

ÜROGENİTAL SİSTEMİN KONJENİTAL ANOMALİLERİ



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Potter yuzu

Böbrek Anomalileri



➤ Bilateral renal agenez

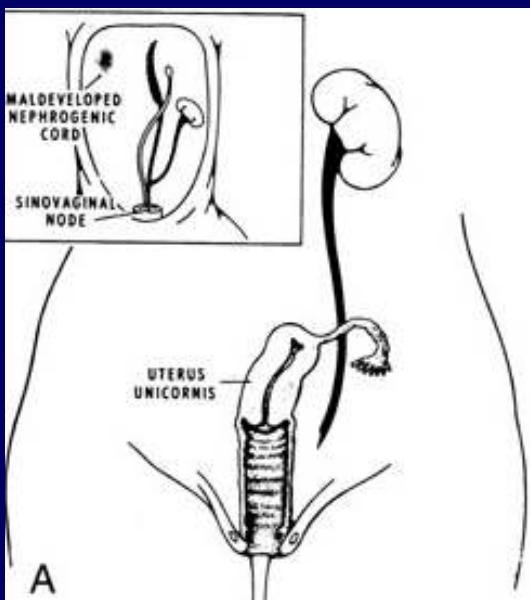
➤ Unilateral renal agenez

➤ 1/500-1000

➤ Kızlarda Muller kanalı anomalileri

➤ Erkeklerde vaz deferens agenezi

➤ Çoksayıda böbrek



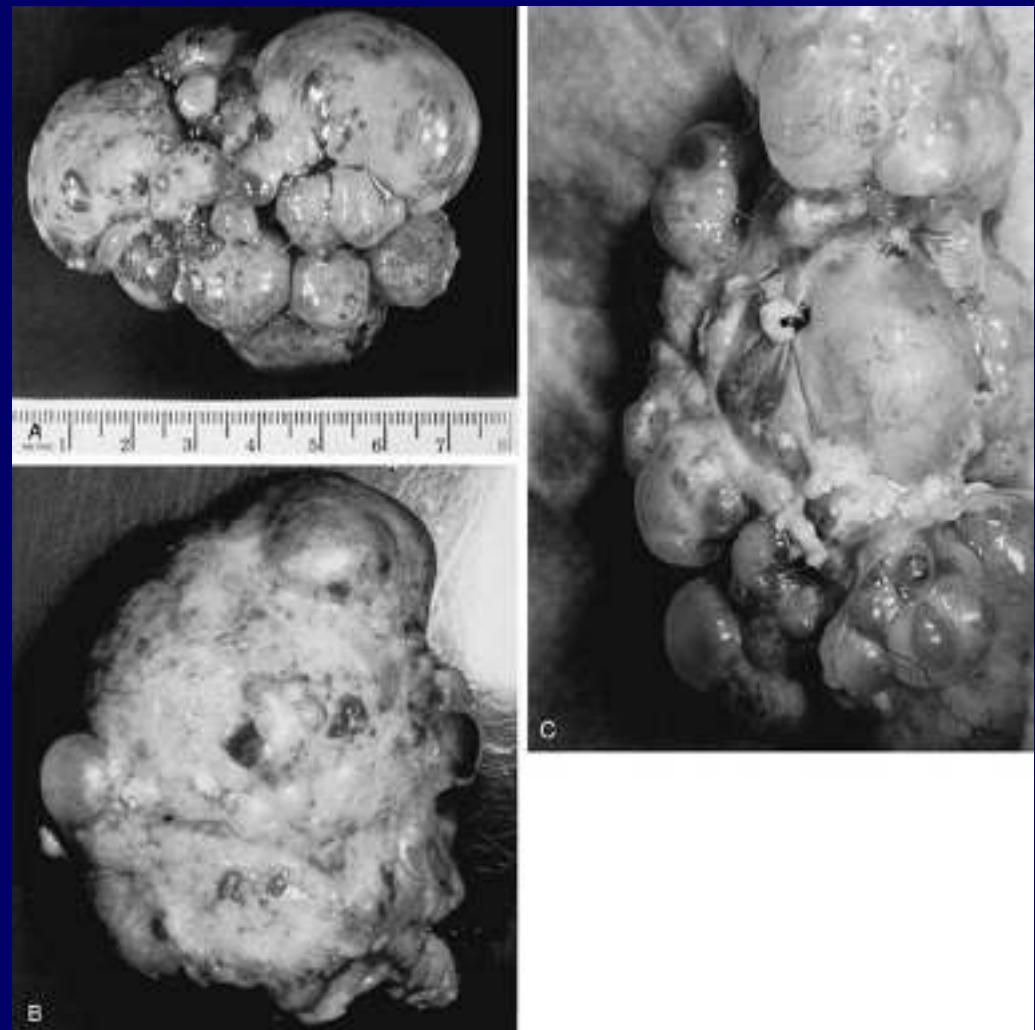
Böbrek Anomalileri

- Displastik böbrek: Farklı dokular
- Hipoplastik böbrek: Böbrek dokusu
- Segmental hipoplazi (Up-Askmark böbreği)

Bobrek Anomalileri

Kistik Hastalıklar:

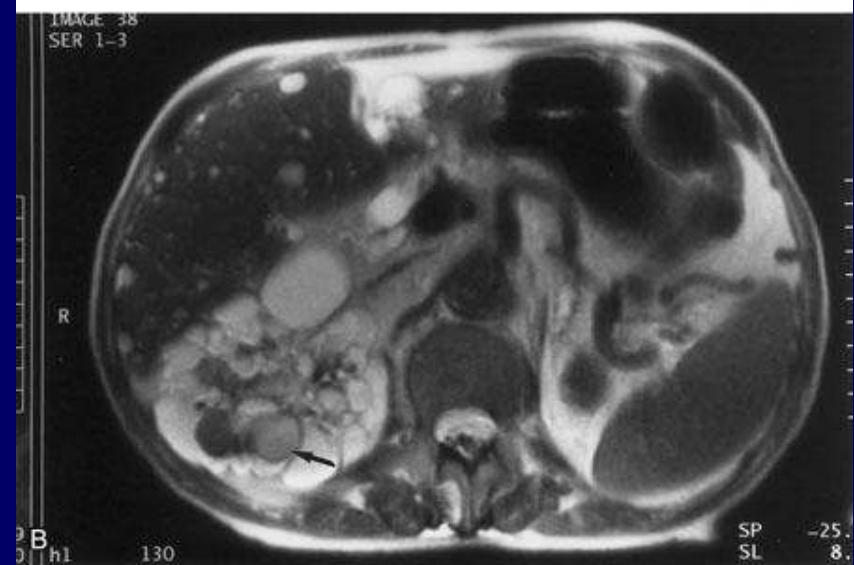
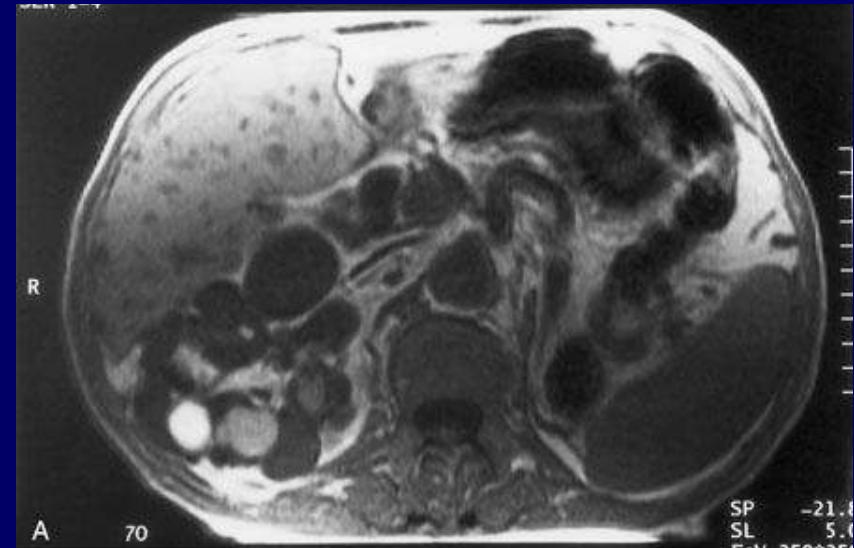
- Multikistik Displastik Bobrek
- Unilateral
- Uzum salkımı kistler
- Atretik ureter
- Abdominal kitle
- Spontan rezolusyon



Bobrek Anomalileri

➤ Kistik Hastalıklar:

- Otozomal Dominant Polikistik Bobrek
- 1/400-1000
- 30-50 yaş
- K/ciger pankreas
dalak A/ciger
berry anevriz.
- KBY



Bobrek Anomalileri

➤ Kistik Hastalıklar:

➤ Otozomal Resesif Polikistik Bobrek

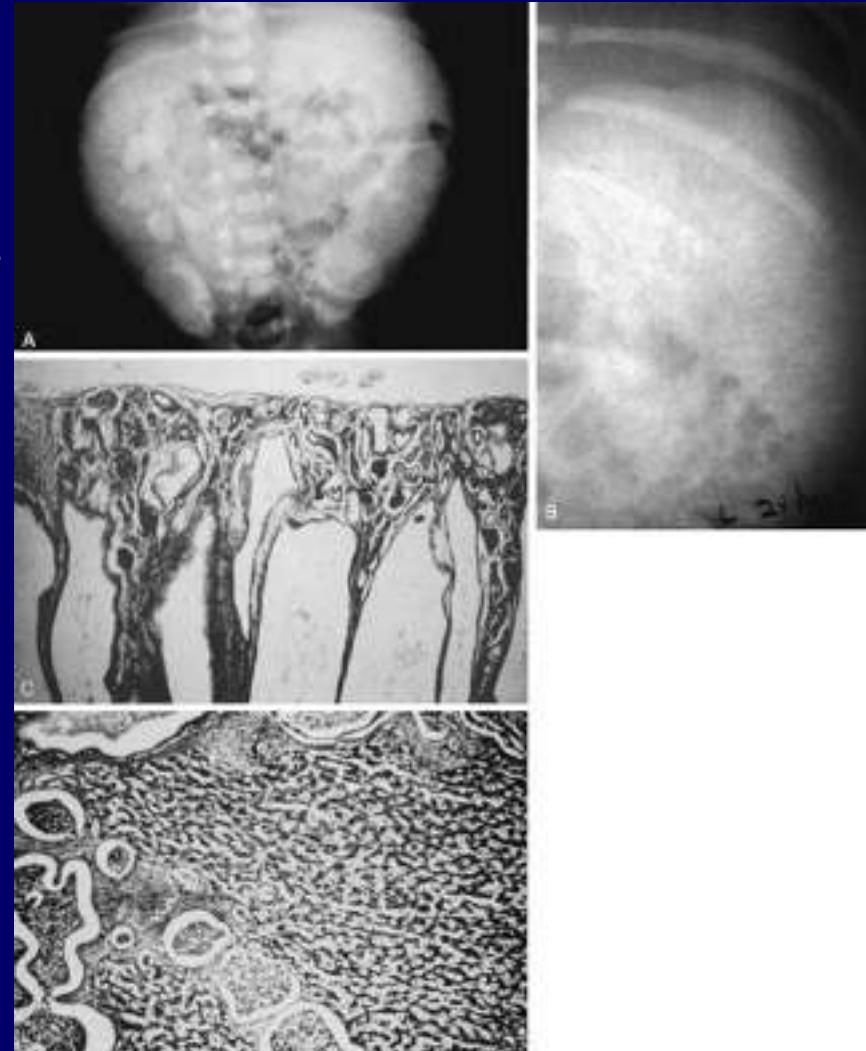
➤ 1/40.000

➤ Perinatal, neonatal,
infantil, juvenil

➤ Konjenital periportal fibrozis
hepatik fibroz

➤ KBY-portal hipertansiyon

➤ Yüksek mortalite



Bobrek Anomalileri

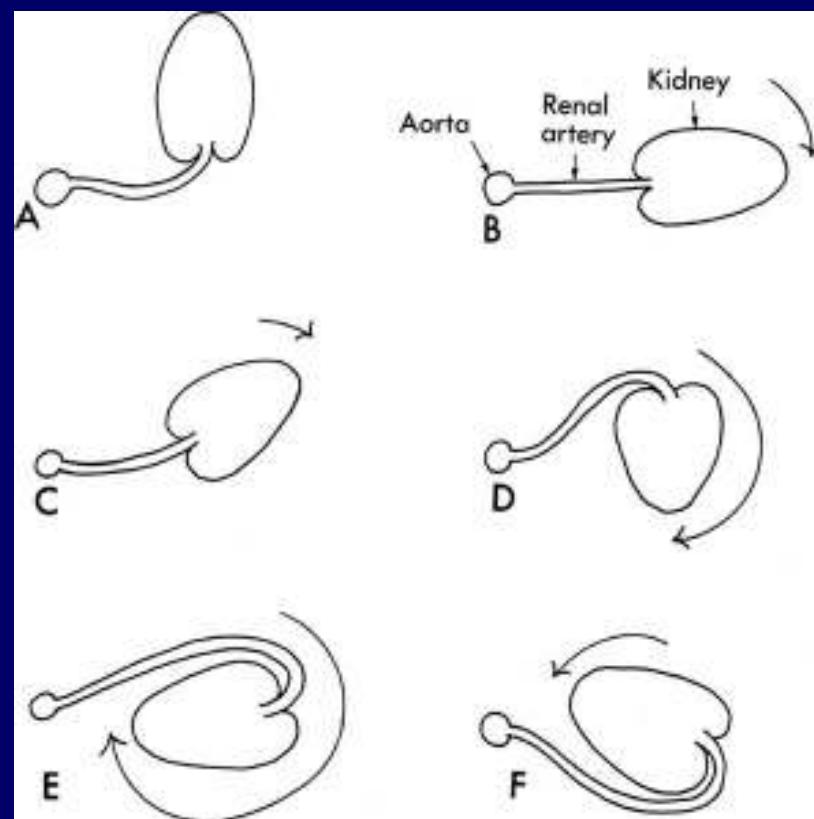
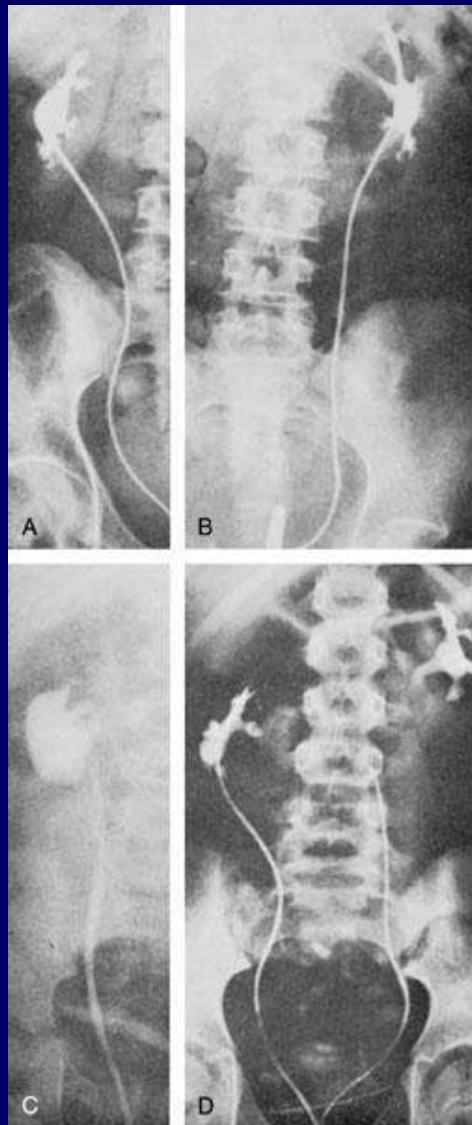
➤ Kistik Hastalıklar:

- Meduller Sunger Bobregi
- 1/500-20.000
- 20 yas sonrası
- Kollektor tublerin dilatasyonu ve çevresinde kistler
- Hematuri-tas-IYE
- IVP
- Normal yaşam beklenisi



Bobrek Anomalileri

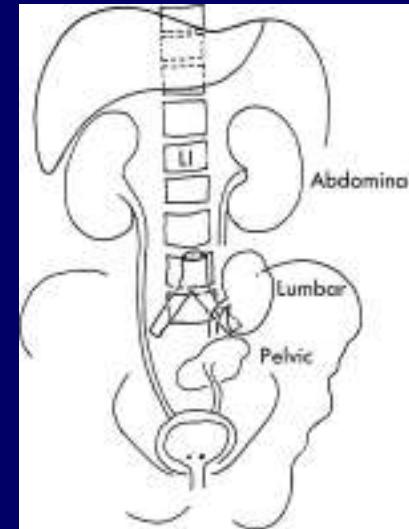
➤ Malrotasyon (en sık vertikal aks malr.)



Bobrek Anomalileri

➤ Renal ektopi (en sık pelvik bobrek)

- Tas
- UP bileske darlığı
- Vezikoureteral reflu



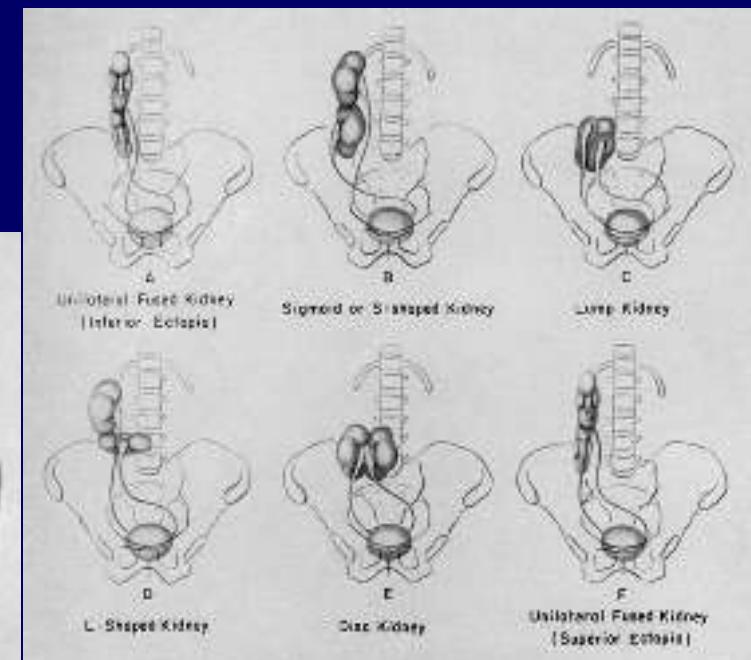
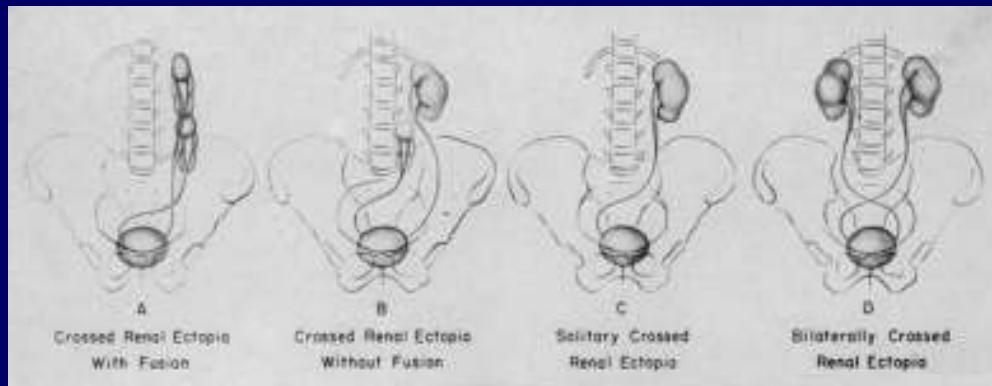
➤ Intratorasik bobrek



Bobrek Anomalileri

➤ Fuzyon anomalileri

- Ureter orifisleri normal lokalizasyonda
- En sık ektopik bubrek diğer bubrekin inferorunda



