degree of dilation.

d) All children with reflux should be placed on prophylactic

antibiotics at one-fourth the therapeutic dose

e) Grades I–III (minimally dilated) are usually treated medically initially; grades IV–V usually require surgical correction. Low volume VUR (grades I–III) resolves at a rate of 17% per patient-year, whereas grade IV VUR resolves at a rate of 4% per patientyear.
f) Reimplantation of the refluxing unit by the Cohen technique is the standard surgical management, with nearly complete success; duplicated ureters are reimplanted in their common distal muscular sheath.
Recently, subureteric injection as well as laparoscopic and vesicoscopic approaches have been used with good success in preliminary reports.
g) Breakthrough infections, failure to comply with the antibiotic prophylaxis regimen, persistent reflux into puberty in females, progressive scarring, and worsening renal function are all considerations that favor surgical intervention, but there are no absolute indications for surgery for reflux.

5. The incidence of ureteral ectopia is approximately 1 per
1900; ectopic ureters are duplex in 80% of females, more often single in males; there is a 3:1 female predominance, and approximately 10% are bilateral.

a) The cause is a failure of the ureteral bud to separate from the mesonephric duct, probably due to its ectopic origin on the duct

c) Associated findings.

i) Renal dysplasia correlates with the degree of ectopy.

ii) Contralateral duplication accompanies single ectopic ureter in 80%.

iii) Incontinence and ureteral obstruction are variable findings; incontinence may be due to an orifice located below the sphincter in females or to failure of bladder neck development.

iv) Bilateral single ectopic ureters in which the orifice is distal to the bladder neck lead to poorly developed bladder and incontinence due to outlet incompetence and failure of bladder cycling.

d) Management is most often removal of the renal segment and ectopic ureter; rarely the segment may be salvageable by ureteroureterostomy or reimplantation.

6. Ureterocele occurs with a frequency between 1 per 500 and 1 per 4000 in autopsies, accounting for approximately
1% of pediatric urologic admissions and is bilateral
in 10–15% of cases. Females 4:1 over males.
a) Develops due to a combination of an abnormal ureteral
bud with either a stenotic orifice or involvement
of the distal ureter in the expansion of the vesicourethral canal. The ureter is duplicated in children
(80%), with the ureterocele draining the upper pole ureter. Presence of a simple ureterocele subtending a single ureter is less common in children.

b) Associated anomalies include contralateral ureteral duplication in 50%; renal segmental dysplasia; renal fusion and ectopia; reflux (50%); and, rarely, incontinence.
c) Classification is based on location of the orifice and is typically defined as intravesical or ectopic.
d) Cecoureterocele, a subclassification of ectopic ureterocele, differs in that a "cecum" extends beyond the orifice down the urethra; it may be associated with poor bladder neck development and incontinence.

e) Management is varied.
i) Puncture of the ureterocele as newborn.
ii) Upper pole nephrectomy with decompression of the ureterocele.
iii) Same as ii with lower excision of the ureterocele with common sheath reimplantation and bladder neck reconstruction.
iv) Same as i with delayed excision of ureterocele plus reimplant with bladder neck reconstruction if vesicoureteral reflux persists or occurs following
i. It appears that whether one takes an approach that favors removal of the upper tract moiety only or puncturing the ureterocele, the development of de novo VUR is relatively the same (10–15%).

LOWER URINARY TRACT

A. Exstrophy/Epispadias—Spectrum of Anomalies

1. Origin is failure of the cloacal membrane to migrate toward the perineum at 4 weeks' gestation, preventing ingrowth of lateral mesoderm and coalescence of genital tubercles.

a) The most consistent finding is some degree of separation

of symphysis pubis.

b) Epispadias (30%) may be penopubic with incontinence in males (55%), penile (20%) with or without incontinence, or balanitic (5%) or may occur in females with incontinence (20%). It consists of a dorsal meatus with a distal mucosal groove, flattened glans, or bifid clitoris; in males, there is a variable dorsal chordee with shortening of the corporal bodies in severe forms (penopubic).

c) Nearly all cases of epispadias require complete disassembly with or without complete separation of the distal urethra from the glans (Mitchell) technique.

d) Classic exstrophy (60%) occurs in 1 per 50,000 births with 3:1 male predominance; the bladder and the urethra are open dorsally, and the penis is short or the clitoris is bifid.

Exstrophy may be managed in stages or by primary single repair. A staged closure begins with bladder closure in the newborn period.

a) Penile lengthening by freeing corpora from pubic bone attachments and tubularization of the bladder neck is accomplished during the first stage.

b) In cloacal exstrophy, the omphalocele and vesicoenteric fissure must be dealt with by lateral closure of the bowel end colostomy and omphalocele repair. The bladder halves are approximated in the first stage.

The second stage is epispadias repair, in most cases at approximately 1–2 years of age..

The third stage in those with functioning, sufficiently large bladders is achieving continence by bladder neck tubularization (60% success).

a) Those who fail this are candidates for augmentation plus intermittent catheterization.

b) Most cloacal exstrophy patients have undergone early ileal loop diversion, but a few may be reconstructed along the same principles.

6. Second option is complete penile disassembly with bladder closure and bladder neck and epispadias repair all done at a single stage (Mitchell repair).

