



DSD

Disorders of **S**exual **D**ifferentiation

Disorders of **S**exual **D**evelopment

Differences of **S**exual **D**evelopment

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Outline

Basics



Approach



Summary



Classification



Surgical
Reconstruction



Basics

Atypical sexual development (congenital)

1 in 300

Have ambiguous genitalia
(including hypospadias, UDT)

1 in 15,000

Are due to CAH (70%)

1 in 5,000

Have true ambiguous
genitalia

1 in 20,000

Have 46 XY disorders

- **Incidence estimates** vary due to confusing terminology

“Hermaphrodite”



- **Greek Mythology:** Hermaphroditos was a son of Hermes and Aphrodite (gods of sexuality). Salmacis fell in love with him and prayed to be united forever. They were then merged into an androgynous form

Terminology has been controversial

- 19-20th century: hermaphrodite
 - misleading
 - stigmatizing
 - clinically problematic
- 1917: intersex >> 2006: DSD
 - Some still prefer to use the term intersex

Mismatch between the 3 components of sex

Genotype (genetic information)

Chromosomal sex



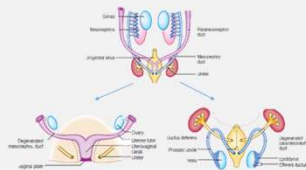
Determines gonadal sex

- **SRY gene**
- Other (*SOX, WTI, SF1*)
- ***DAX1 gene**

• **Immutable**

Phenotype (observable traits)

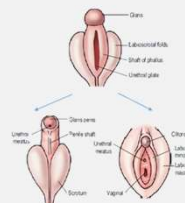
Gonadal sex



Determines Anatomic sex

- **Testosterone**
 - Internal structures
 - Androgen imprinting (Postnatal surge)
- **DHT**
 - External genitals have more affinity
- **MIS**
 - Paracrine effect (ipsilateral gonad)

Anatomic sex



• **Modifiable (hormone, surgery)**

Gender (perception)

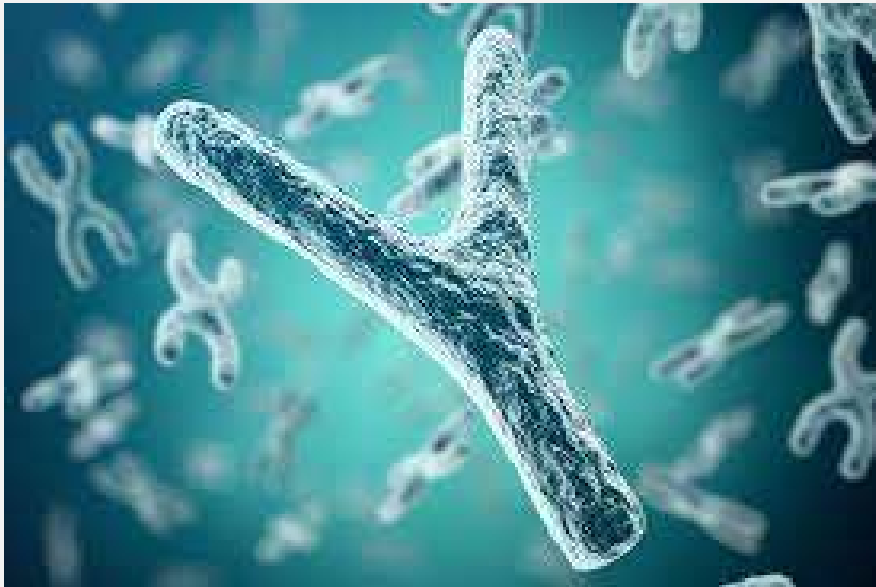
Gender Identity & trait



Determined by....

- **Social Influence**
 - parents' chosen gender of rearing, learning, socialization
- **Physiology**
 - Genes, brain structure, prenatal androgen exposure
- ****Gender dysphoria**

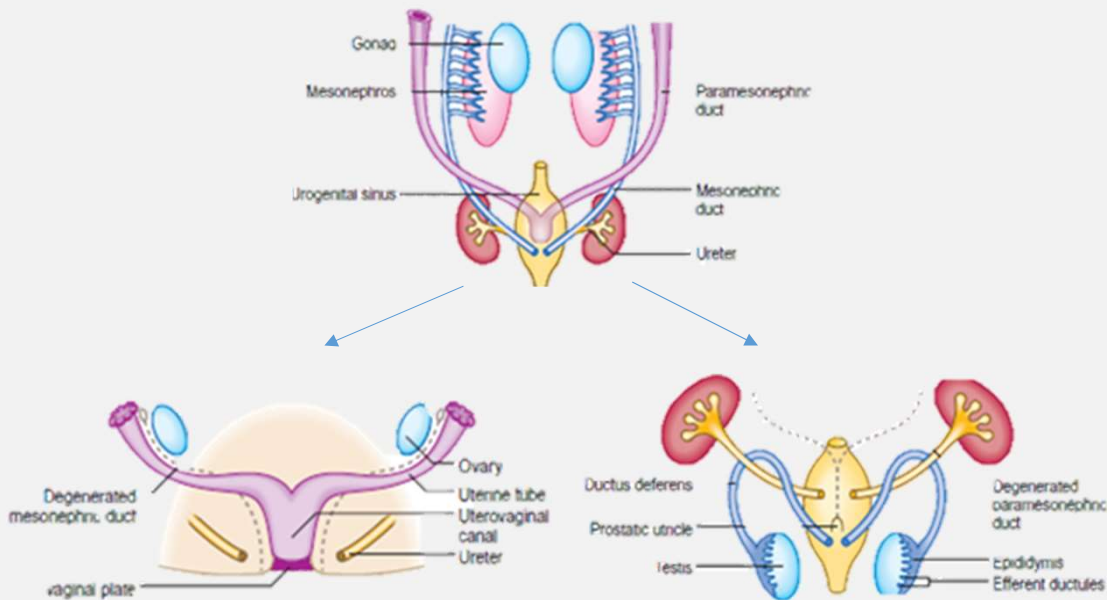
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Determines gonadal sex

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 - **DAX1* gene
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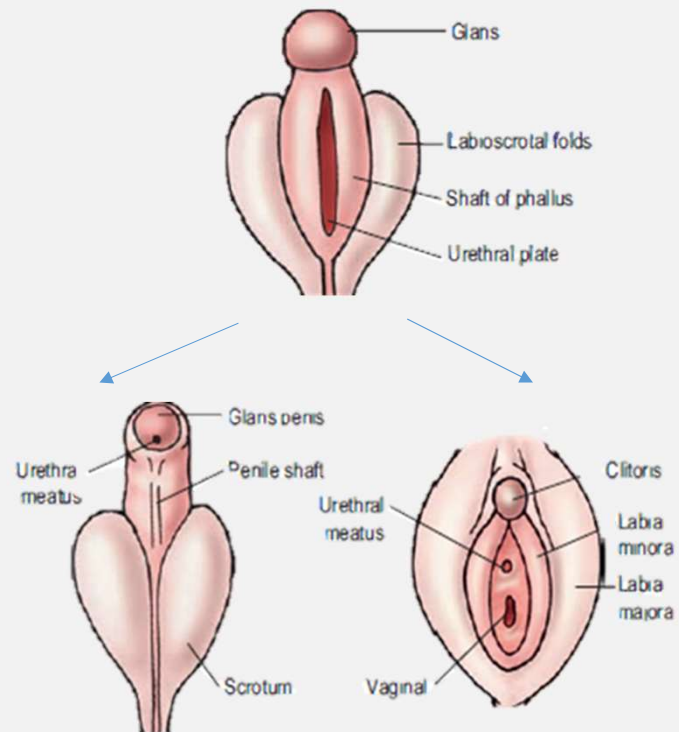
Gonadal sex



Determines Anatomic sex

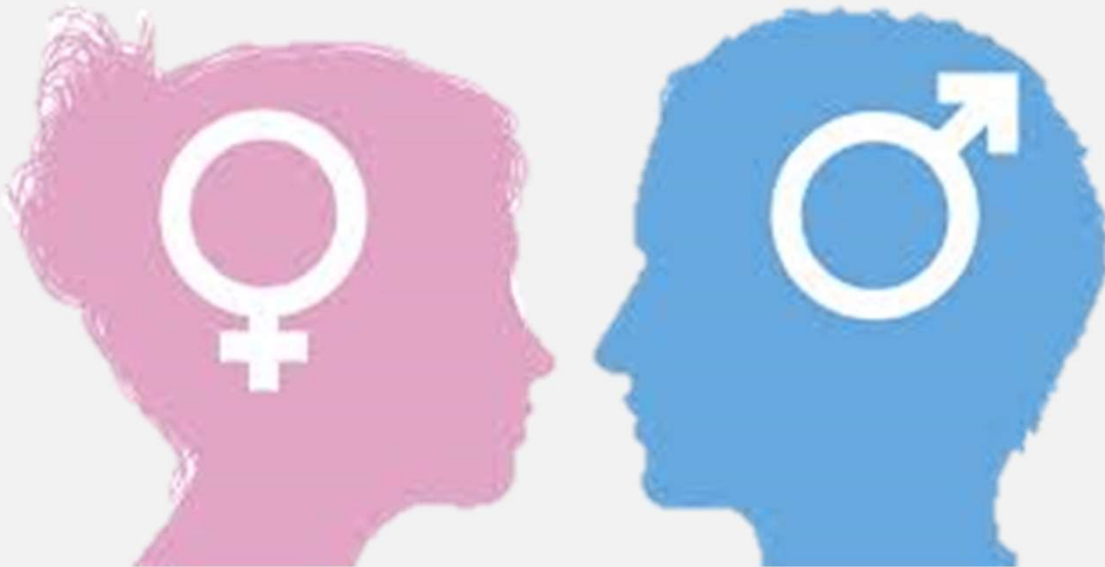
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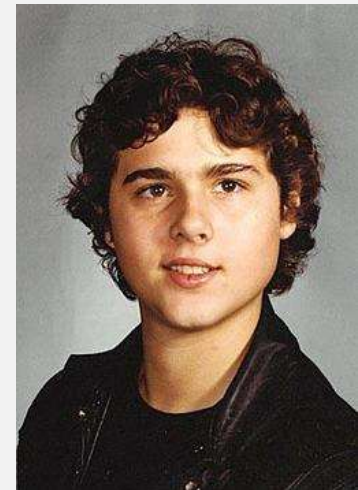
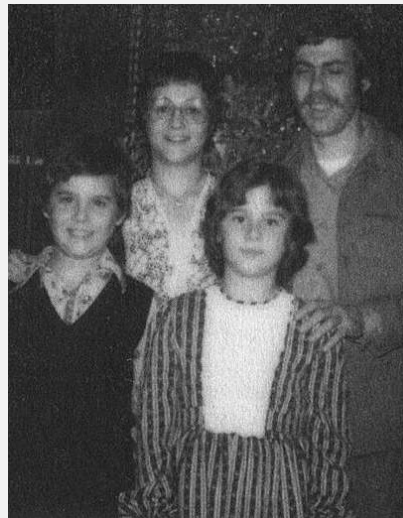
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Gender identity: Nature or Nurture?