Bobrek Anomalileri

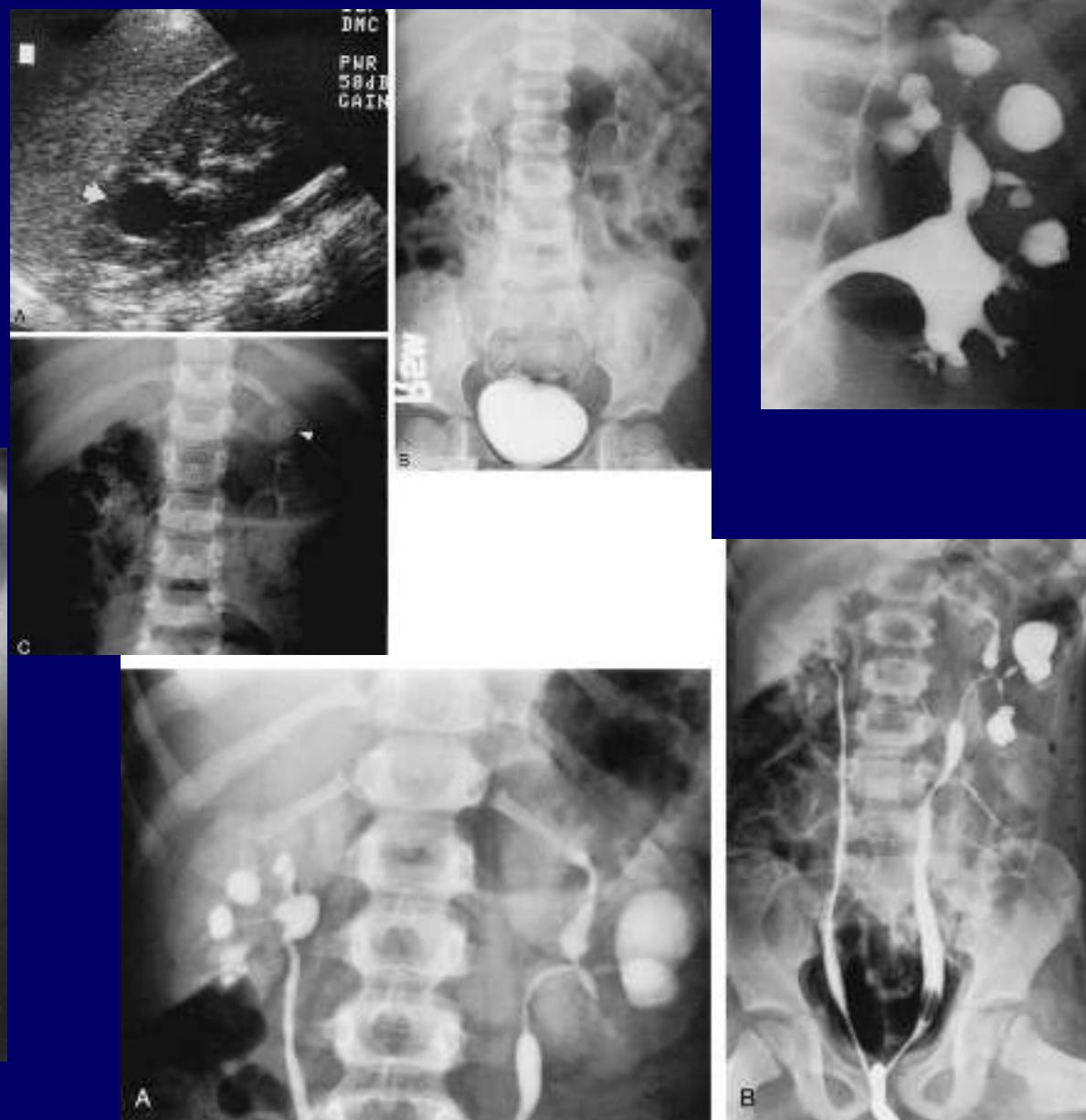
➤ Atnali bobrek

- 1/400
- Alt pollerde fuzyon (%5 ust pol)
- Ekstansiyonda istmus basisi ile agri (Rovsing belirtisi)
- Tas, UPJ darligi, VUR, hematuri, IYE, Neoplazi(?)
- Dusuk bobrek, mediale donmus aks, ureterler orta hatta yakin



Kaliks Anomalileri

- Kalisiyel divertikul
- Konjenital hidrokaliks
- Konjenital kalikosis



Kaliks Anomalileri

- Ekstrarenal kaliks
- Infundibulopelvik Disgenez



Renal Pelvis Anomalileri

➤ Intrarenal-Ekstrarenal pelvis



➤ Bifid Renal Pelvis

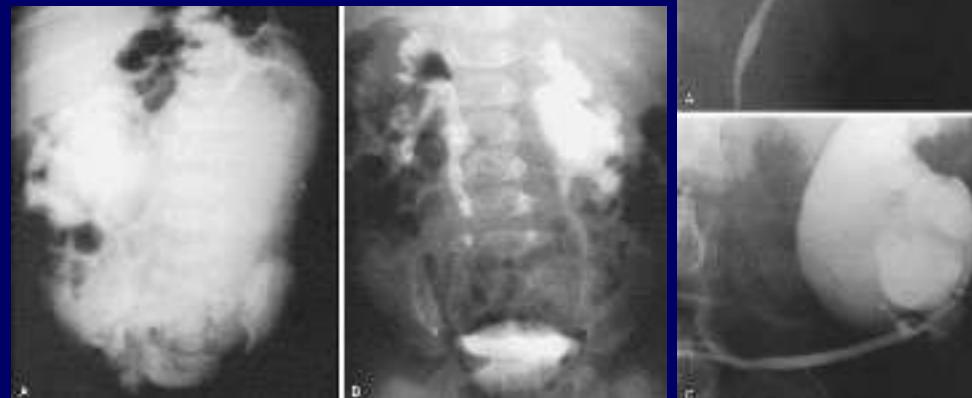
➤ Ureteropelvik Bileske Darligi (UPJ)

➤ %10 bilateral

➤ Intrinsik (adinamik segment)
ve Ekstrinsik (damar basisi)

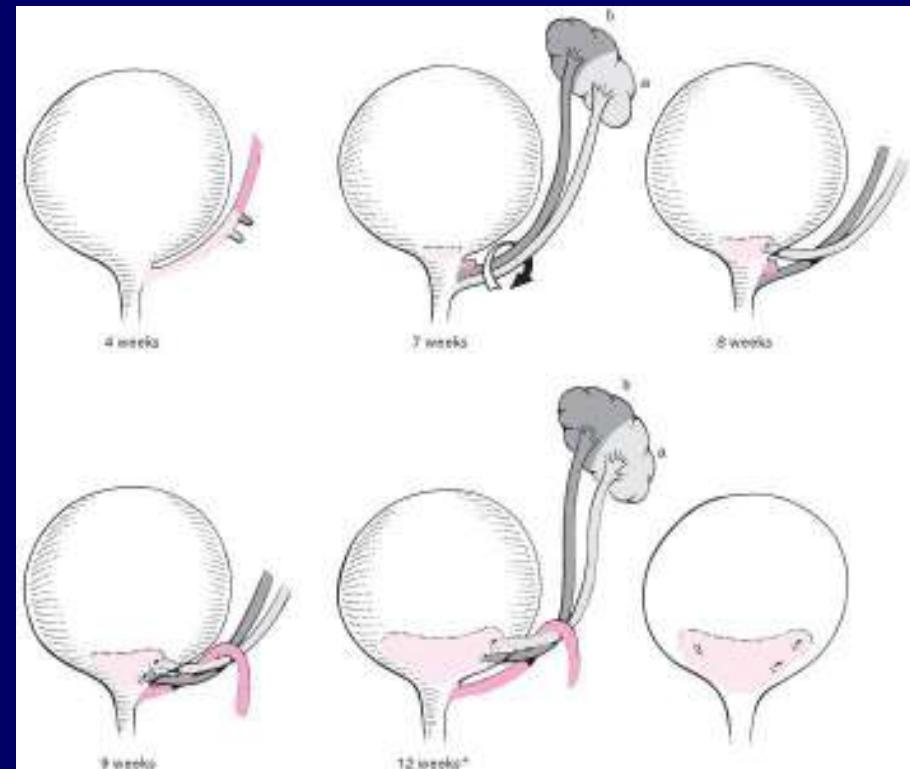
➤ Tas, Enfeksiyon

➤ Cerrahi veya takip



Ureter Anomalileri

- Ureter atrezisi ve hipoplazisi
- Ureter Duplikasyonu
 - Üst pol ureteri alta-Alt pol ureteri uste (Weigert-Meyer kanunu)
 - Alt pol ureterinde VUR-üst pol ureterinde ureterosel



Ureter Anomalileri

➤ Megaureter

- Obstruktif-refluksif-nonobstruktif/nonrefluksit
- %15-25 bilateral



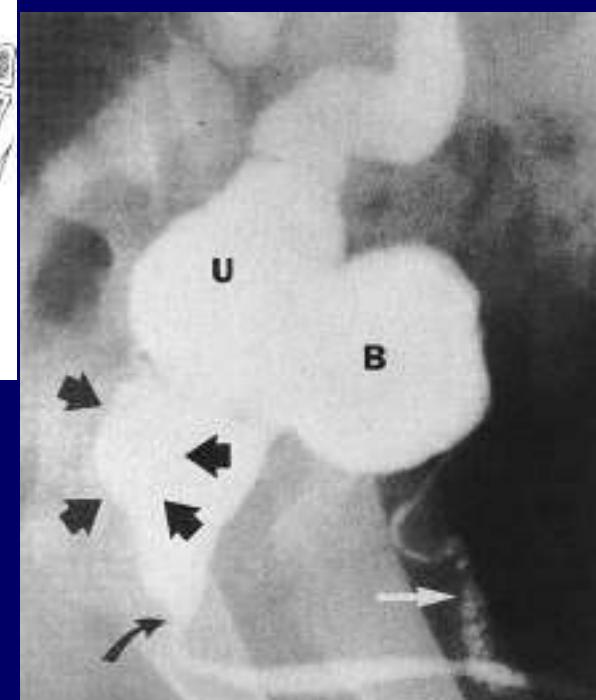
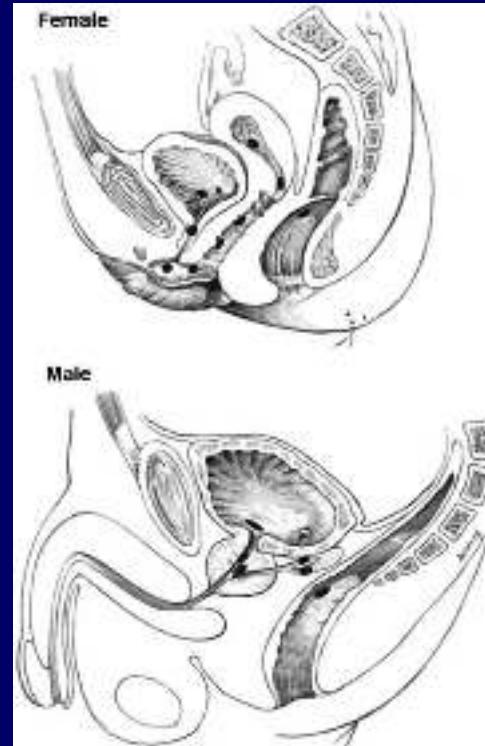
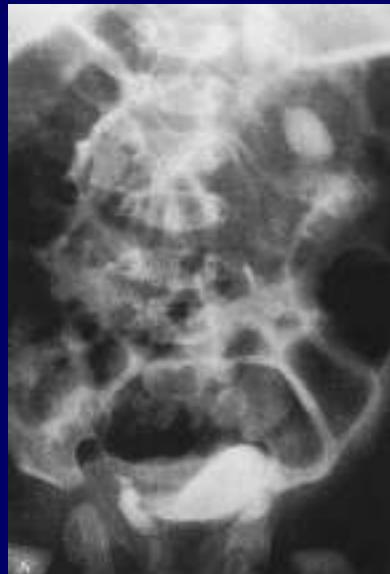
➤ Retrokaval Ureter