7. All patients require careful follow-up throughout life with survey of the upper tracts by IVU or ultrasound, monitoring of acid base balance, renal function tests, and supportive counseling.

Urachus

Patent urachus and persistence of portions of the urachus

as cysts result from failure of fibrosis of the cranial

embryonic bladder segment; they are excised when symptomatic.

If infected, primary drainage, antibiotic coverage,

and secondary resection are appropriate. In a few cases,

the urachal segment may undergo malignant transformation

(adenocarcinoma).

Posterior Urethral Valves

1. Incidence. In boys, 1 per 5000 to 8000; >50% are diagnosed in the first year of life, generally with more severe obstructions.
2. Proposed cause is failure of regression of the terminal segment of themesonephric duct, which is normally represented by the plicae colliculi. Type II valves are nonobstructing normal folds in the prostatic urethra; type III valves represent either more marked anterior fusion of the valve leaflets or congenital urethral membrane (a separate embryologic entity). Recent observations suggest that types II and III are variations of type I valves.

3. Associated findings.

a) Vesicoureteral reflux (40%, approximately one half bilateral) resolves in approximately one third of cases generally within 2 years. Persistent unilateral reflux is usually associated with a nonfunctioning kidney, most commonly the left one.

b) Severe renal dysplasia is common in those with severe obstruction.

c) Severe hydroureteronephrosis.

d) Acute renal failure and acidosis in the newborn are obstructive phenomena; chronic renal insufficiency from dysplasia may occur.

4. Diagnosis.

a) Antenatal diagnosis.

b) UTI or poor stream in an infant or older child; incontinence occasionally in an older child.

c) A newborn with palpable bladder and kidneys and urinary ascites.

d) VCUG is the diagnostic study; ultrasonography and renal scan are employed to assess the extent of upper tract damage and postoperative recovery.

5. Management.

a) In the sick infant, bladder drainage with a small feeding tube (6 F) per the urethra is maintained while acidosis and sepsis are treated; VCUG may be done with this catheter in place.

b) The healthy infant or older child may undergo transurethral incision of valves initially; the sick infant when creatinine stabilizes and sepsis resolves.

c) Cutaneous vesicostomy can be used as a temporizing measure in a very small infant but is rarely required with today's endoscopic equipment.

d) Nonfunctioning kidneys with refluxing ureters should be preserved. The ureters may be used as tissue for augmentation of the bladder if needed at the time of renal transplant.

e) Ureteral tailoring and reimplantation are almost never indicated and are often fraught with failure.

f) Antibiotic prophylaxis is maintained as long as reflux persists or upper tract emptying is slow (usually through adulthood).

Megalourethra

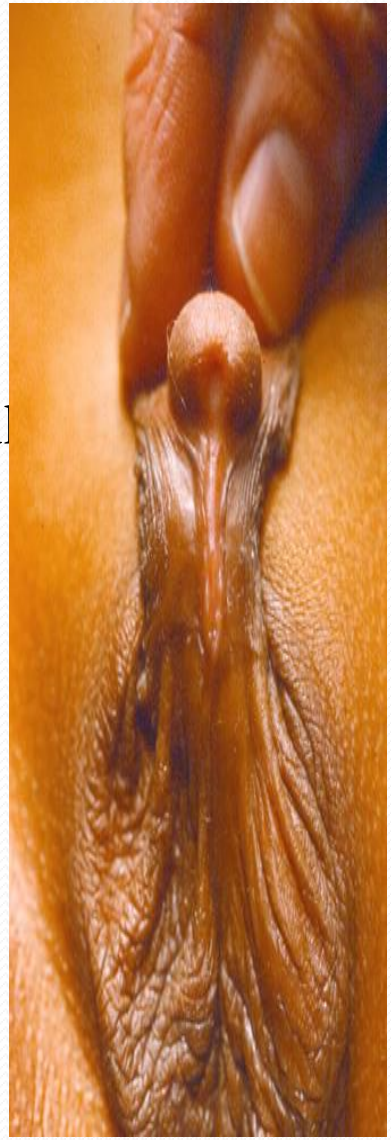
1. This rare lesion is usually associated with prune belly syndrome.
2. Occurs in two types.