- **The Case of John/Joan:** In the 1960s, identical male twins were born to a Canadian couple. One was reassigned as female after his penis was severely injured during circumcision in infancy. John Money reported the reassignment evidence that gender identity is primarily learned. Milton Diamond later reported child's realization he was not a girl crystallized between ages of 9 and 11 years and he transitioned to male at age 15. He committed suicide after suffering years of severe depression, financial instability, and a troubled marriage.

At what age should we perform gender confirmation surgery?

Discussion with parents

- **Consensus statement** (2006) continues to advocate **early reconstruction at 3-6 mo**
 - *** surgery b/n 12mo-adolescent is not recommended*
- **Support groups** : **better to wait for child to choose** (?)



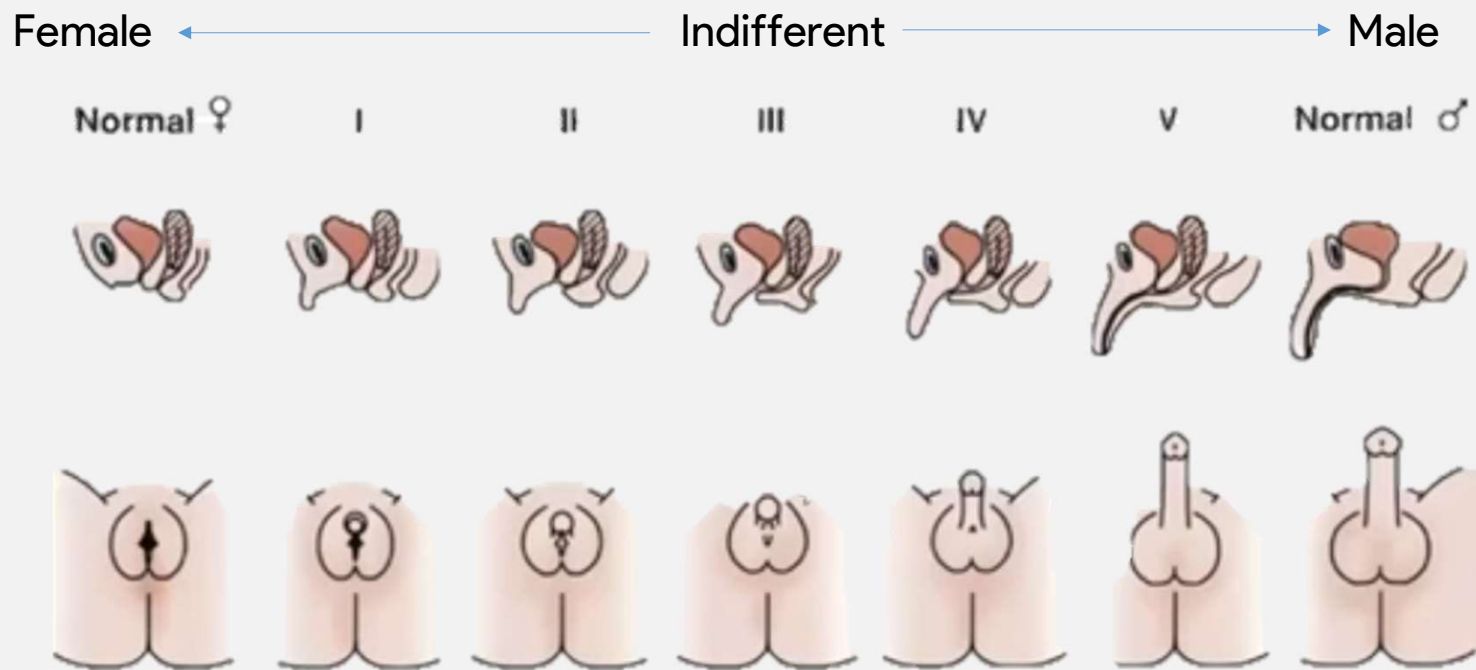
Approach

Suspicion of DSD



- Not always **overt ambiguity** (confusing/unsure). Can be **apparent male** with severe hypospadias, micropenis, non palpable gonads or **apparent female** with clitoral hypertrophy, posteriorly fused labia, palpable gonad. **May present later** during inguinal hernia repair, delayed puberty, large breast, amenorrhea, infertility.

Prader scale



- **Primarily designed to rate virilization in CAH.** Some have adopted it to describe the range of differentiation of genitalia, with normal infant presentation being shown on either end of the scale.

Evaluation

- Number of **perineal openings** (vagina, anus)



- **Rectal exam** (palpable uterus)

- **Gonad palpation** (labioscrotal/inguinal)



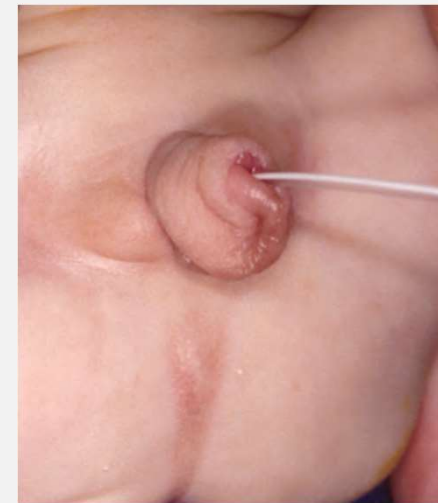
- Palpable gonad rules out 46XX DSD
- Gonad asymmetry indicates MGD & ovotesticular DSD

- **Specific Measurements**



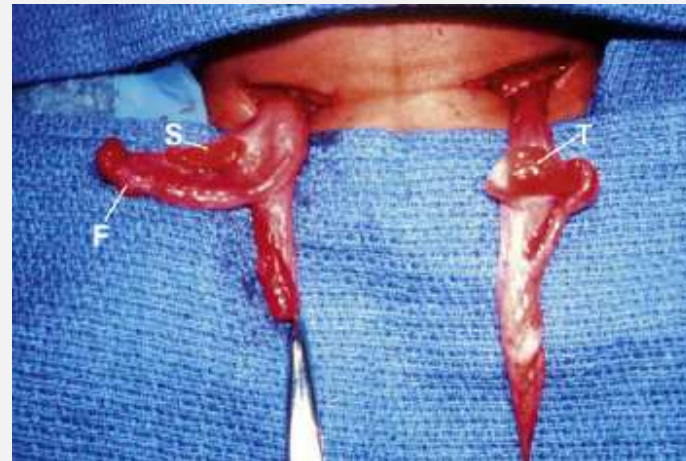
- Stretched phallus length (clitoromegaly/micropenis)
 - Anogenital ratio (labial fusion)
- **Bronzing** of the areola or scrotum (CAH)
 - Evaluate **family history** (genital ambiguity, fetal death)

Presence of Anus (Cloaca Vs DSD)