- Ters J belirtisi



Ureter Anomalileri

➤ Ektopik Ureter



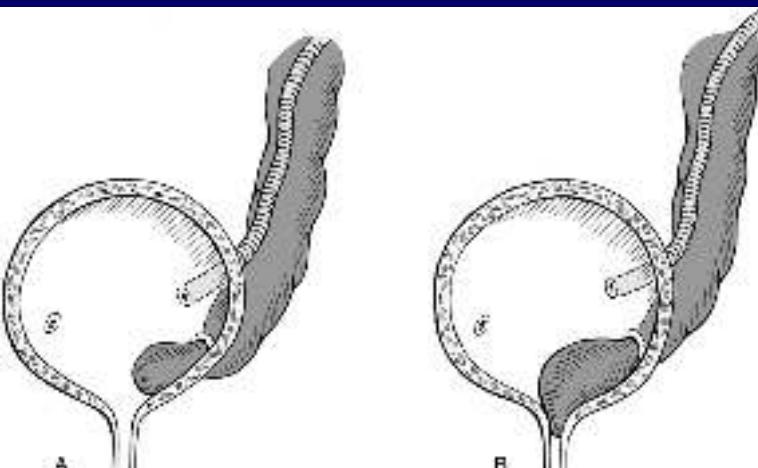
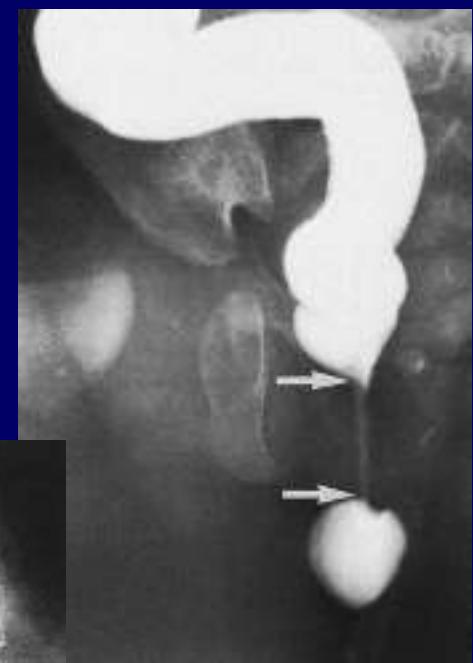
Ureter Anomalileri

➤ Ureterosel

➤ Ureterin mesane icindeki mukoza ve basal membrandan olusan kistik dilatasyon

➤ Intravezikal -Ekstravezikal

➤ Kobra bası belirtisi



Mesane Anomalileri

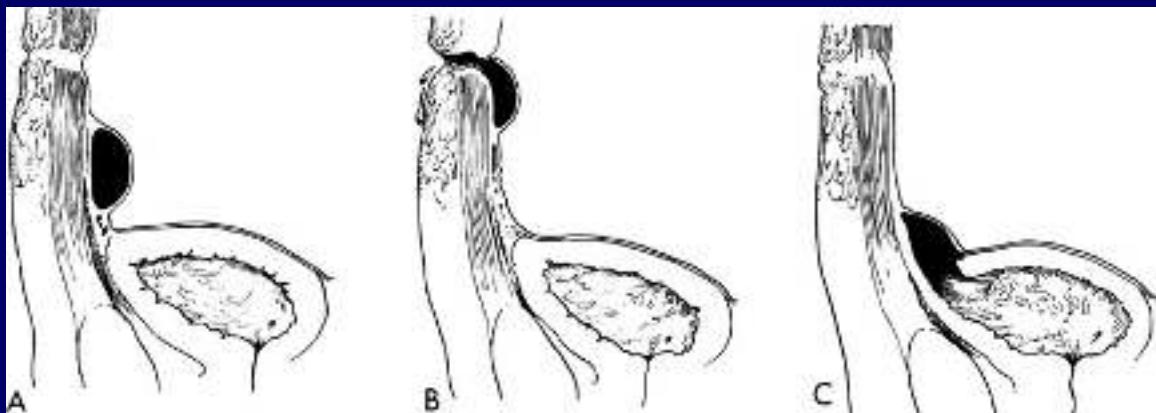
➤ Ekstrofia vezikale

- 1/30.000
- Epispadias ile beraber
- Adenokanser riski



➤ Patent Urakus

- Urakus divertikulu
- Urakus kisti
- Urakus fistulu



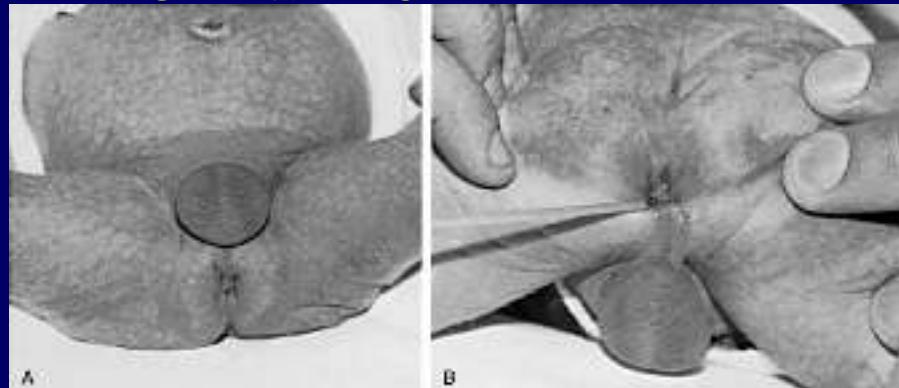
Mesane Anomalileri

- Prune-Belly Sendromu
 - Karin kaslarinin yoklugu
 - Uriner sistem anomalileri
 - Bilateral kriptorsidizm



Penis ve Uretra Anomalileri

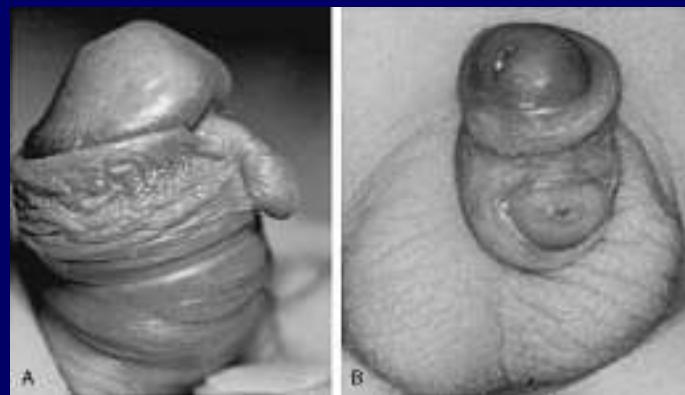
- Penis agenezi(Afallia)



- Mikropenis (yenidogan $<1.9\text{cm}$)
 $<2, 5 \text{ SD}$



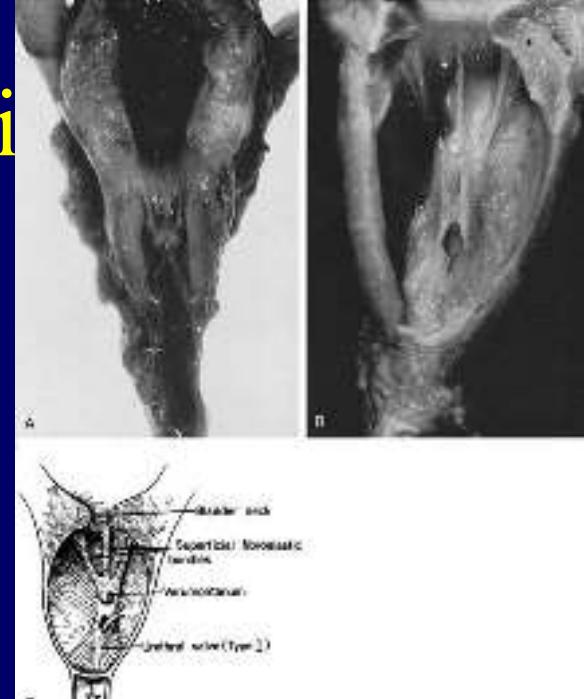
- Penil duplikasyon



Penis ve Uretra Anomalileri

➤ Posterior Uretral Valv

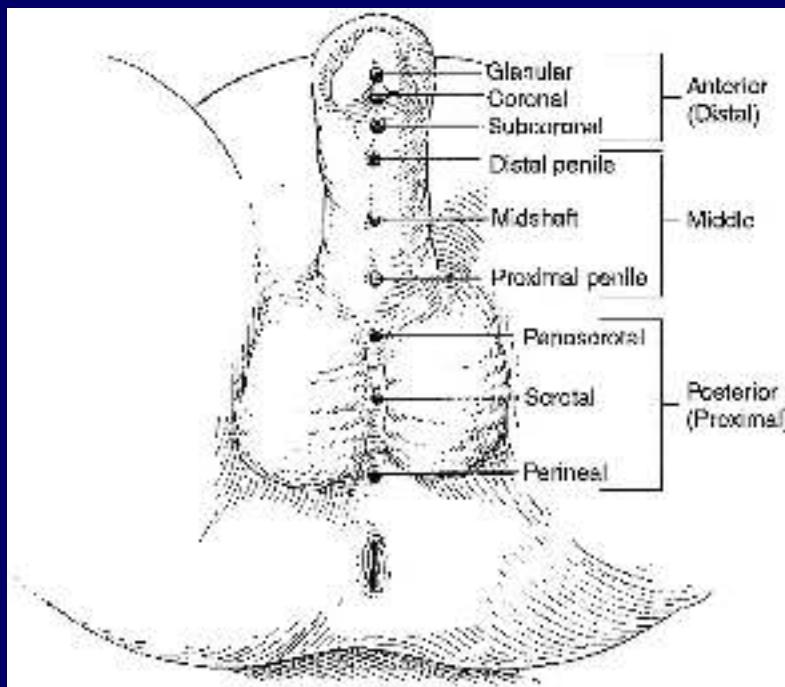
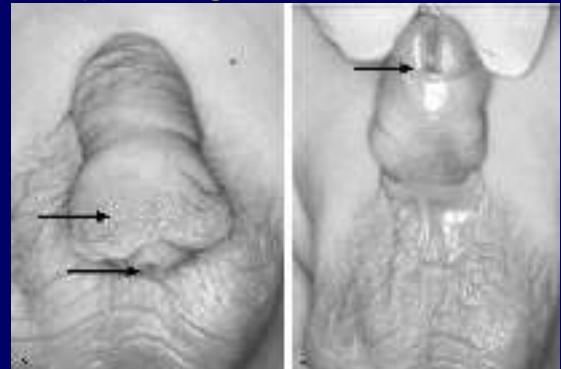
- Retrograd işlem rahat ama iseme güç
- Voiding sistouretrografi
- Endoskopik tedavi ya da vezikostomi
- Kronik böbrek yetmezliği