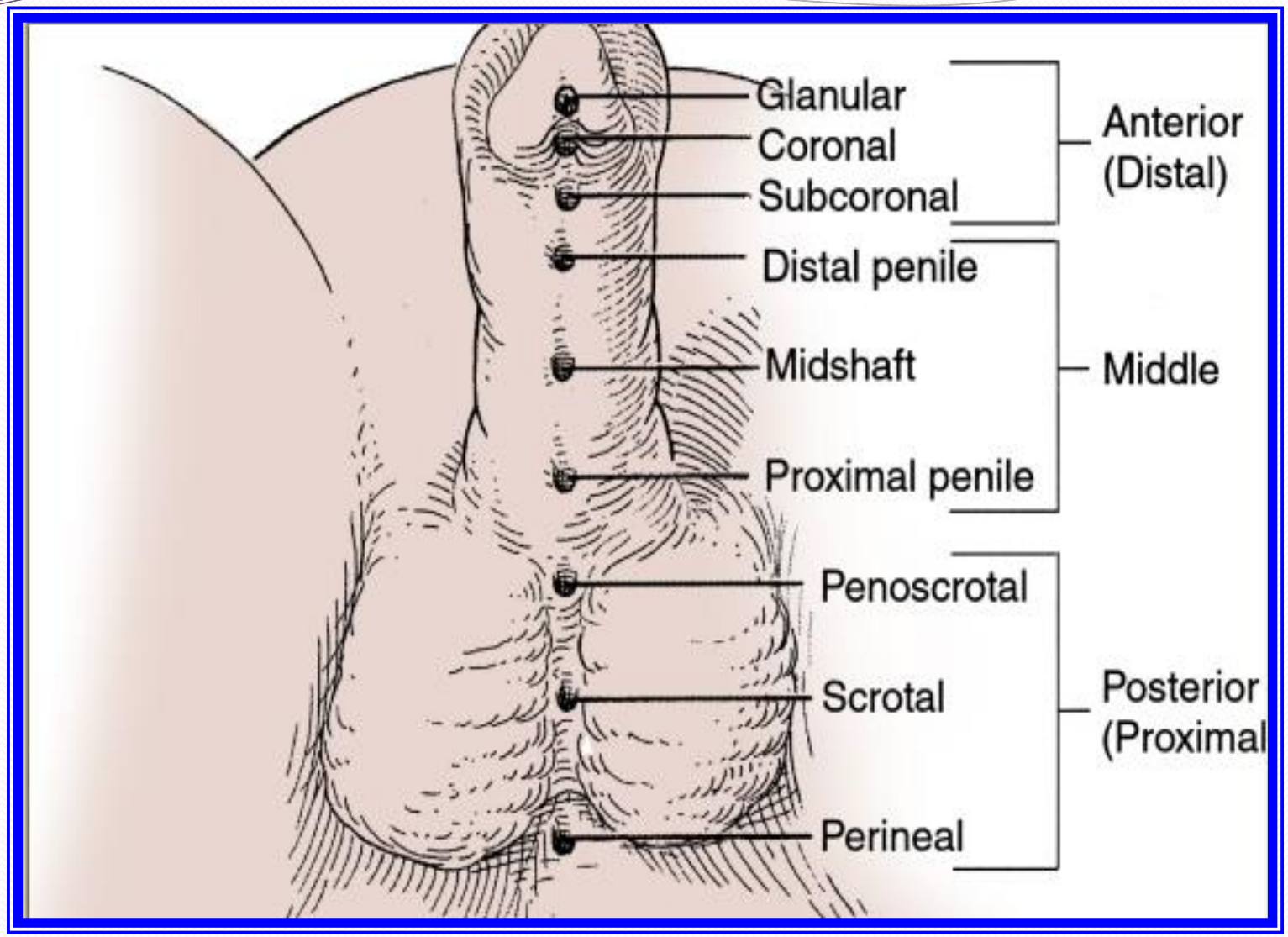
a) Scaphoid type is a deficiency of corpus spongiosum allowing ballooning of the urethra during voiding; it can be repaired with hypospadias techniques.

b) Fusiform type involves deficiency of corpora cavernosa as well as corpus spongiosum, resulting in elongated flaccid penis with redundant skin. This form is seen usually in stillborn infants with other cloacal anomalies and is difficult to correct because of the lack of adequate corporal tissue.

Hypospadias

Hypospadias occurs in 1 in 300 live boys; there is a 14% incidence in siblings and an 8% incidence in offspring.
1. Caused by failure of the mesodermal urethral folds to converge in midline; chordee results from failure of urethral plate disintegration or fibrosis of inner genital folds (which form the spongiosum and dartos fascia).
2. Associated findings.
a) Blunted human chorionic gonadotropin response to gonadotropin releasing hormone and low androgen receptor levels in a few cases.
b) Undescended testes in 9.3% (30% with penoscrotal or more proximal meatus). Up to one third of boys with hypospadias and undescended testes have an intersex state, usually genetic mosaicism.