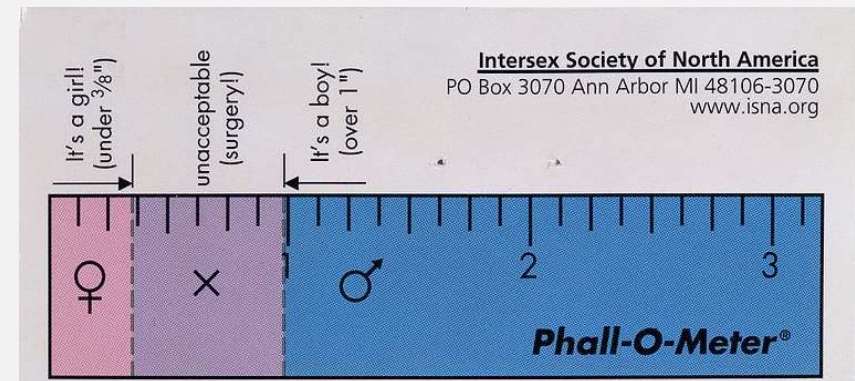
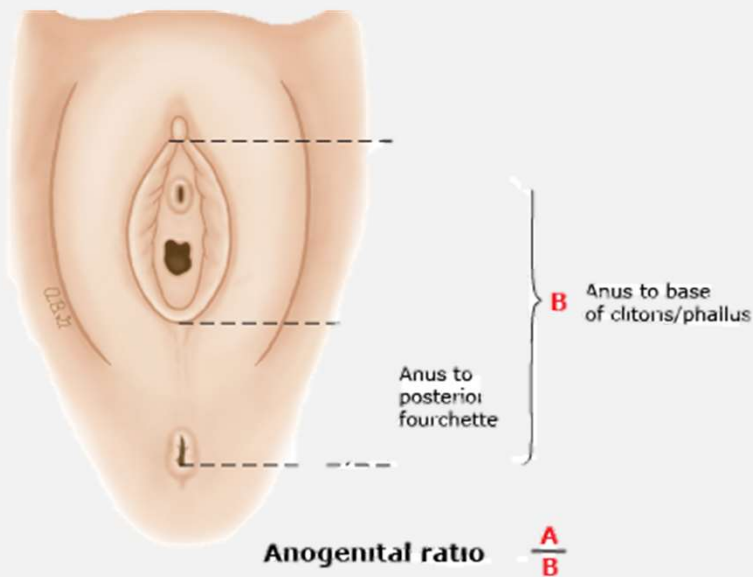
- Cloaca can have bizarre external genitalia such as **blank-appearing (doll-like) perineum**, complete **genital transposition** or **enlarged phallic structure (psuedophallus)** that gives the genitalia an ambiguous appearance. Cloacal opening can be at introitus or at tip of phallus

Gonad asymmetry



- **Asymmetry** may indicate **mixed gonadal dysgenesis** (typically with rt descended testes and lt streak gonad with Mullerian remnants) or **ovotesticular DSD** (ovary one side, testis on the other with correlating ipsilateral ductal structures). *ovotestis may be present on one or both sides (usually Mullerian correlate)

Measurements



- **Stretched phallus length:** Clitoromegally >9 mm or Micropenis < 2.5 cm in full term infant.
- **Anogenital ratio:** >0.5 (independent of gestational age and size) indicates posterior labial fusion

Workup

- **Chromosomal evaluation:** **Karyotype**
- **Genetic evaluation:** micro array, gene testing, whole genome sequencing
- **Laboratory:** gonadal Hormones, **adrenal steroids, electrolyte**
- **Imaging:** **Pelvic ultrasound** (presence of uterus and gonad)
- **Gonadal Biopsy** (if findings would influence gender of rearing)
- **Endoscopy** (in urogenital sinus)

Treatment

- Prevent the **salt-wasting crisis** with CAH
- **MDT** (pediatric surgeon, endocrinologist, geneticist, neonatologist, psychologist)

Family communication

Psychological emergency (parents)

- **Congratulate** family on the new addition.
- If the baby is healthy, **reassure** the family
- **Avoid premature designation of gender**, naming of the baby, and completion of the birth certificate; counsel that it will take days or weeks to conduct the initial evaluation
- emphasis should be on **helping the parents understand** that the atypical genital appearance, although uncommon, is biologically understandable.

Psychosexual support

- Discuss the concept of an **evolving gender identity**
- Psychosexual gender must be **monitored by skilled professionals** during childhood.
 - **Multistage counseling**: birth, two years, school entry, around puberty, annually during adolescence
 - **Detailed and honest**. Pubertal development, menses and fertility, sexual function
- give access to **advocacy groups**
- controversy on the correct timing to **share information with the affected child**.

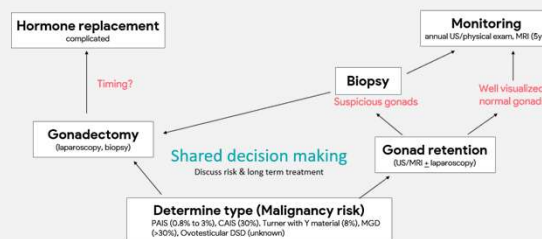
Gender Assignment

- Decision doesn't only depend on type of DSD
- For some DSD identity is reasonably predictable
 - 95% of CAH identify as female & have reproductive potential
 - **Mixed gonadal dysgenesis** is advised to be male
 - Most CAIS identify as female
 - Most 46-XX-RD2 deficiency identify as male
- Genital surgery does not "cure" a DSD.
 - **Good function**: preservation of sexual function and any reproductive potential
 - **Normal appearance**: stable gender identity, psychosocial well-being
 - **Least number of operations**
- Decisions should be made on a case-by-case basis considering unique character of each child/family

Assisted fertility

- Assisted reproductive technology is successful in a few patients
- Identification of germ cells suggests a higher fertility potential
- Delaying gonadectomy allows autonomous decisions but may decrease fertility

Gonadectomy vs Gonad preservation



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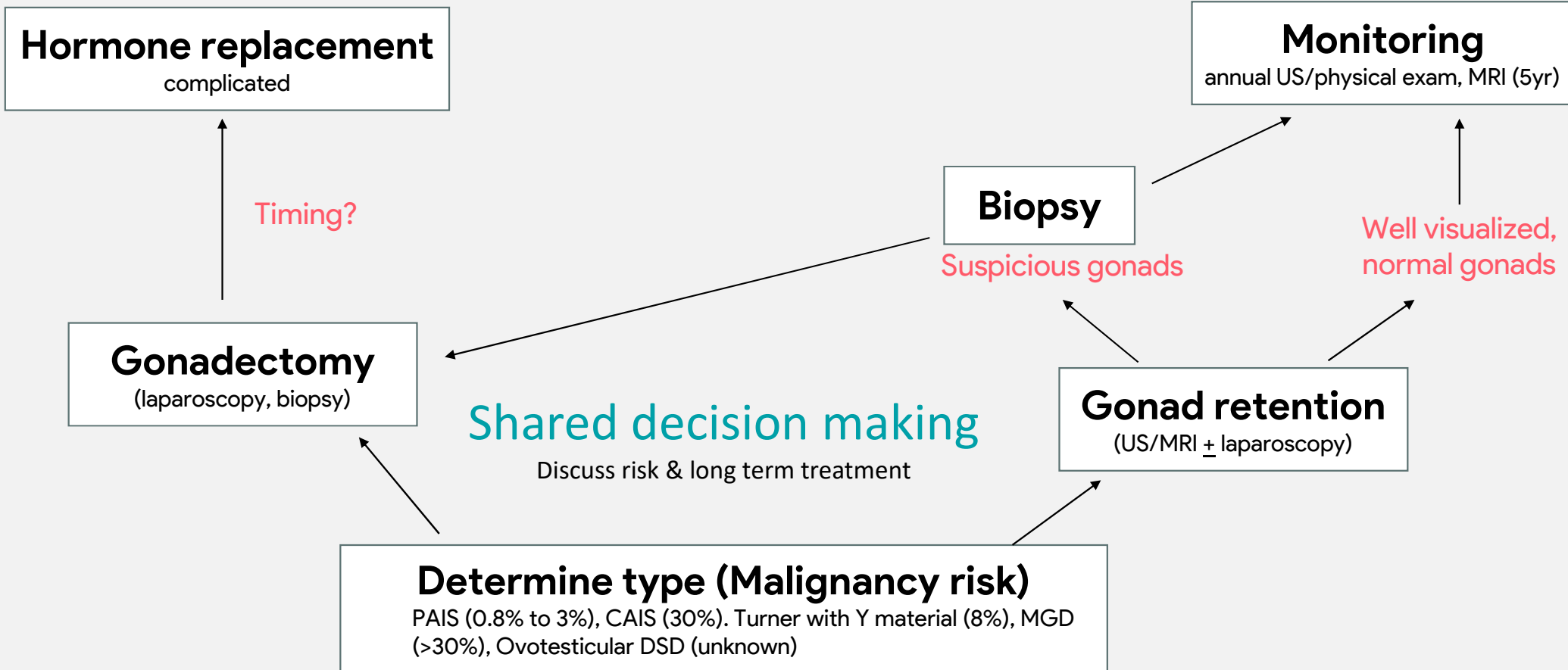
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Assisted fertility

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Classification

Consensus Statement on Management of Intersex Disorders

Peter A. Lee, MD, PhD^{a,b}, Christopher P. Houk, MD^c, S. Faisal Ahmed, FRCPC^d, Ieuan A. Hughes, FMedSci, FRCPC^{b,e}, In collaboration with the participants in the International Consensus Conference on Intersex organized by the Lawson Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology

46XX DSD

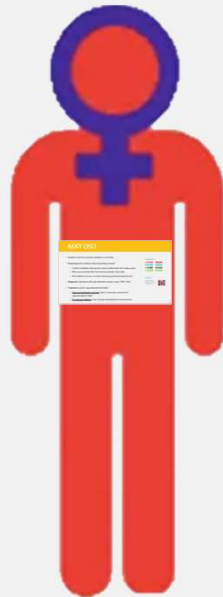
(excess androgen)



46XY DSD

(Androgen Deficit)

*MIS deficiency



Sex chromosome DSD (45X/46XY)

MGD & ovotesticular DSD

Other?