Penis ve Uretra Anomalileri

➤ Hipospadias

- Ventral urethral mea
- En sik anterior yerlesimli
- 1/300
- Idrar akiminda bozulma, zor ejakulasyon, kordi, kozmetik
- Inmemis testis ile beraberse interseks?
- Sunnet kontraendikedir



Penis ve Uretra Anomalileri

- Epispadias
Kontinans problemi



- Uretral divertikul
- Megalouretra

Genital Anomaliler

➤ Skrotal hipoplazi
(testis yoklugu ile beraber)



➤ Skrotal ektopi



➤ Bifid skrotum ve
skrotal transpozisyon



Genital Anomaliler

➤ Testikuler Hipoplazi

➤ Kryptorsidizm

Term %3.5

Pre-term %30

3. Aya kadar testislerin cogu iner!!!

3. ay %1

Eriskin %0.7

Palpal Kryptorsid testis (%80)

➤ Inmemis testis (desensus duraklamasi)

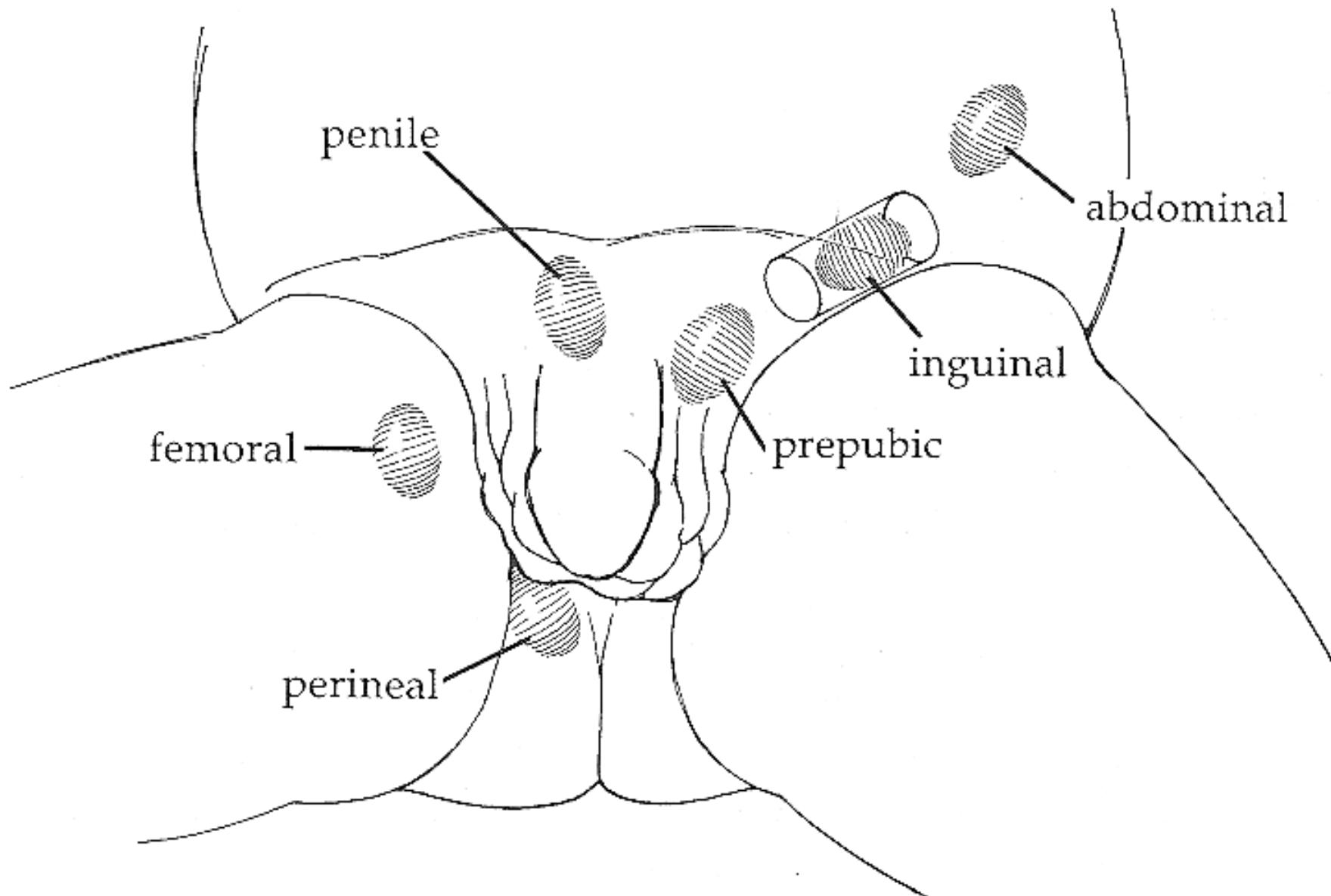
➤ Ektopik testis (Desensus sonrasi hatali yerlesim)

superfisiyel inguinal pos!!!!!, femoral, pubik, penopubik, penil, perineal, veya kontralateral skrotum

➤ Retraktif testis (hiperaktif kremasterik refleks)

Ectopic

Partially Descended



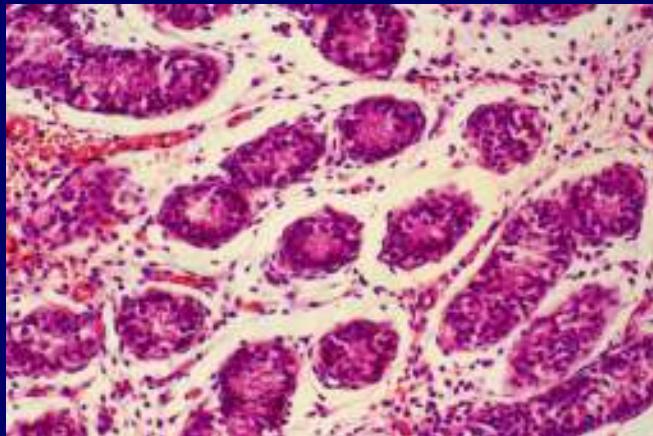
- **Unilateral (68%) vs. Bilateral (32%)**
- **Sag (70%) vs. sol (30%)**
- **Ust uriner sistem anomalisi yok**
- **%30-79 epidimal veya vas anomalileri ile beraber**

- **Palpe edilemeyen Kriptorsid testes (20%)**
 - **Intraabdominal:** genelde internal ringde ama her zaman abdomende
 - **olmayan (4%):** torsiyona bagli (yokolmus) veya agenez
- **Bilateral inmemis testis (10-25%)**

Konjenital adrenal hiperplazi gibi interseks hastaliklarinin ekrate edilmesi gereklidir