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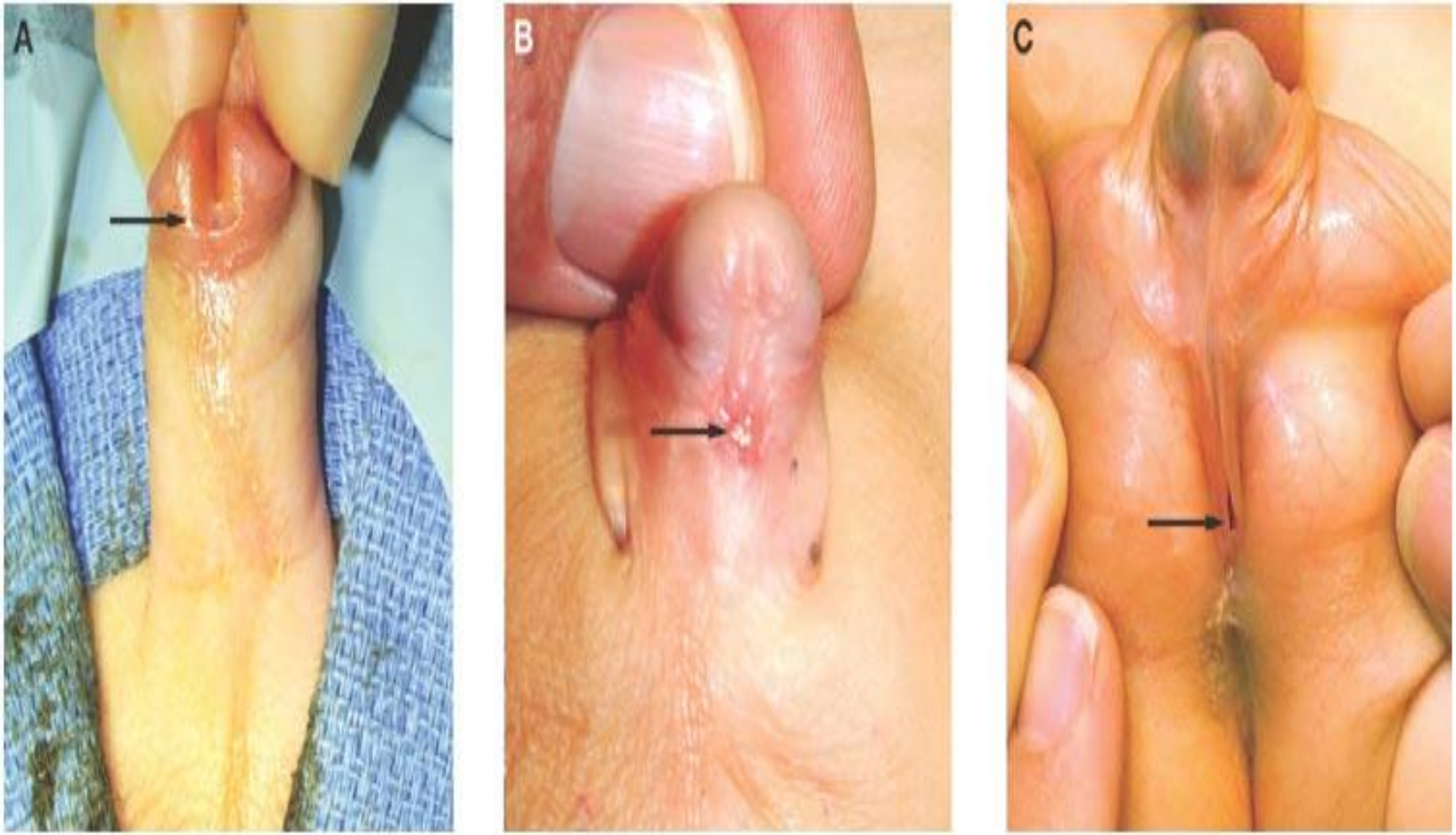
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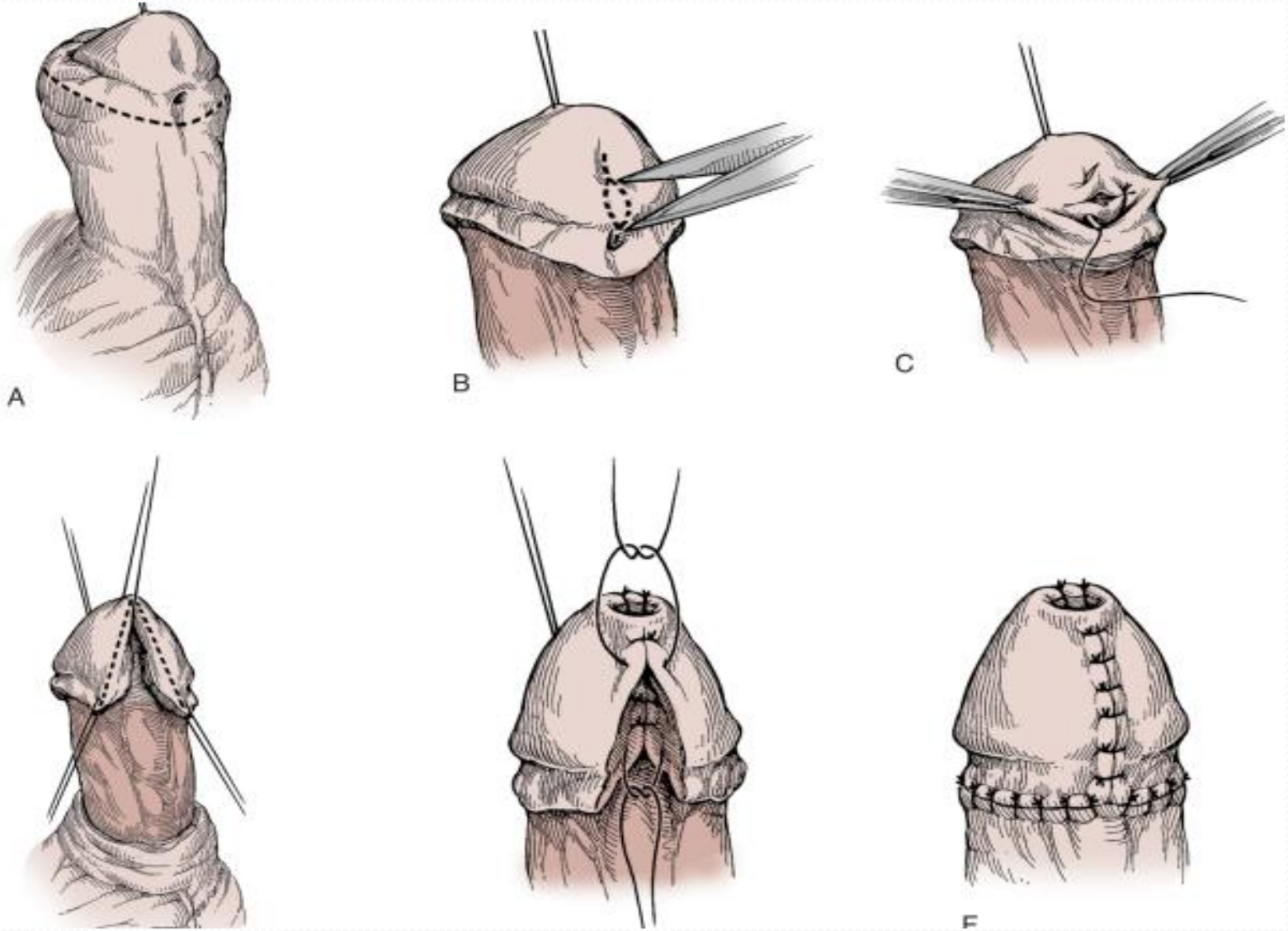
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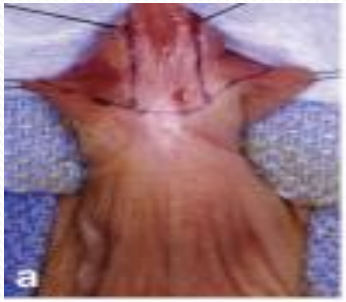