46XX DSD

- **Cause:** 95% CAH, remaining due to maternal androgen excess
- **Phenotype:** Normal Mullerian structure (No palpable gonad)
 - Variable virilization, high/low urogenital sinus
- **Diagnosis:** 17-OHP (elevated except type 4), other (11-DOC), electrolyte
- **Treatment**
 - Supportive management of fluid and electrolyte abn
 - hydrocortisone (glucocorticoid)
 - fluorhydrocortisone (mineralocorticoid)
 - Feminizing genitoplasty (?male for prader V with late diagnosis)

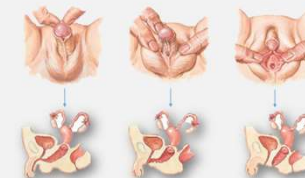
Androgen Excess

Type	Deficiency	Level of block	presentation
1	21-hydroxylase	Cortisol	Virilization only
2	21-hydroxylase (classic: 90%)	+ mineralocorticoid	+ dehydration & hyperkalemia
3	11 β hydroxylase	+ Mineralocorticoid below level of deoxycorticosterone	+ fluid overload, hypertension, inc Na, hypokalemia
4	3 β hydroxylase	+ cholesterol to testosterone	severe salt wasting, lethal, can occur in males

• group of inherited autosomal recessive genetic abnormalities



Persistent urogenital sinus

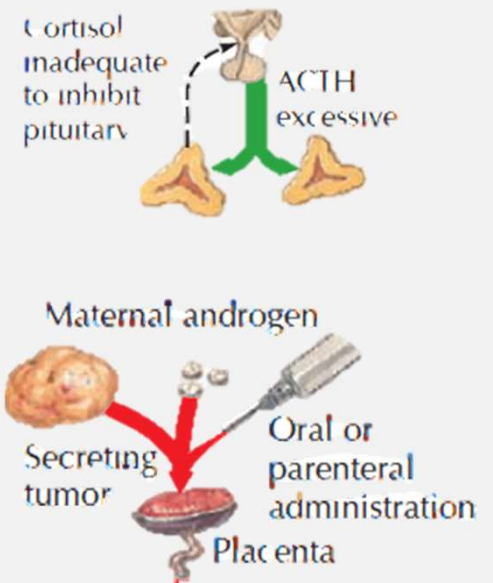


• Urogenital sinus is commonly associated with genital ambiguity (CAH). Urogenital sinus opening may occur anywhere from a near-normal introital location to the tip of a well-formed glans. Different confluences have been described but most important factor is length of urethra and common channel.

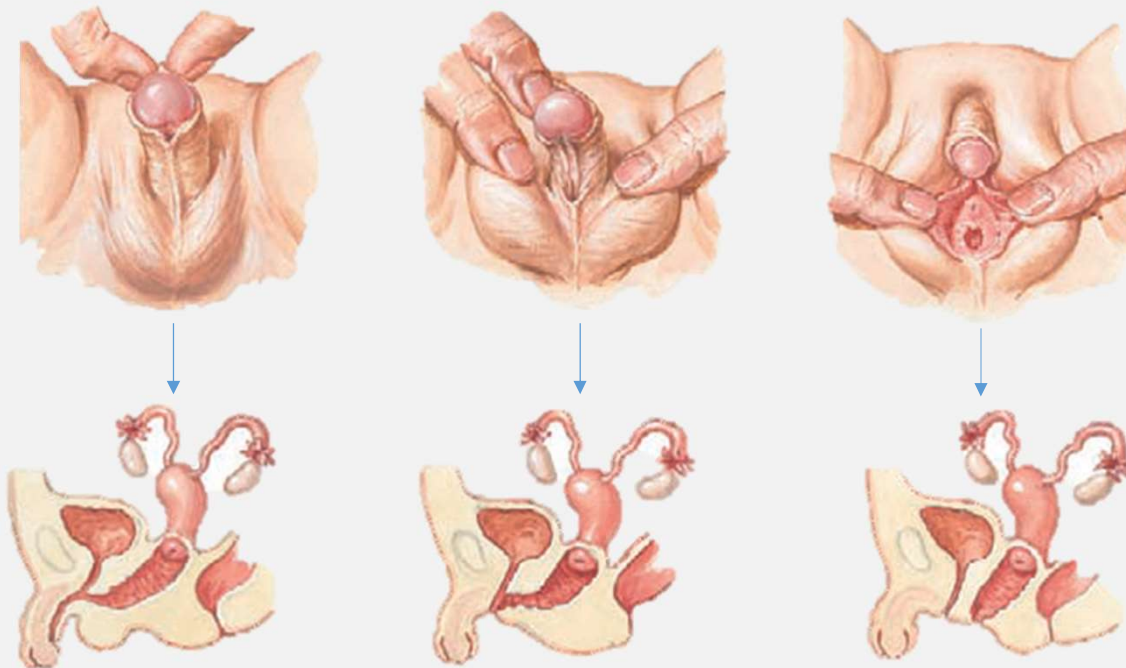
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Congenital Adrenal Hyperplasia



Persistent urogenital sinus



- **Urogenital sinus** is commonly associated with genital ambiguity (CAH). **Urogenital sinus opening** may occur anywhere from a near-normal introital location to the tip of a well-formed glans. Different confluences have been described but most important factor is **length of urethra and common channel**.

46XY DSD

- **Cause:** problem in production, reception or conversion
- **Phenotype:** Have testicular tissue (sometimes palpable)
 - Variable feminization (hypospadias, female genitalia with blind-ending vagina)
 - MIS is usually secreted (No female internal genitalia, amenorrhic)
 - Mild virilization may occur at puberty (excess gonadotropin/testosterone)
- **Diagnosis:** testosterone, LH, hgG stimulation, receptor assay, T:DHT (>10:1)
- **Treatment:** gender assignment is individualized
 - Androgen insensitivity syndrome: based on response to testosterone (must be female if CAIS)
 - 5a reductase deficiency: have androgen imprinting (male recommended)

Androgen Deficit



*MIS deficiency

- Both Wolffian and Mullerian structures
- Orchiopexy difficult b/c vas adherent to mullerian
- Excision of mullerian risk damage to vas

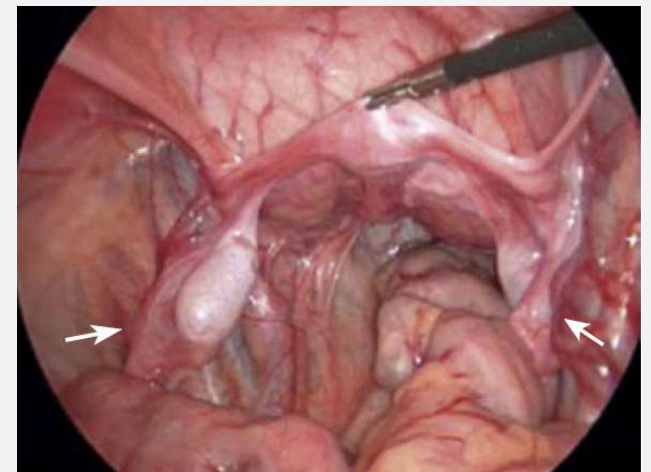


Androgen Deficit

01	Testosterone production (Adrenal)	<ul style="list-style-type: none">• Severe CAH
02	Testosterone production (testicular)	<ul style="list-style-type: none">• Gonadal dysgenesis
03	Testosterone reception	<ul style="list-style-type: none">• Androgen insensitivity syndrome
04	Conversion to DHT	<ul style="list-style-type: none">• 5a-reductase deficiency

*MIS deficiency

- **Both Wolffian and Mullerian structures**
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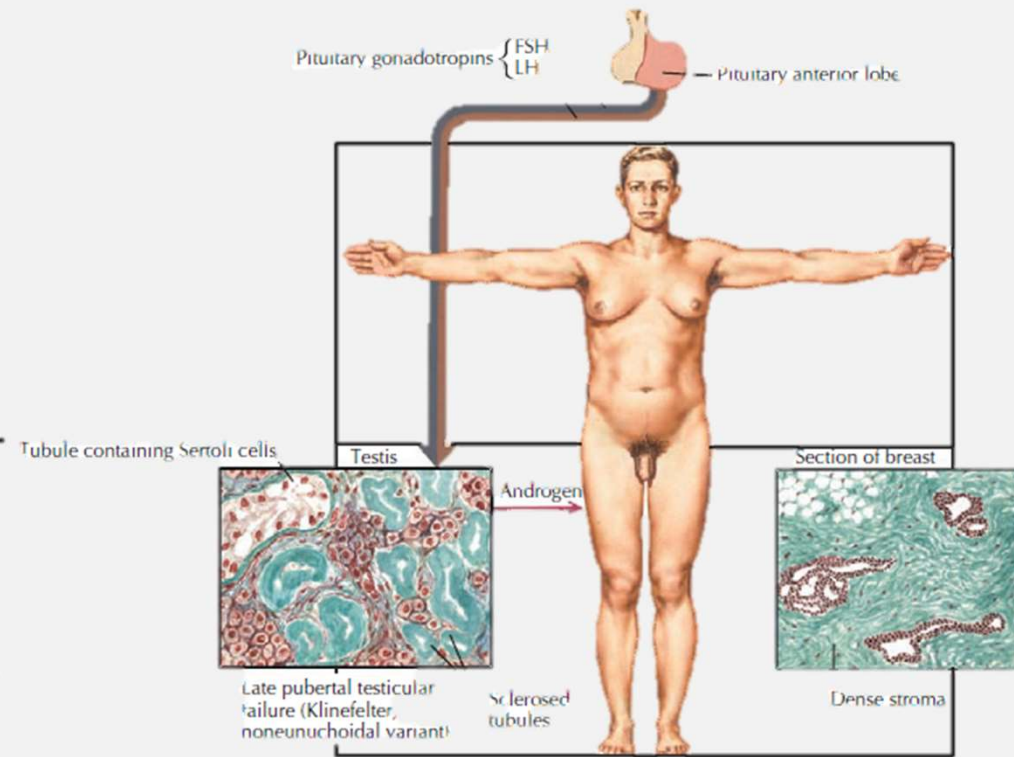
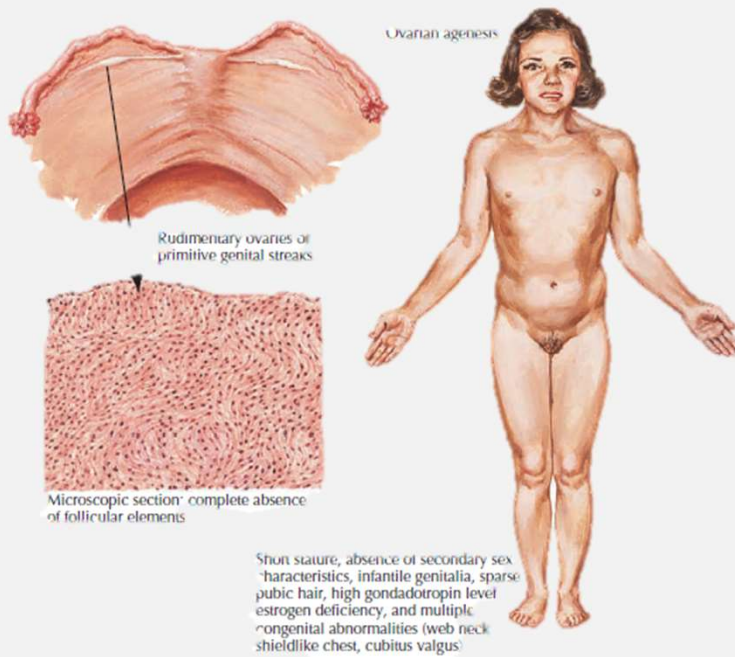
Mixed Gonadal Dysgenesis