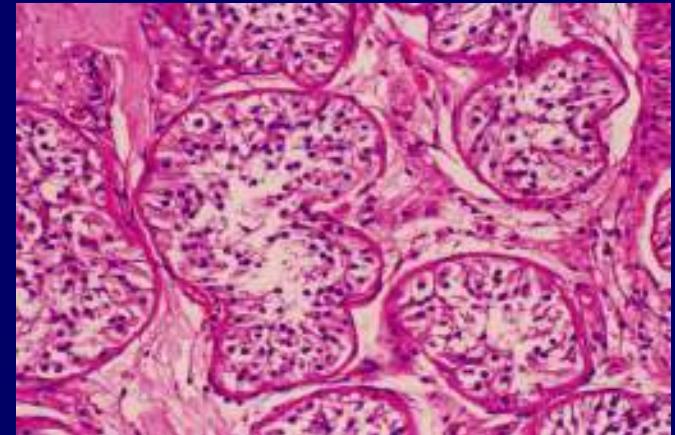
Inmemis testis histolojisi

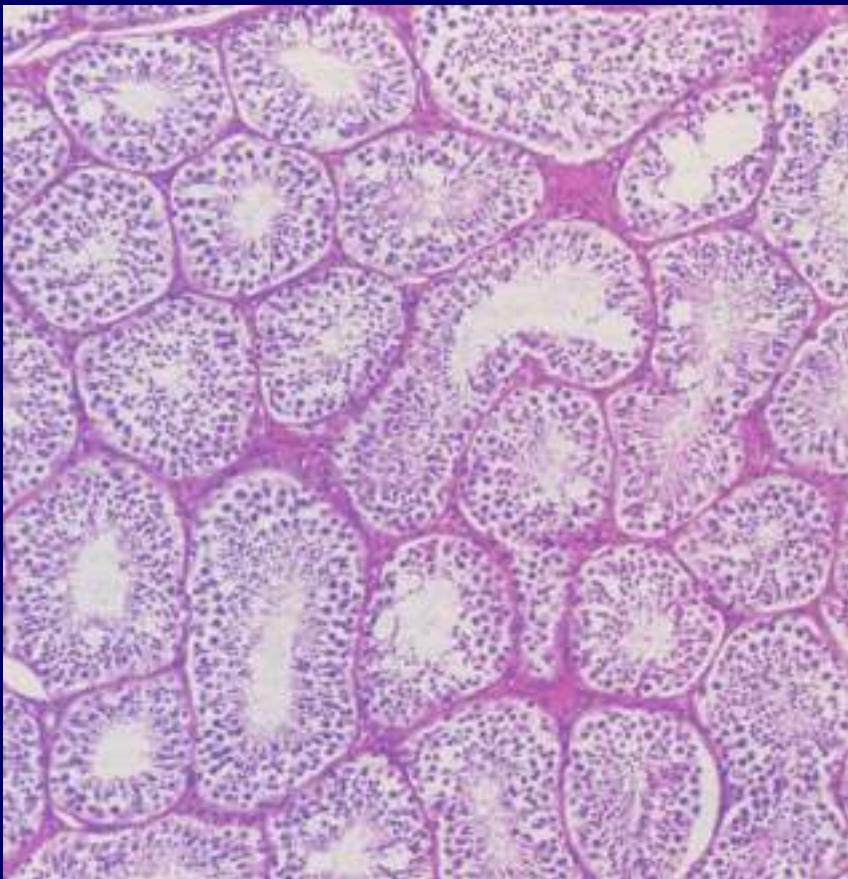
Fetal



Prepubertal

Pubertal

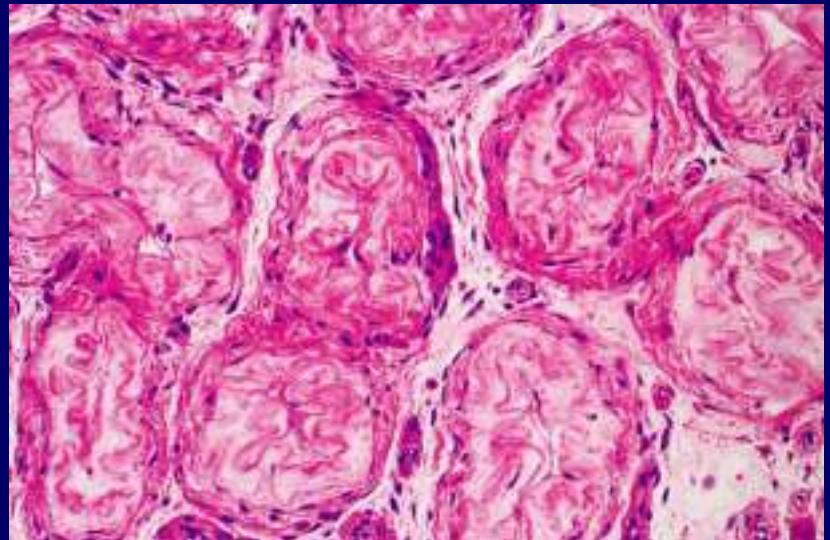




Normal

Inmemis t. histolojisi

Postpubertal UDT

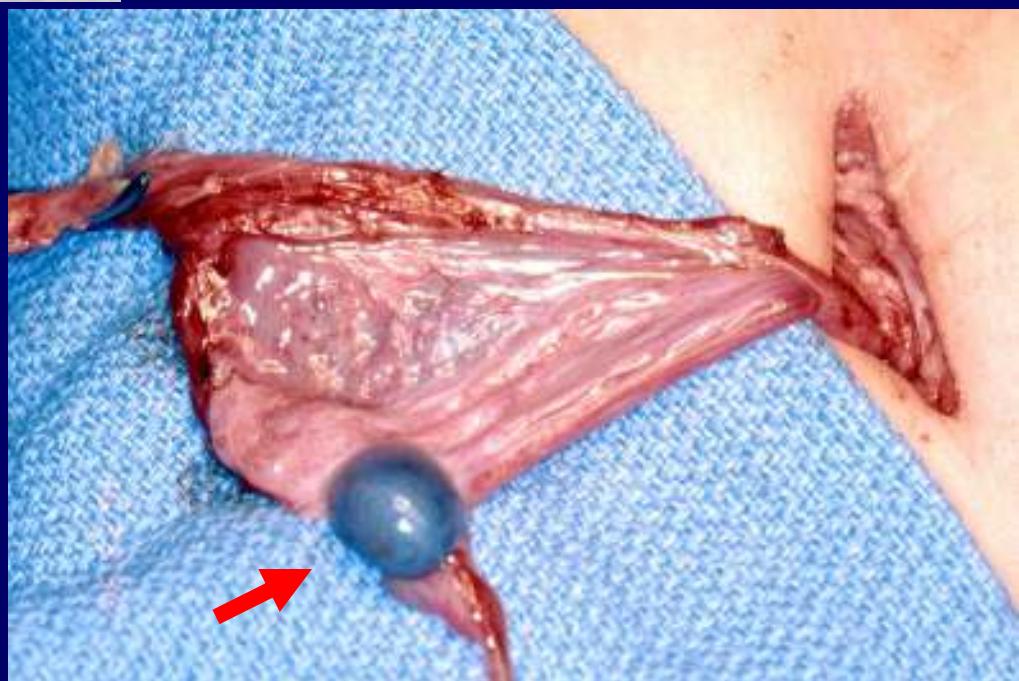


Tedavi Endikasyonları

- **Fertilite:**
 - Histolojik degisimler 18 aydan sonra izlenir
 - Orsiopeksi sonrasi bile %50-70 fertilite problemleri olur
- **Malignensi:**
 - 40x risk artar
 - Orsiopeksi riski azaltmaz ama erken taninmasini saglar



Inmemis testiste torsiyon



Tedavi Endikasyonlari

- **Inguinal herni:** %90ninda herni vardir
- **Torsiyon:** riski yuksektir
- **Travma:** lokalizasyondan oturu
- **Kozmetik**

Tedavi

- genellikle 1-2 yas arasi
- Hormonal:
 - HCG: 1500 units/m²; 19% basari orani
 - LHRH: 1.2mg/d; 21% basari orani
 - 10-25% tekrar cikar

tedavi

- cerrahi: acik veya laparoskopik
 - basari orani:
 - Abdominal 74%
 - Internal ring 82%
 - Kanalikuler 87%
 - Eksternal Ring 92%

Inmemis Testiste Laparoskopi

- **USG ve MRI dan daha hassas** (Al-shareef et al., 1996)
- **Kesinlik 88-100%** (Froeling et al., 1994)

Damarlar ve Vas Cikista

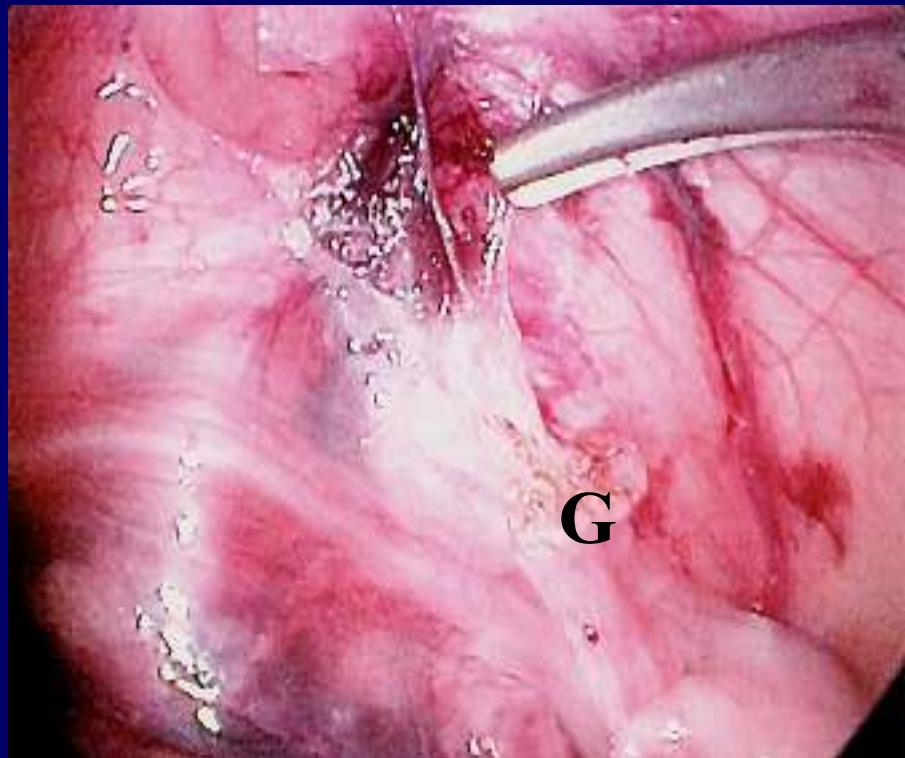


Cikista
Gubernakulum

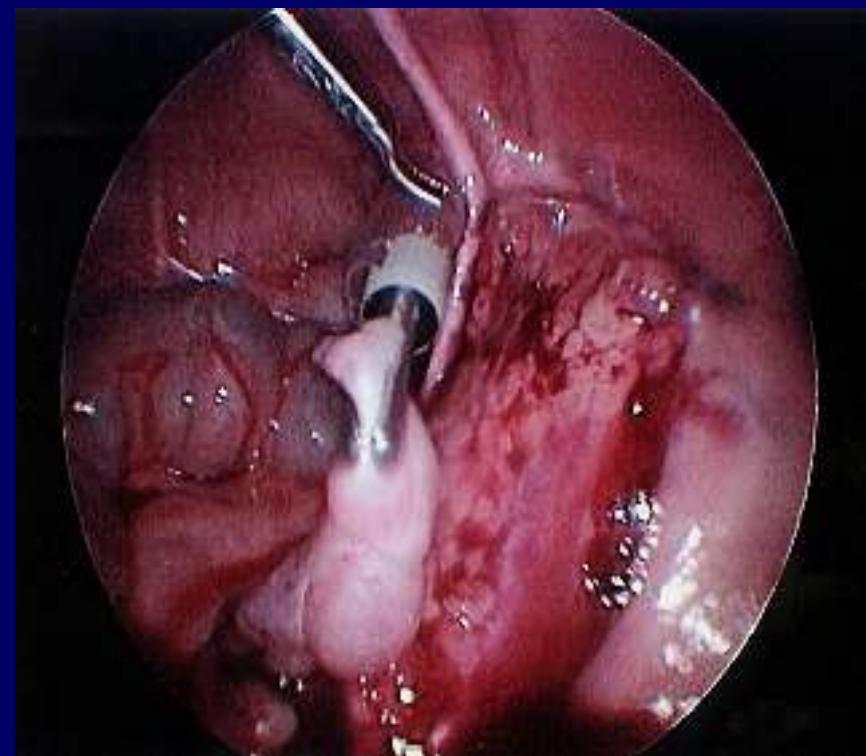


Atretik damarlar ve Vas





Laparoskopik Orsiopeksi



**Yenidogan muayenesinde penoskrotal
hipospadias saptadiniz. Bundan
sonraki basamakta yapacagini sey
nedir?**

- A. Testislerin palpasyonu
- B. Uterus icin abdominal palpasyon
- C. Uterus icin abdominal USG
- D. Serum testosterone seviyesi
- E. Ileri arastirmaya gerek yok



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**Skrotumda testisleri palpe
edemediniz. Bir sonraki basamagini
ne olmalıdır?**

- A. Serum elektrolitlerinin tayini
- B. Kromozomal analiz
- C. Uterus icin abdominal USG
- D. Serum OH-progesterone seviyesi
- E. Pediatrik urolojiye hipospadias
onarimi icin 6 aylikken gonderme

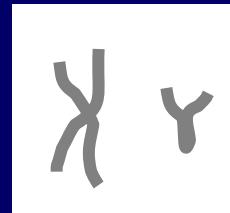


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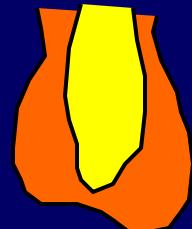
Kromosomal seks