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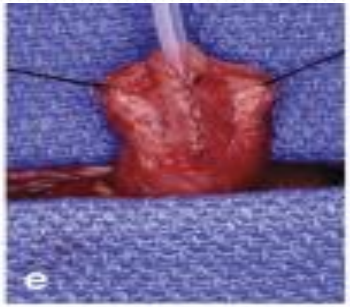

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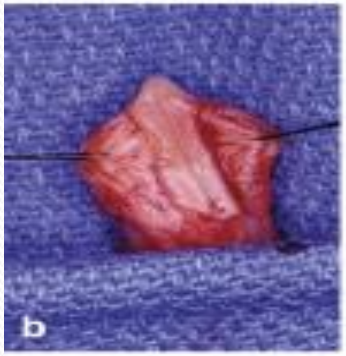

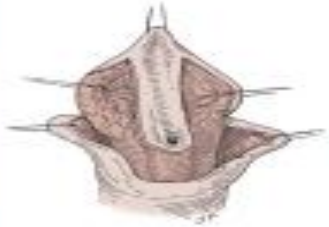

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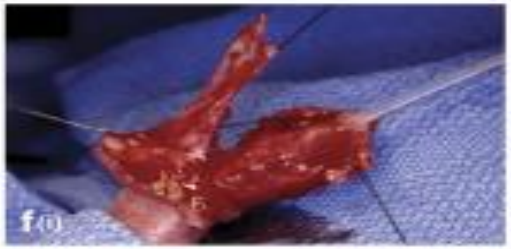