- **Cause:** 45X/46XY mosaicism
- **Phenotype:** testis on one side and streak gonad (ovary) on other
 - stigmata of Turner syndrome are variably present
 - Ambiguous but masculinized. Testes usually undescended
- **Diagnosis:** turner features, chromosome and finding of streak gonad
- **Treatment:** historically raised as female b/c short & early gonadectomy
 - Advise male rearing b/c Y chromosome & virilized (hypospadias repair)
 - Risk of gonadoblastoma in both gonads (20%) so remove streak gonad with hemiuterus and surveillance of scrotal testes with biopsy at puberty
 - Risk of wilms tumor & glomerulopathy (DDS)

Ovotesticular DSD

- **Cause:** 46XX (translocation of SRY), 45X/ 46XY (mosaicism)
- **Phenotype:** Both male and female gonads
 - Testes and ovotestes are usually palpable. Ovaries are not
 - Ambiguity with a tendency towards masculinity
- **Diagnosis:** mosaic karyotype or ductal structures
- **Treatment:** decision very complex, diagnosis requires gonadal biopsy
 - Males require hormonal replacement b/c progressive testicular fibrosis,
 - Males need surveillance (1-10% testicular tumor)
 - Females don't need hormones and rarely described to be fertile
 - Discordant gonad should be removed early (retained testes cause virilization)

45X (Turner), 47XXY (Klinefelter)



Surgical Reconstruction

Male gender confirmation

- Orchidopexy / orchidectomy
- Removal of persistent Mullerian structures (if symptomatic)
- **Hypospadias repair or Phalloplasty**



Local flap



- Pedicle flaps: scrotal dartos- musclocutaneous flap

Free flap



- Anterolateral thigh (ATL), thoracodorsal artery perforator (TDAP), Radial forearm (RFFF)

Transplant



- Vascularized composite allotransplantation (VAC)

Outcome

Complex Reconstruction (Flap)

- Good aesthetics and sensation but poor intercourse and ability to void standing
- No inherent erectile function; inflatable penile prosthesis for erection
- Implant complication
- Urethral stricture and fistula

Transplantation

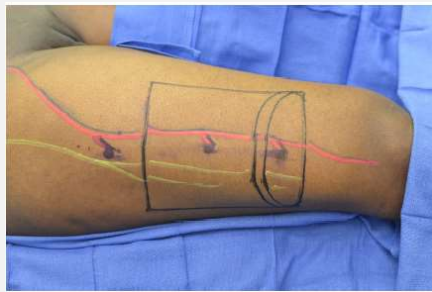
- Good aesthetics, sensation as well as intercourse and voiding
- reliability and safety
- medication, toxicity

Local flap



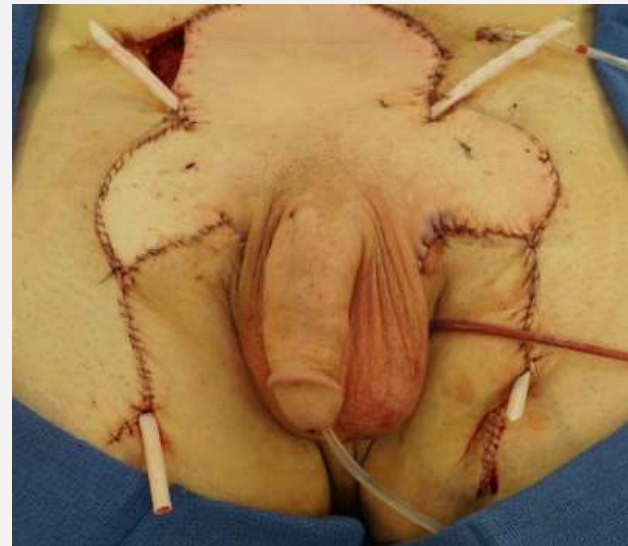
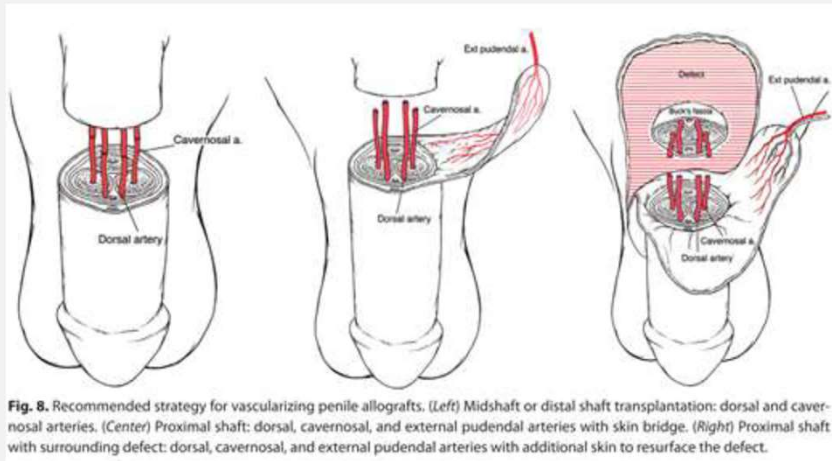
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Free flap



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Outcome

Complex Reconstruction (Flap)

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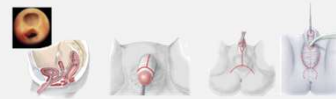
Transplantation

- Good aesthetics, sensation as well as intercourse and voiding
- reliability and safety
- medication, toxicity

Female gender confirmation

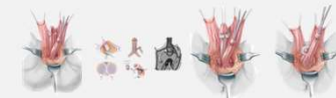
- **Orchiectomy: Timing?**
 - Before puberty: risk of malignancy. Risk of virilization/masculinization
 - after puberty: testis synthesizes estradiol (facilitate feminine development)
 - ****Avoiding orchiectomy** (surveillance only) to avoid hormonal side effect
- **Feminizing genitoplasty**: used to be two stage until recently. Timing?
 - Early : easier repair b/c vagina thicker & lower (maternal estrogen)
 - Can use prepuce for vaginoplasty if max virialized (dec stenosis)
 - Peripubertal : for very small and very high vagina b/c high rate of stenosis (better compliance with dilatation at older age)

Incision



• 10% endoscopes used to inspect penile shaft. Telescope withdrawn from sheath and fatty capsule are incised into vagina and bladder. Skin flaps are raised. Gonadotropin-releasing hormone (GnRH) and progesterone are administered. Incision extended vertically on dorsal & ventral aspect. Vertical incision extended proximally to well-invested & shaped penile flap, similar to scrotal orchiectomy.

Clitoroplasty



• Phallus is degloved and two flaps are pulled aside. Backs flaps incised on ventral aspect of each corpus. Tunic along median line of corpora and lateral ligament. Lower clitoral corpus is removed and preserved. It then is sutured to the penile shaft. The remaining clitoral corpus is sutured to the penile shaft. The remaining clitoral corpus is sutured to the penile shaft. The remaining clitoral corpus is sutured to the penile shaft.

Vaginoplasty



Labioplasty



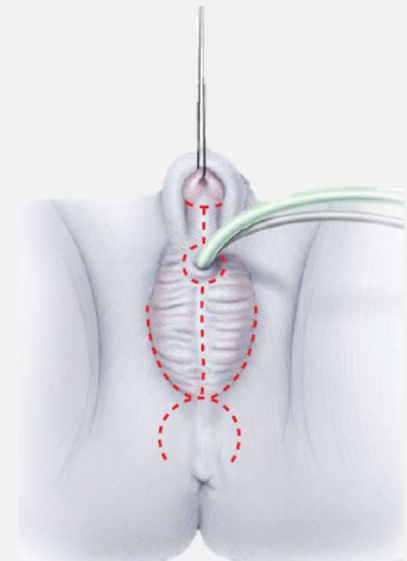
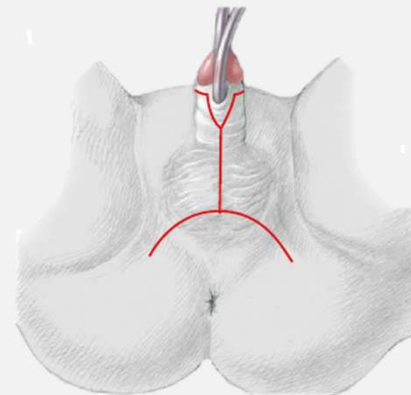
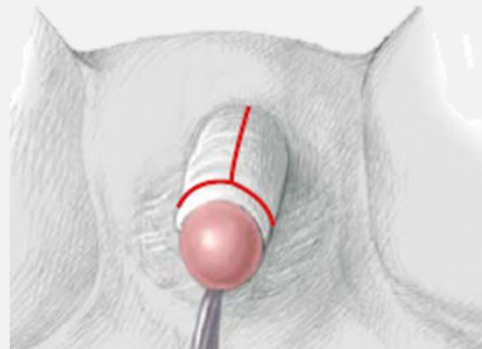
• Prepuce skin is brought about with G-shaped advancement flap, leaving some skin for prepuce hood. It is then sutured to the penile shaft. The remaining prepuce skin is sutured to the penile shaft. The remaining prepuce skin is sutured to the penile shaft. The remaining prepuce skin is sutured to the penile shaft.