Gonadal seks



Fenotipik seks

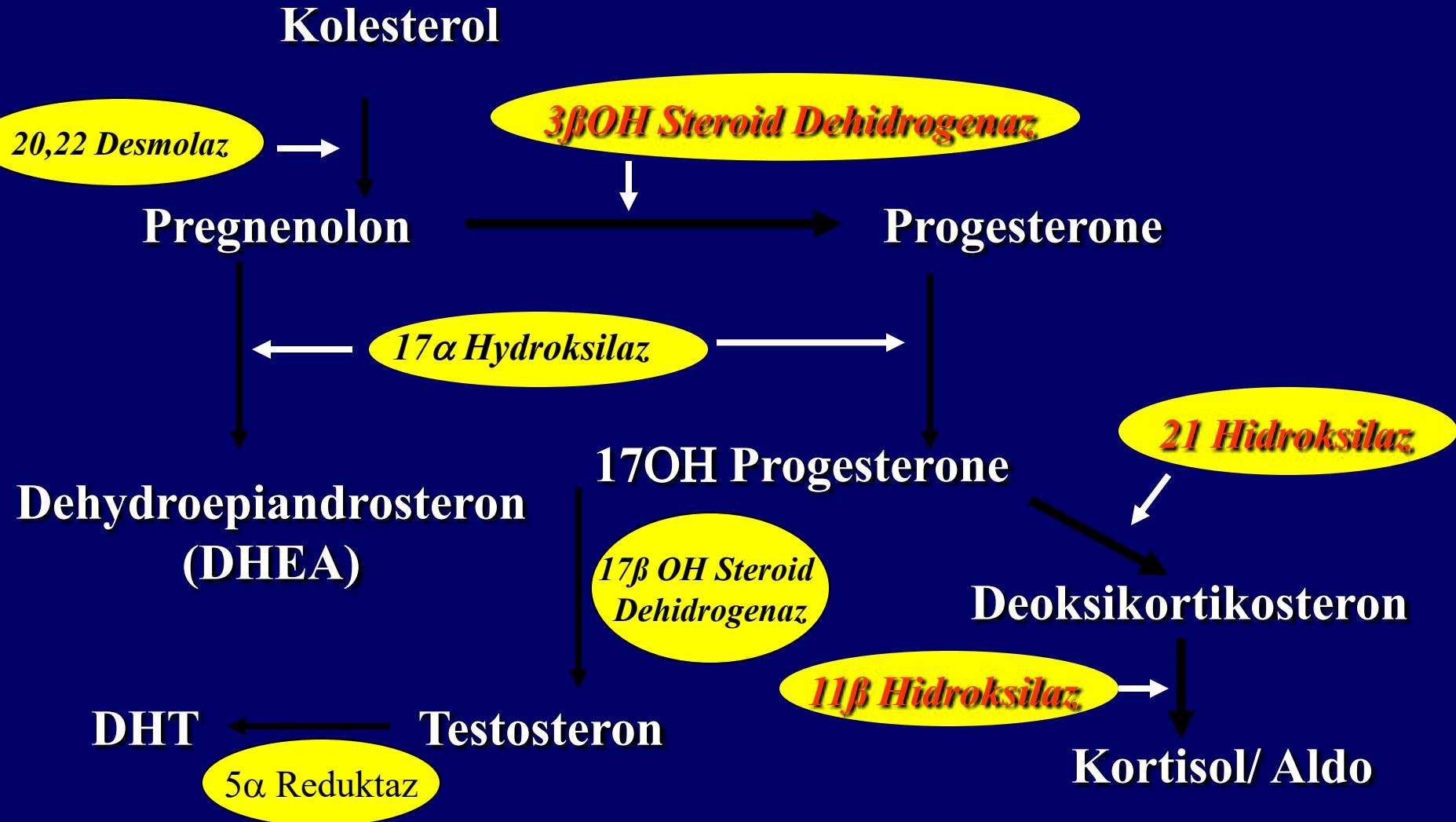


Yenidoganlarda Tanimlanamayan Genital

- **Disi Psodohermafrodit**
 - Konjenital Adrenal Hyperplazi
- **Erkek Psodohermafrodit**
 - Androjen Aktivitesi Anomalisi
- **Karisik Gonadal Disgenez**
- **Gercek Hermafrodit**



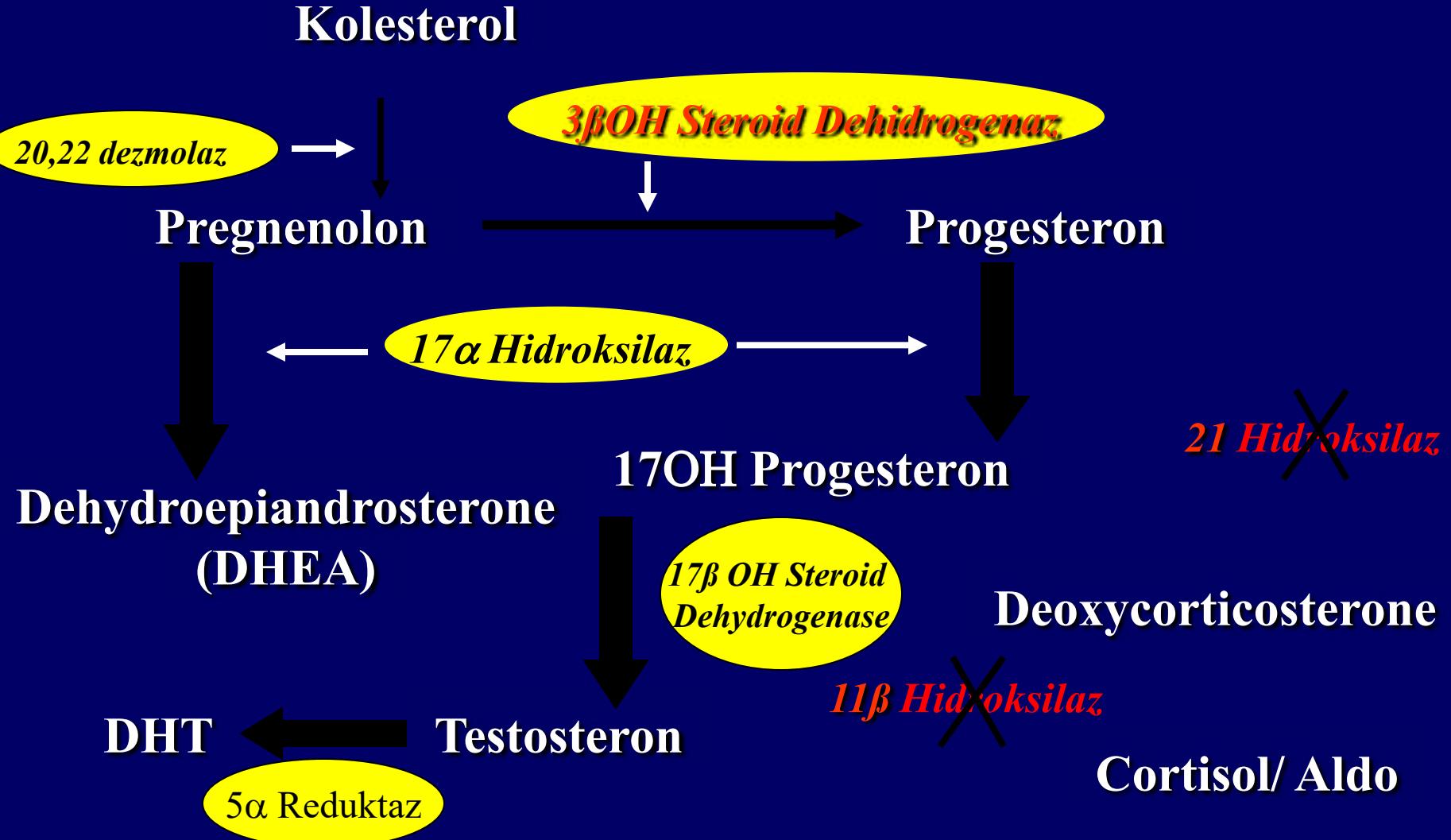
Kortizol Sentezinde Enzimatik Yollar



Disi Psodohermaphrodit: 46 XX

- Ekzojen androgen karsilasmasi
- Conjenital Adrenal Hyperplazi
 - steroid sentez yolundaki 5 enzimden biri genlerinde mutasyon sonucu, kortizol olusumda bozukluk
 - 21 and 11b-hidroksilaz maskulinize disiye neden olur ancak etkilenmis erkeklerde kendini gosteremez

Disi Psodohermaphroditizmde Enzimatik Blok





Pre-op



Intra-op



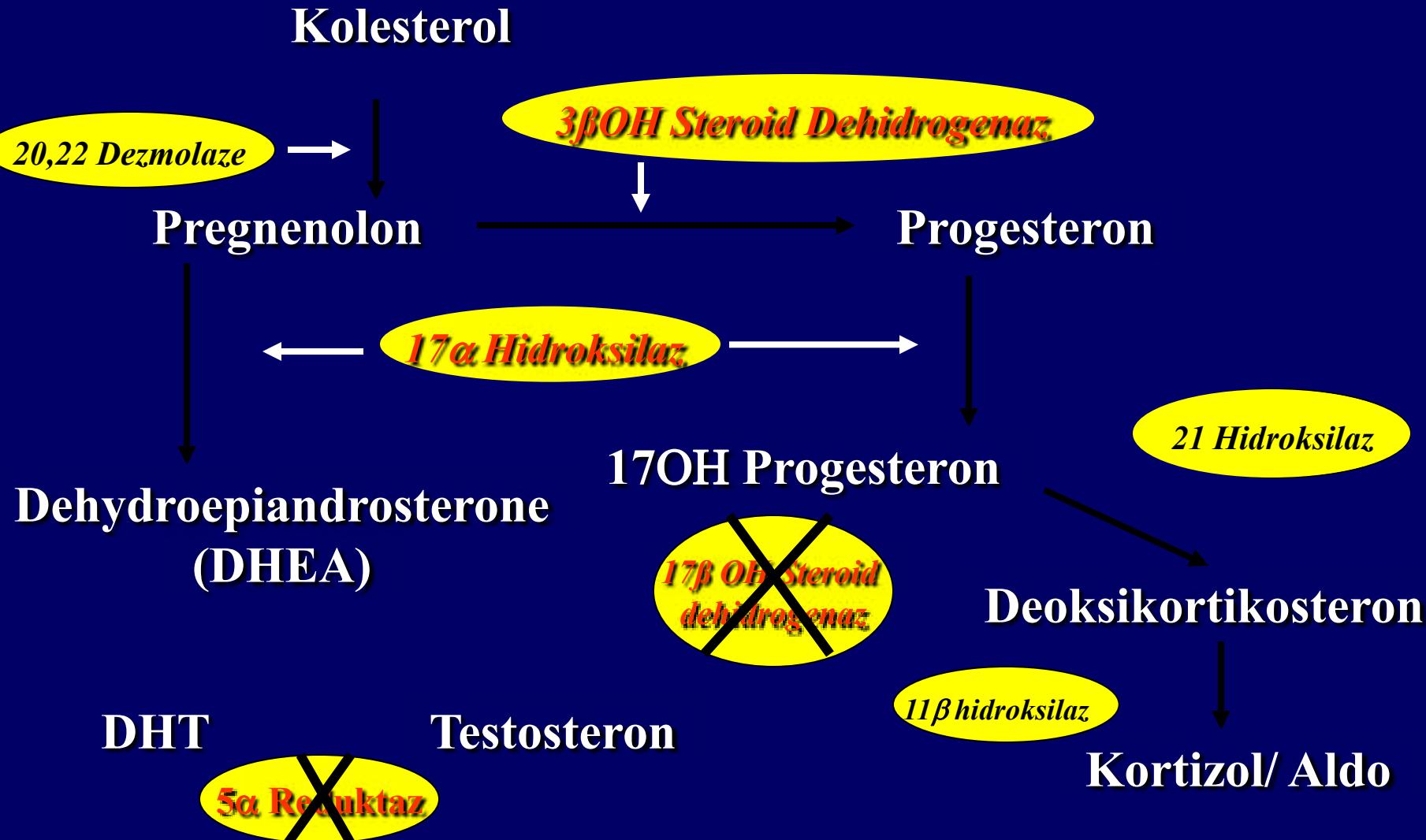
Post-op

KAH

Erkek Psodohermafrodit: 46 XY

- Testosteron yapilamaz (enzymes defect)
- Testosteron dihydrotestosterona
cevrilemez
- Androjen reseptör defekti
- Persistan Mullerian Duct sendromu