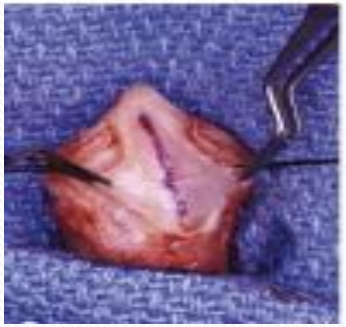

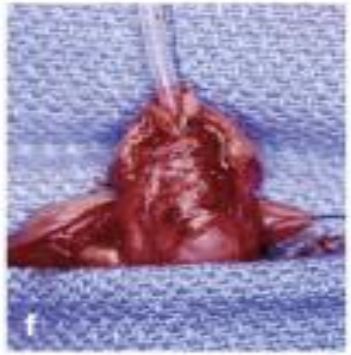

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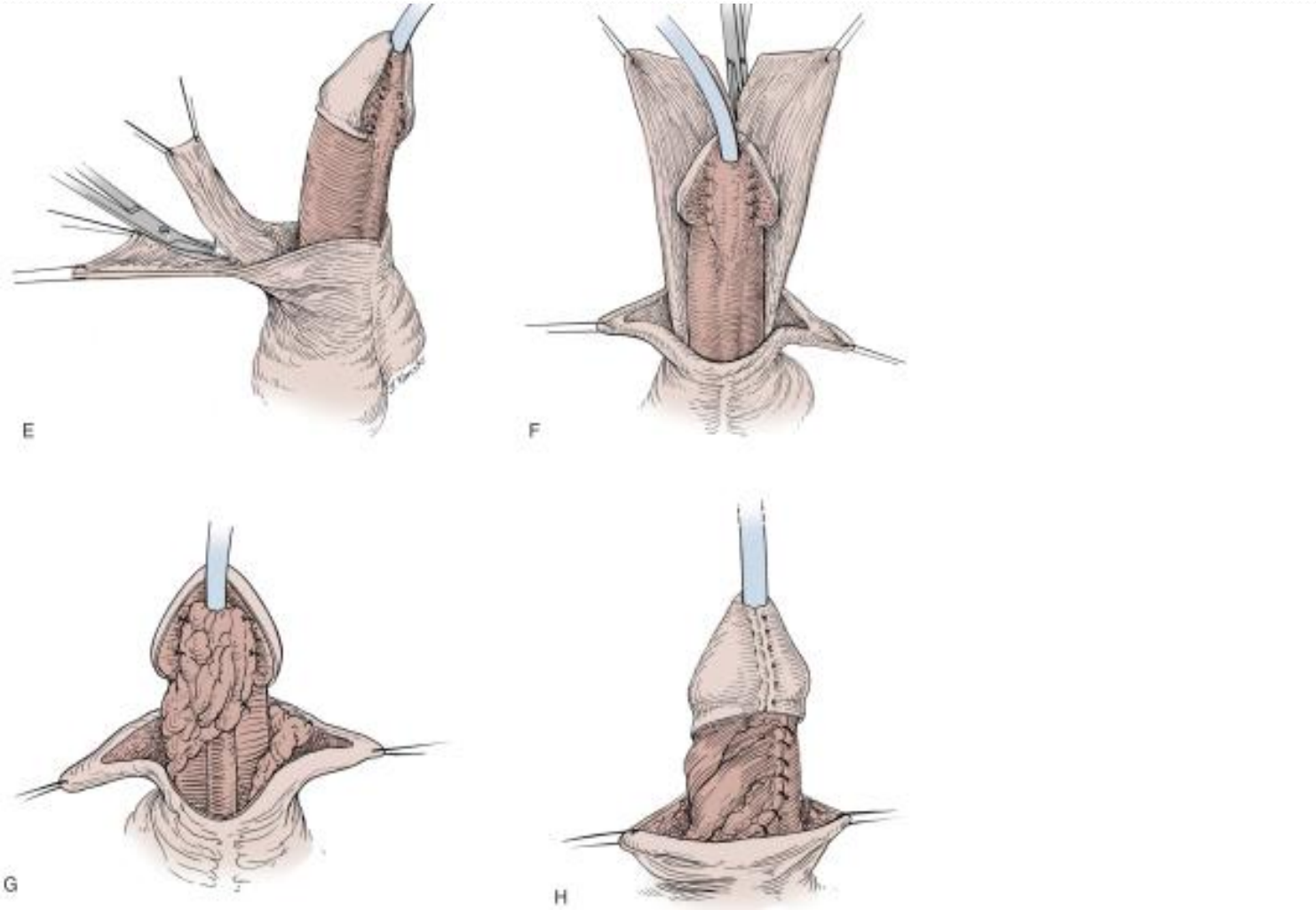
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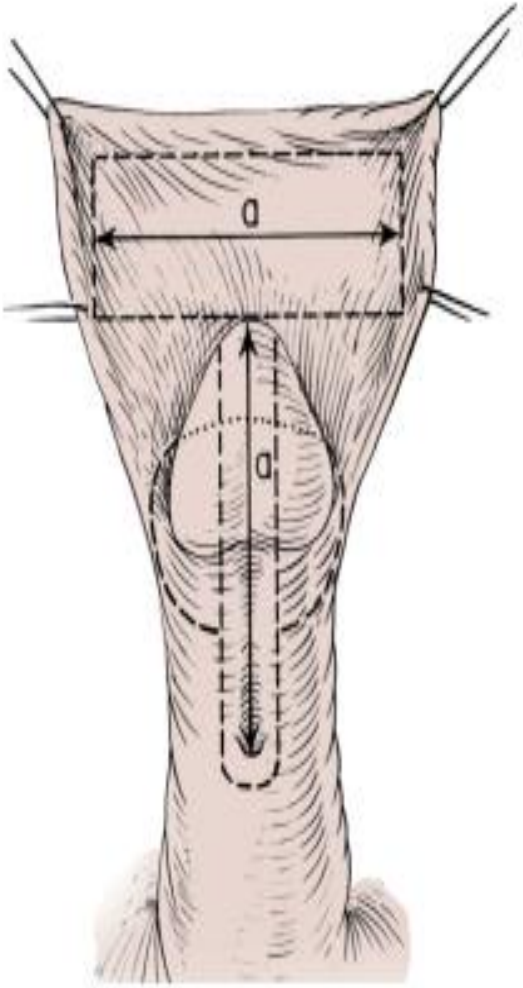
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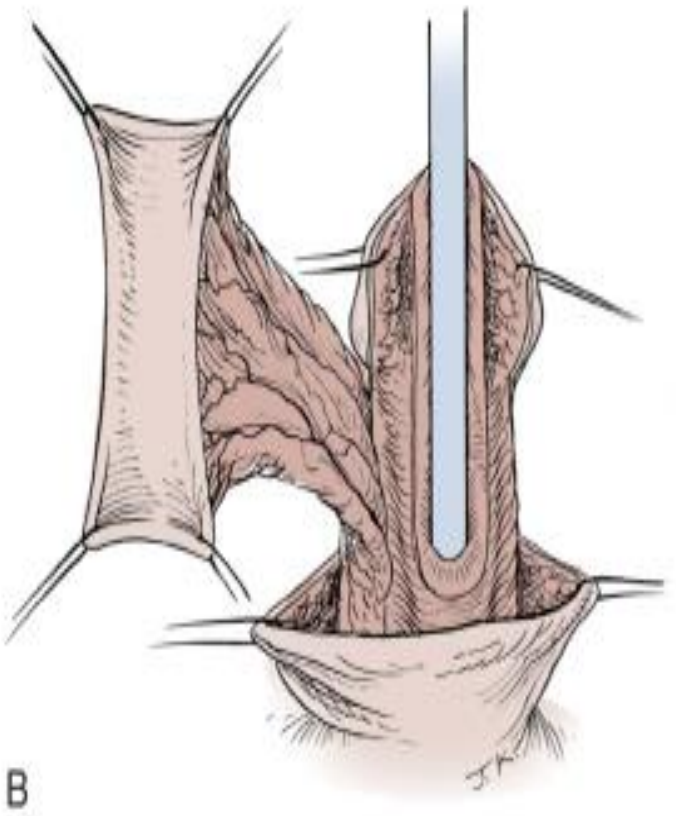
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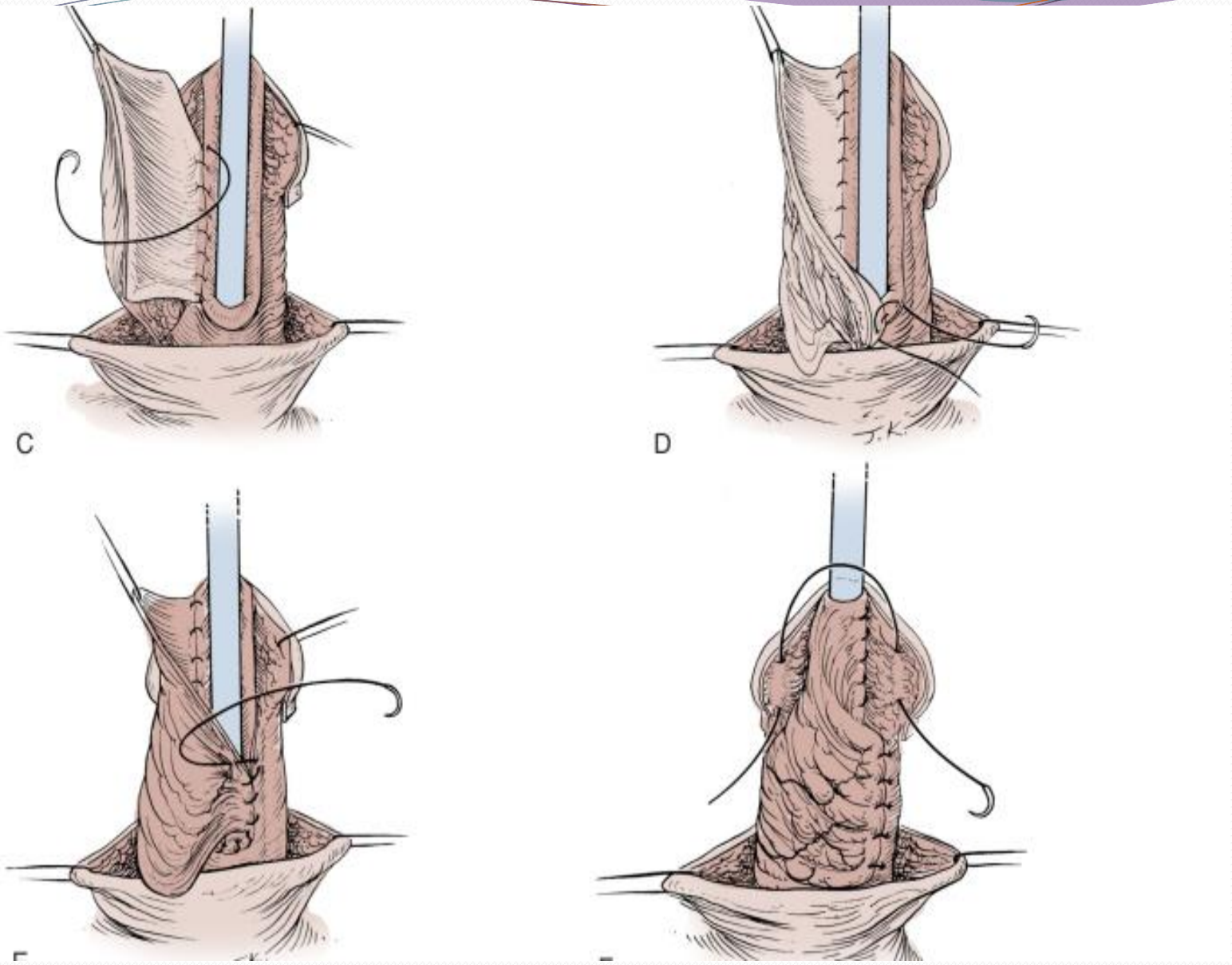
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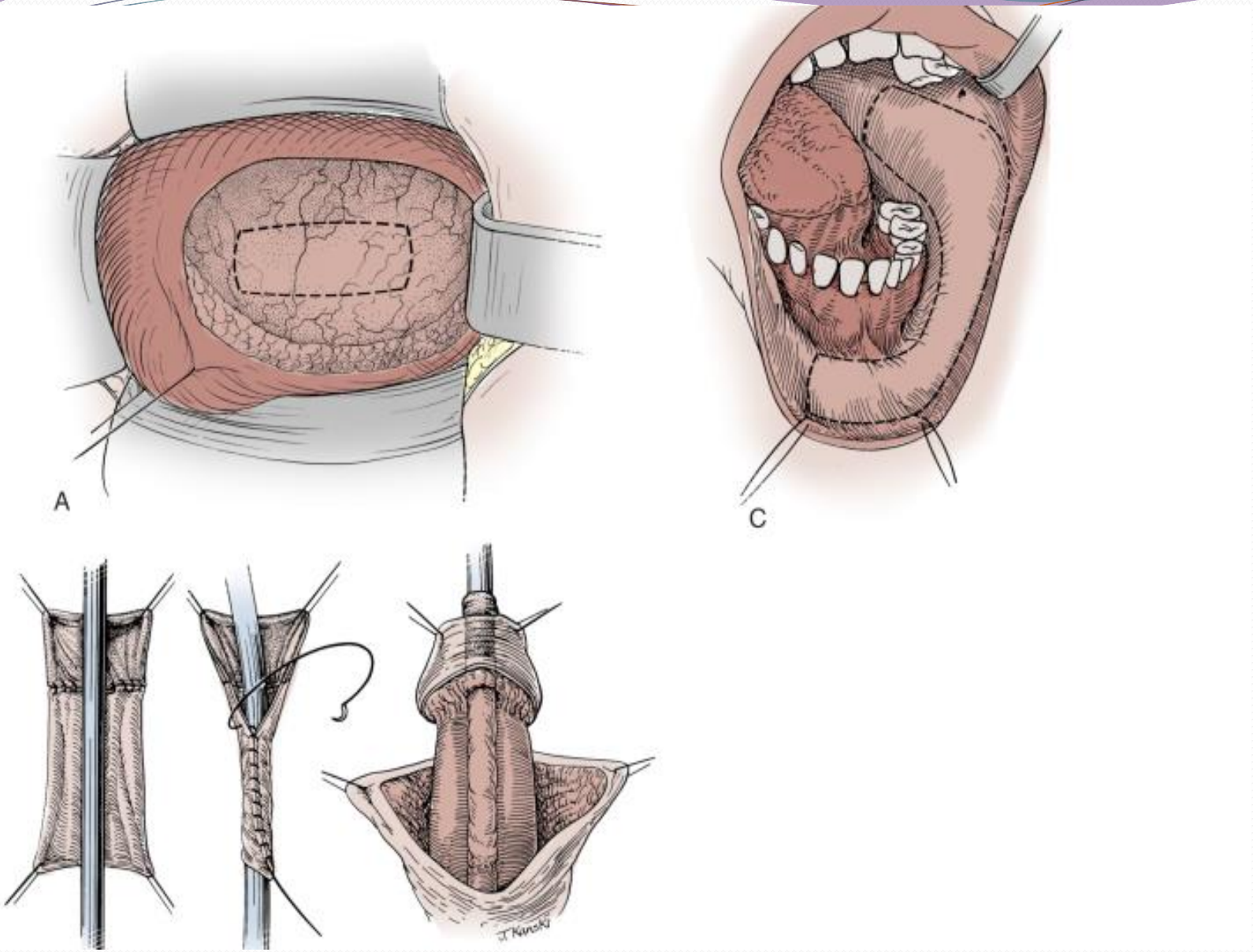
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c) Inguinal hernia in 9%.
d) Upper tract anomalies in 46% when associated with imperforate anus, 33% when meningomyelocele is present, 12–50% when one other system anomaly is present, 5% with isolated hypospadias (screening intravenous pyelogram [IVP] not needed for simple hypospadias).
3. Classification (simplified)
a) Hypospadias without chordee (straight erections, meatus between midshaft and corona).
b) Hypospadias with chordee.
i) Meatus penile or penoscrotal after release of chordee.
ii) Meatus scrotal or perineal.
c) Chordee with hypospadias.
i) With normal urethra.
ii) With short or hypoplastic urethra.