Outcome

- Normal or nearly normal-appearing and functioning genitalia
- Successful coitus is often reported (7 sexual satisfaction, lubrication, and erotic sensitivity, nonpainful coitus)
- **Vaginal stenosis** - require revision procedure after puberty in (25% to 100%)
- **Stress incontinence** may be an issue with TUM (descent of the bladder neck)

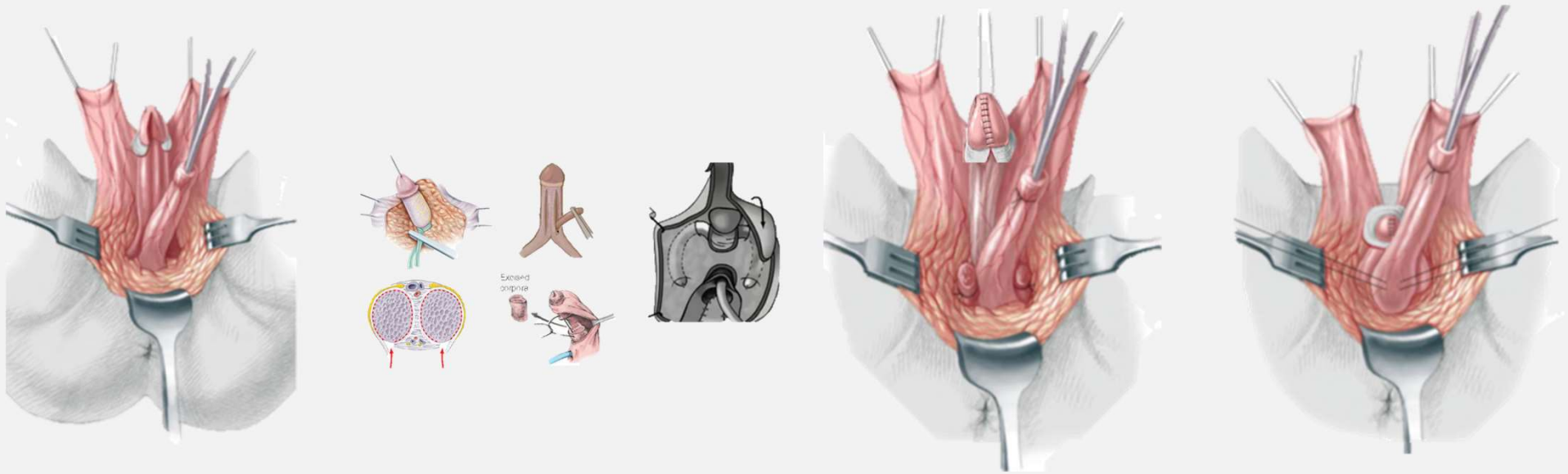


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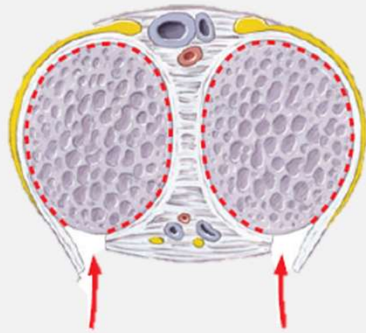
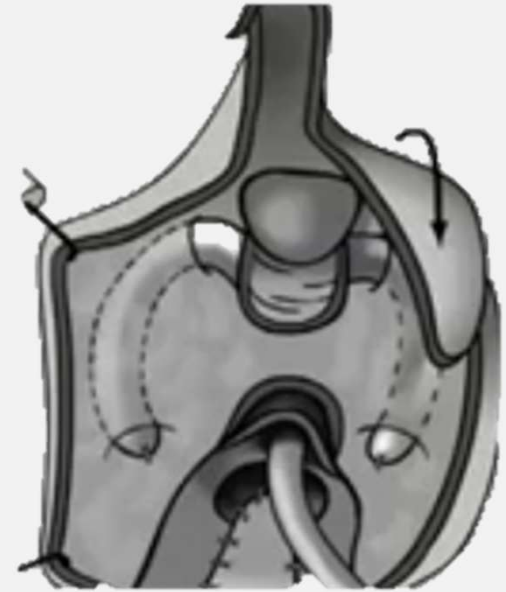
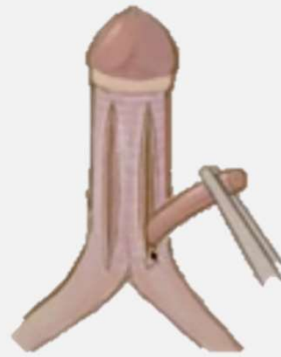
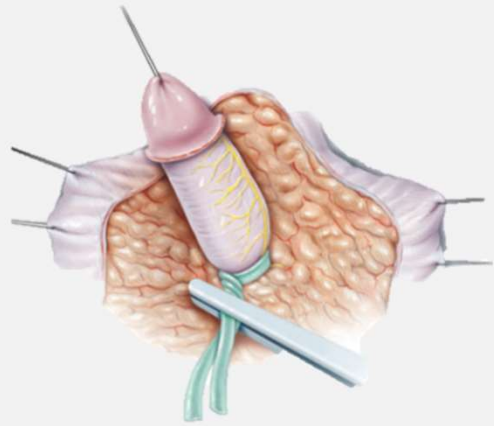


- **10Fr cystoscope** used to inspect urogenital sinus. Telescope withdrawn from the sheath and **foley catheters** are inserted into **vagina and bladder**. Both balloons are inflated . **Circumcision incision** around enlarged clitoris and urogenital orifice leaving 5mm cuff. Incision **extended vertically on dorsal & ventral** aspect. Ventral incision extended proximally to wide **inverted U shaped perineal flap** (extend to ischial tuberosities) – nowadays narrower

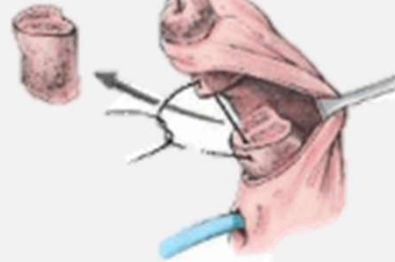
Clitoroplasty



- **Phallus is degloved** and two flaps are pulled aside. **Buck's fascia incised** on ventral aspect of each corpora. Tunica albuginea dissected off corpora and **is suture ligated** (below glans and at pubis symphysis) and removed. If glans is bulky, reduction should be made centrally leaving the lateral sensitive part. **Glans is repaired** with 6-0 suture then sutured to corporal stumps **under pubic arch** with two or three 4-0 sutures.
- **** An alternative technique preserving the corporeal bodies has been described (maintain potential for reversibility)**



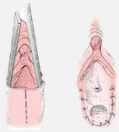
Excised
corpora



Vaginoplasty

Very short sinus

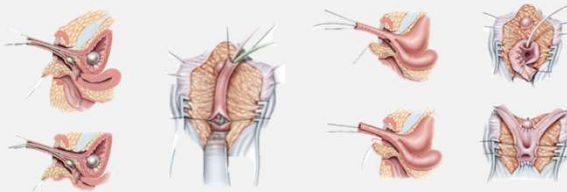
Vaginal cutback



• Rarely used, most believe it is only appropriate for simple labial fusion. This technique involves inclusion of the fused skin posteriorly to the perineum to expose the vaginal orifice. The incised lateral edges are oversewn. While such a procedure is simple and may produce a good result in the agropenial patient, it is only a consideration in patients with a common urogenital sinus on the phallus with confluence of a normal vaginal opening at the level of the perineum. This combination is extraordinarily rare; thus, this procedure is very rarely, if ever, indicated.

longer sinus, normal urethra length

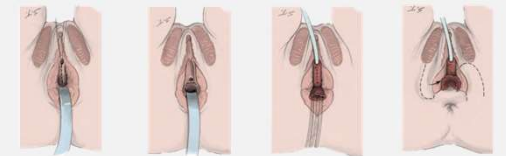
Urogenital mobilization: Partial/ Total



• High UGS: urogenital sinus is brought tension-free as close as possible to the perineum (after separating from corporal bodies). The sinus is incised to expose communication with urethra. Classically, vaginal wall is opened posteriorly into normal caliber vagina and sutured to perineal flap skin. Redundant urogenital sinus split in the dorsal midline for mucosal plate. More recently, we have described opening the sinus laterally allowing the sinus to rotate to create a posterior wall of the vagina, eliminating the need for the posterior skin flap. This latter maneuver requires the mobilized sinus to be long and well vascularized.

longer sinus, short urethra

Urogenital separation: Vaginal Pullthrough



• very high sinus requires separating vagina from the urogenital sinus, and the sinus is used to create a urethra. Mobilized vagina may reach perineum but most require skin flaps.