Erkek Psodohermaphroditizmde Enzimatik Blok

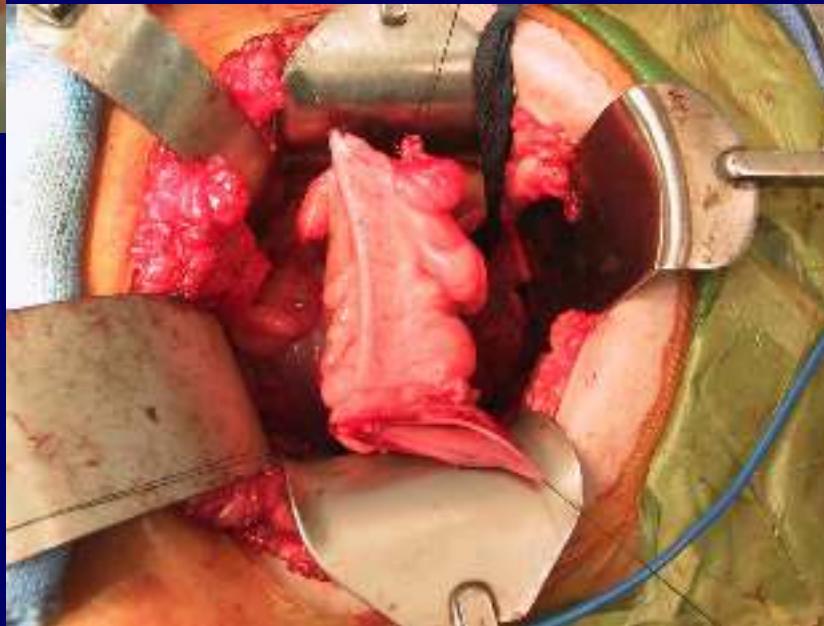


Testikuler Feminizasyon

Sigmoid Vagina



Pre-op



Post-op



Karisik Gonadal Disgenez: 46 XY/ 45 X

- Bir çizgi ve bir disgenetik gonad
- Genital: inkomplet virilizasyondan hipospadias ve inmeis testise kadar
- Tedavi kromozom ve virilizasyonun derecesine bağlıdır.

Karisik Gonadal Disgenez



Pre-op



Post-op



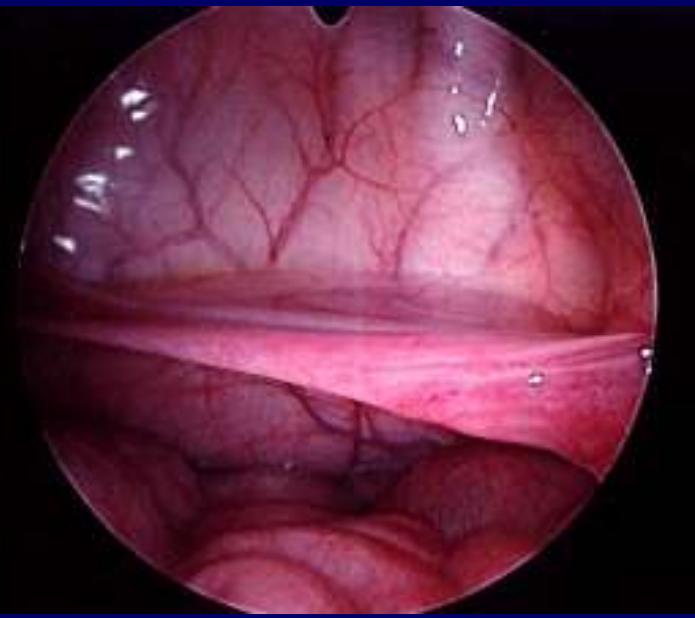
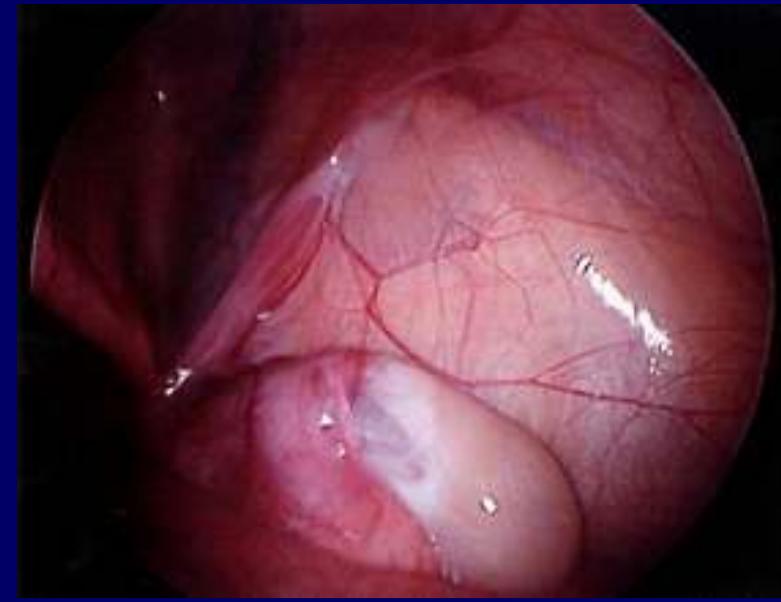
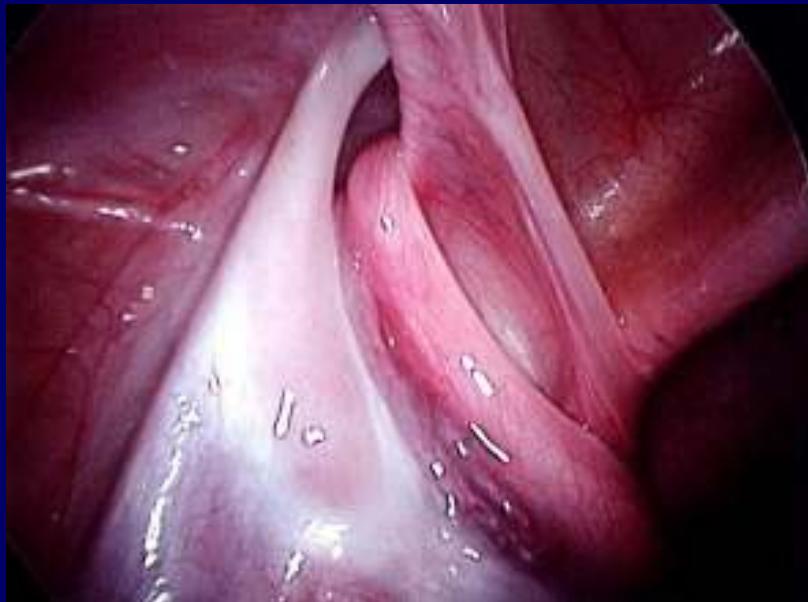
Gercek Hermafrodit: 46XX, 46 XY veya mosaik

- ovotestis, over and testis kombinasyonlari
- Vagina and uterus hastalarin %80-100 de mevcuttur.
- Cinsiyet tayini tartismalidir

Gercek Hermafrodit



Gercek Hermafrodit



Tanımlanamayan Genitalia: İlk Degerlendirme

- **Hikaye: Maternal and Aile**
- **Fizik Inceleme**
- **Kromosomal seks**
- **Metabolik tetkikler**
- **Goruntuleme**
- **Biopsi**

Tanımlanamayan Genital: Fizik İnceleme

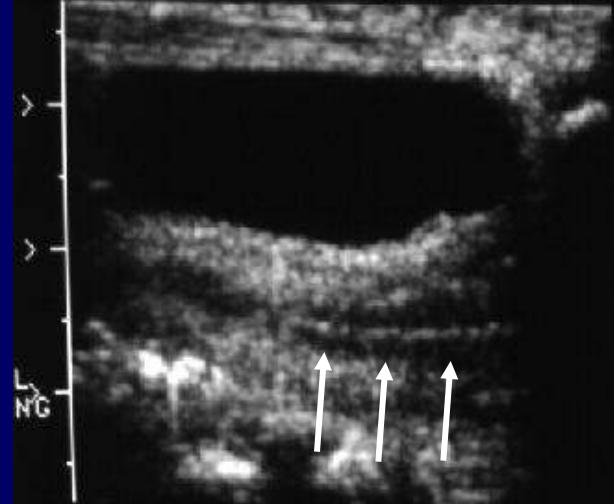
- Genel ozellikler: ? sendromu
- Fallus
 - Konfigurasyon: Kordi, hipospadias, korporal doku
 - boyutlar: gerilmis uzunluk, genislik
- Labioskrotal yapılar
 - fuzyon
 - Pigmentasyon
 - Gonadlar palpe edilebiliyor mu?
- Urogenital sinus/perineum - tek delik?
- Abdominal inceleme - rektal muayenede uterus palpabl mi?



Tanimlanamayan Genital: Kromosomal degerlendirme

- Kromosom tayini hemen
- Barr body guvenilir degil
- Genel sonuc alimi 48-72 saat
- Mosaikism:
 - orn. 46,XY /45,XO
 - En sik gonadal disgenez

- Pelvik ultrasound:
uterus, gonads, adrenals



- Genitogram:



Tanımlanamayan Genital: Tani

	<u>Karyotip</u>	<u>Gonad</u>	<u>Genital</u>	<u>Uterus</u>	<u>17OH P</u>
KAH	XX	over	hipospadias	+	Ú
KGD	XY/XO	testis/cizgi	hipospadias	+/-	
MPH	XY	testis	hipo/micro	-	
G H	XX/mosaik	testis/over	hipospadias	+/-	

Tanimlanamayan Genital: tedavi

- KAH nin metabolik komplikasyonlarinin tedavisi: tuz kaybedici
- ileri degerlendirme
 - Endoskopi: urogenital sinus anatomisi
 - Gonadal biopsi: disgenetik, cizgi, ovotestis
- İleri tarihte genital cerrahi

Genital Anomaliler

➤ Kloaka Anomalileri

- Imperfore anus
- Persistan kloaka