Management.

a) One-stage correction between 4 and 12 months of age is preferred.

b) Glanular hypospadias may be corrected by meatal advancement and glanuloplasty (MAGPI) For coronal repairs, the Snodgrass or TIP

or onlay island flap procedure, depending onmeatal position as well as surgeon preference are most commonly employed.

c) Penile shaft or more proximal hypospadias may be managed by inner preputial transverse island flap

d) Severe penoscrotal hypospadias may require combined island flap and primary (Duplay) closure of the proximal urethra and may need secondary scrotoplasty to improve the cosmetic result.

e) Degloving the penis and mobilizing the urethra may treat skin chordee without urethral involvement.

f) Chordee with hypoplastic urethra requires island flap urethroplasty after chordee release due to bowstring effect of short urethra.

g) Urinary diversion for 2 weeks in all but very distal repairs, with a small Silastic urethral stent ensures adequate bladder drainage.
h) Compressive dressing for 2–3 days is commonly used.
5. Results and complications.
a) Small urethrocutaneous fistulae are the most common complication. These may be closed in layers with 90% success.
b) Postoperative bleeding can usually be stopped with compression.
c) UTI occurs in less than 10% of cases and can be treated with the usual oral agents.
d) Strictures are rare and usually occur at the meatus or the proximal end of the repair and are treated by Y-V meatoplasty or excision; direct vision urethrotomy is sometimes successful for short strictures.
e) When carefully done, the procedures outlined provide functional and cosmetically nearly normal penis and meatus even in the most severe hypospadias cases.