Flap Vaginoplasty



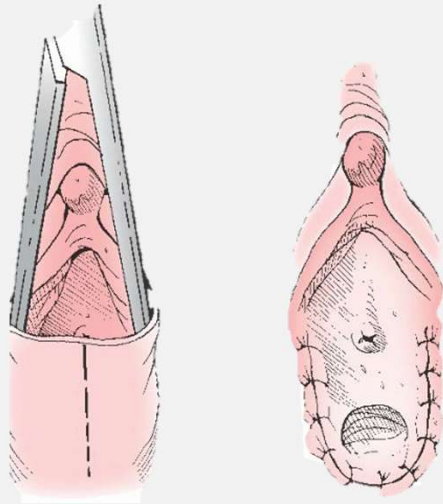
• Low urogenital sinus: flap is mobilized prior to incision of the sinus. Note that the confluence is not changed but the sinus is opened up by insertion of the flap. Released posteriorly to split level of confluence. Further wall is opened and normal caliber. Then UGS is advanced to complete anterior vaginal wall. If severe female hypospadias will result from flap vaginoplasty we would recommend a pull-through procedure. Skin elements may be excised caudally at the base of the flap to create a more normal appearing posterior introitus. Subcutaneous tissue and blood supply should be preserved at the base. Maintaining a U rather than narrow V shape at the tip anteriorly helps to make sure that the end of the flap is not ischemic.

Urogenital separation: Vaginal Replacement



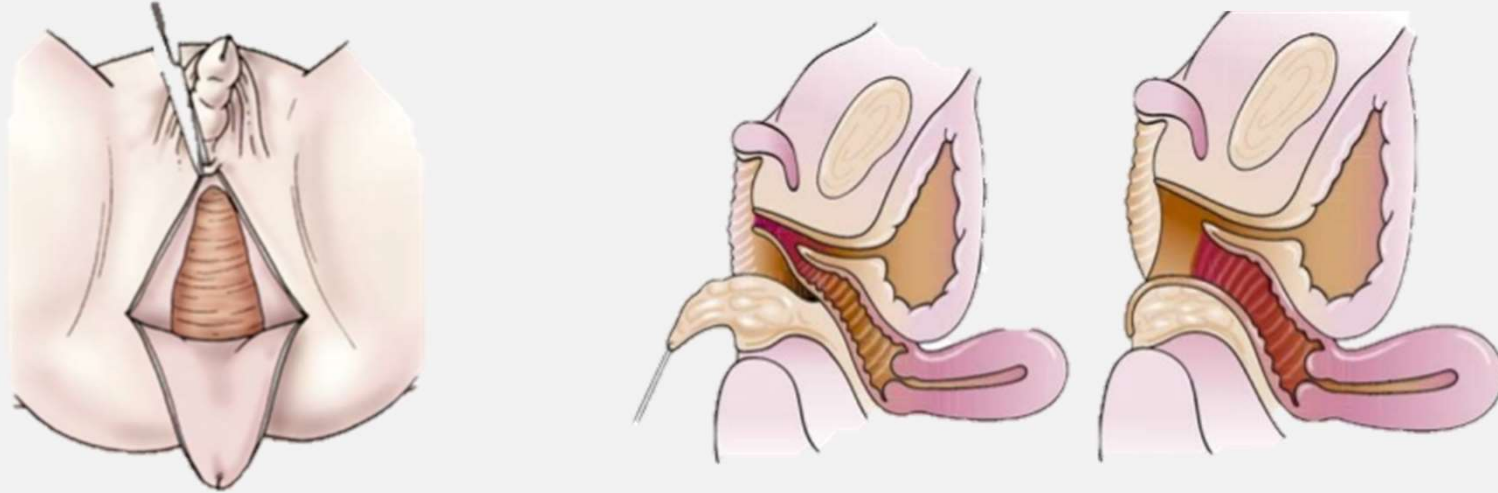
• Vaginal substitution with colon, ileum or split thickness skin graft may be needed in absent/rudimentary vagina and peripubertal obese patient. Microde skin vagina. A split-thickness skin graft is modeled over a stent.

Vaginal cutback



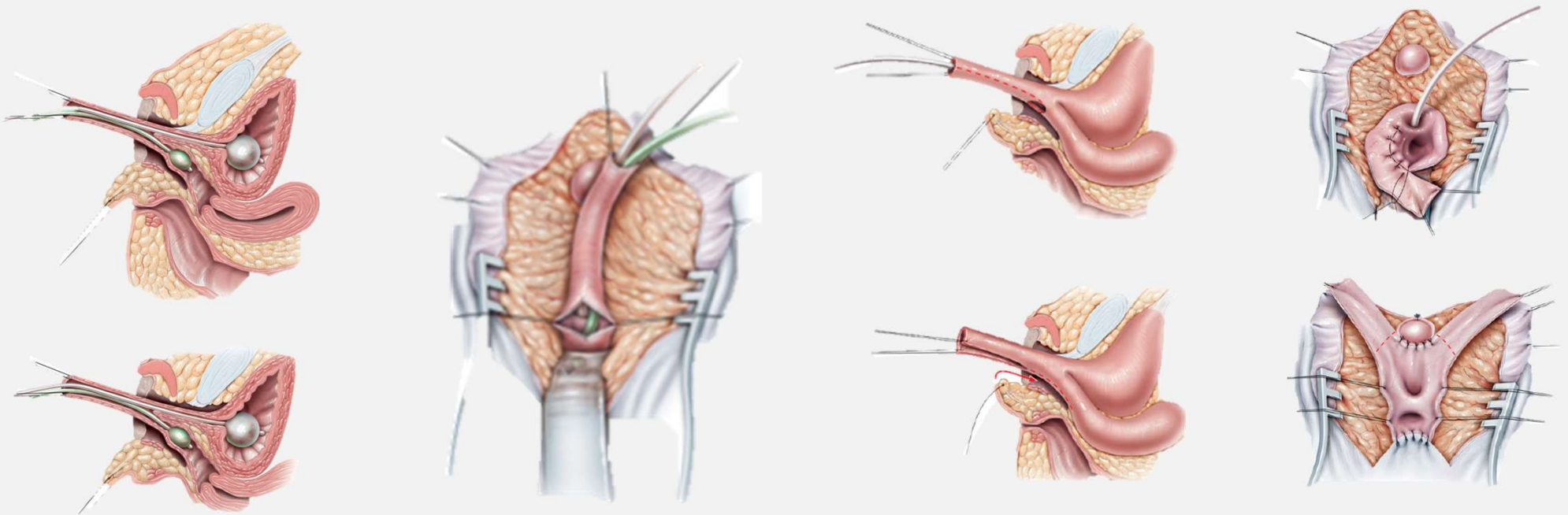
- Rarely used, most believe it is only appropriate for simple labial fusion. This technique involves incision of the fused skin posteriorly to the perineum to expose the vaginal orifice. The incised lateral edges are oversewn. While such a procedure is simple and may produce a good result in the appropriate patient, it is only a consideration in patients with a common urogenital sinus on the phallus with confluence of a normal vaginal opening at the level of the perineum. That combination is extraordinarily rare; thus, the procedure is very rarely, if ever, indicated.

Flap Vaginoplasty



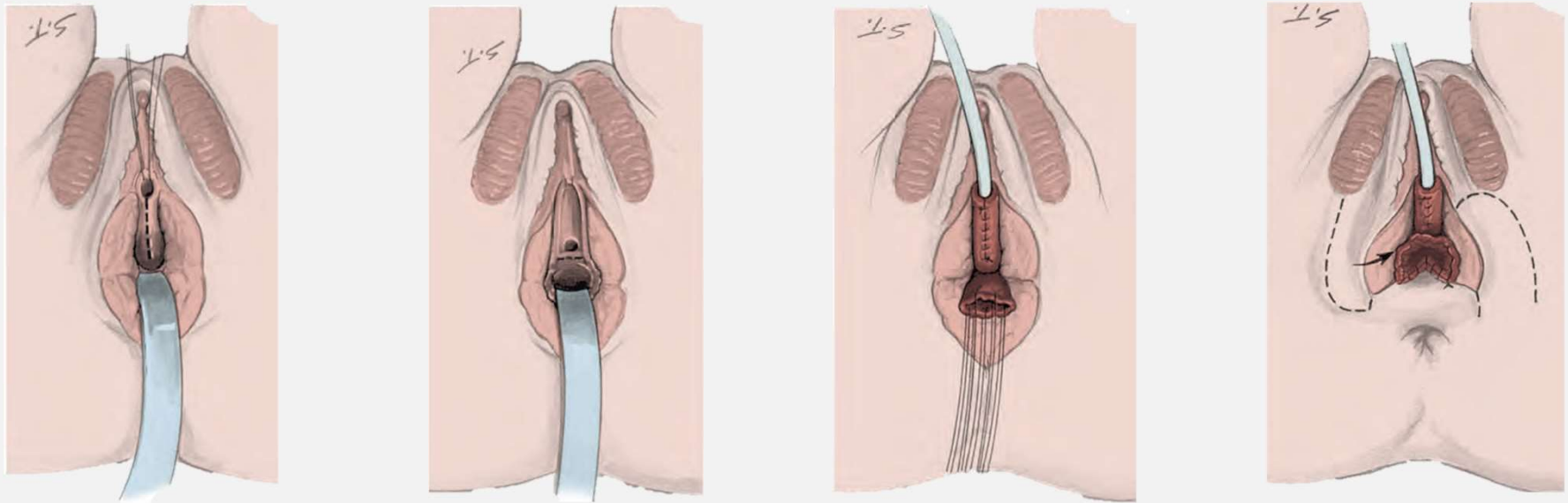
- **Low urogenital sinus: flap is mobilized prior to incision of the sinus. Note that the confluence is not changed but the sinus is opened up by insertion of the flap.** dissected posteriorly until level of confluence. Posterior wall is opened until normal caliber. Then U flap is advanced to complete posterior vaginal wall. If severe female hypospadias will result from 'flap' vaginoplasty we would recommend a 'pull-through' procedure. Skin elements may be excised carefully at the base of the flap to create a more normal-appearing posterior introitus. Subcutaneous tissue and blood supply should be preserved at the base. Maintaining a U rather than narrow V shape at the tip anteriorly helps to make sure that the end of the flap is not ischemic

Urogenital mobilization: Partial/ Total



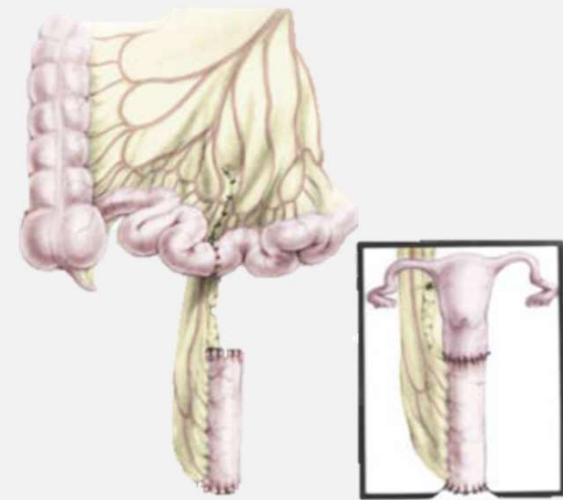
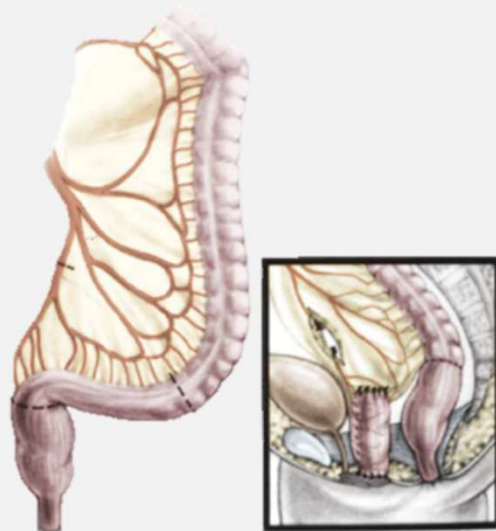
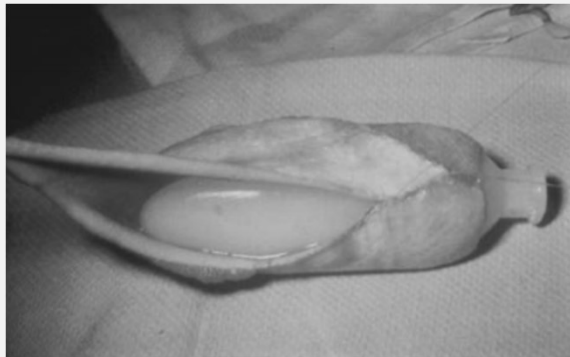
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Urogenital separation: Vaginal Pullthrough



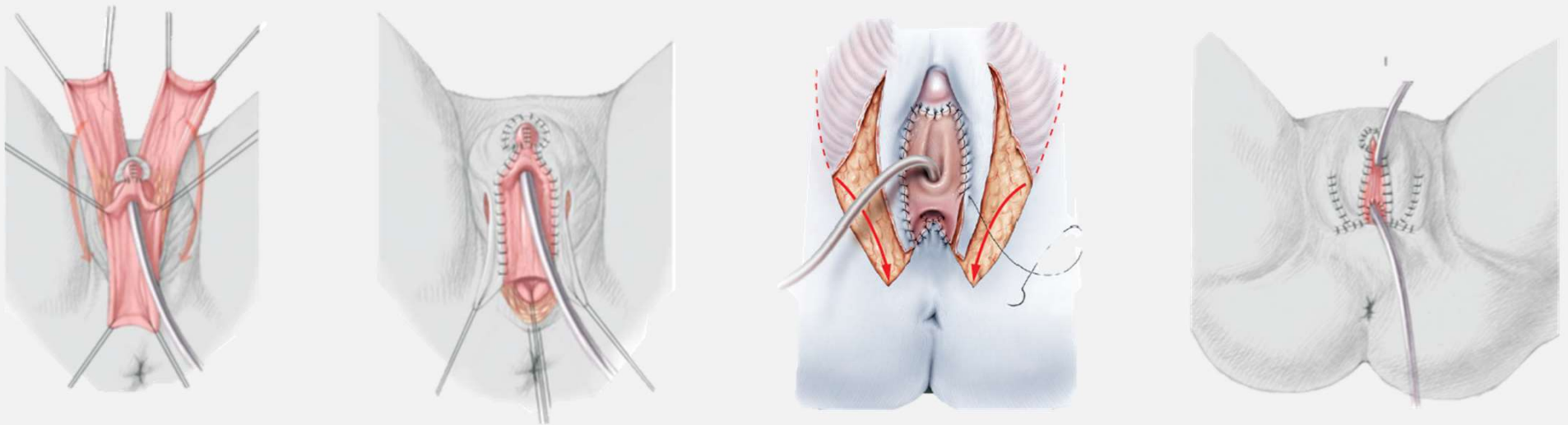
- very high sinus requires **separating vagina from the urogenital sinus**, and the sinus is used to create a urethra. **Mobilized vagina may reach perineum** but most require **skin flaps**.

Urogenital separation: Vaginal Replacement



- Vaginal **substitution with colon, ileum or split thickness skin graft** may be needed in absent/rudimentary vagina and peripubertal obese patient. McIndoe skin vagina. A split-thickness skin graft is modeled over a stent.

Labioplasty

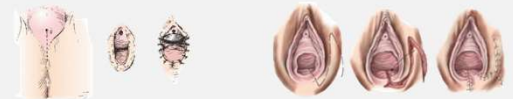


- Preputial skin is brought down with V-shaped advancement flap, leaving some skin for preputial hood. **It is** then sutured to mucosal plate to create labia minora. **Vaginal** wall is sutured to mucosal plate (ventral and latera) and to tip of inverted u flap (posteriorly). Labia majora is moved inferiorly by Y-V plasties and sutured to lateral side of preputial skin to complete the labioplasty.
- Elastic wrap legs (with foam b/n knees and ankles) for 1-2d. compression dressing and vaginal tampon/drain removed after 48–72 h. catheter is removed after 7 days.

Outcome

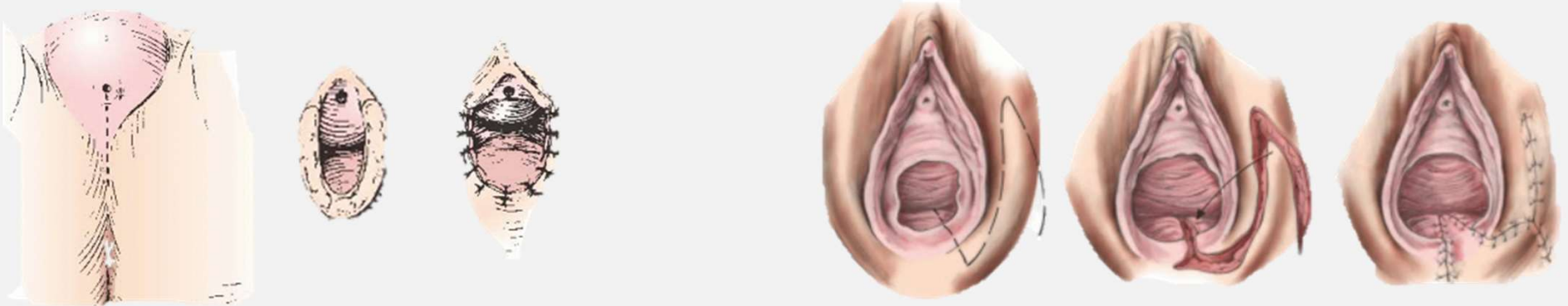
- Normal or nearly normal-appearing and functioning genitalia
- **Successful coitus** is often reported (? sexual satisfaction, lubrication, and erotic sensitivity, nonpainful coitus)
- **Vaginal stenosis** – require revision procedure after puberty in (25% to 100%)
- **Stress incontinence** may be an issue with TUM (descent of the bladder neck)

Surgery for vaginal stenosis



* Narrowed vaginal outlet may require a minor revision (midline incision and closing it at 90 degrees to the incision) e. In some instances it may be necessary to create labiocutaneous flaps to enlarge orifice.

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Summary

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- Infant with **genitalia** that is **atypical or discordant with chromosomal sex** is classified as DSD
- **CAH** is the most common and **MGD** is second most common form of neonatal ambiguous genitalia
- First steps to determine the cause of the DSD are a **karyotype and a pelvic ultrasound**
- For common types of DSDs, **sex of rearing and genital surgery** are reasonably **predictable**
- **long-term support** to address psychosexual/social adjustment, decision-making, effects of sex steroid exposure, bone density and potential of malignancy in retained gonadal tissue.

Thank You

References

- Holcomb & Ashcraft's pediatric surgery, 7th E, 2020
- [Uptodate](#) updated 2019
- Campbell Walsh urology, 11th E, 2016
- Netter's pediatrics, 1st E, 2011
- [Consensus statement on DSD](#), 2006

