Surgical Treatment for Ambiguous Genitalia

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Genetic information passed to the fetus plays a critical role in gonad development. If this information is skewed by maternal adrenal problems or medications ingested by the mother during fetal development, abnormal sexual development may occur in the fetus. Abnormalities in fetal gonad development can lead to a condition known as ambiguous genitalia (more commonly referred to as "hermaphroditism").

Males have an XY chromosome configuration and females an XX configuration. In normal development, existence of the Y chromosome stimulates secretion of the protein H-Y antigen, which causes testes to develop rather than ovaries while external genital tissue simultaneously develops as a scrotum rather than labia.[1] By week 8 in fetal development, Leydig's cells differentiate and begin to secrete testosterone, which affects the embryonic ducts. (The embryonic wolffian ducts, which transport urine from the early kidney, ultimately become the sperm ducts, which include the epididymis, vas deferens, and seminal vesicle, while the mullerian ducts become the fallopian tubes, uterus, and upper vagina.) The presence of testosterone triggers deterioration of the mullerian ducts, causing the labial tissue to fuse and form a scrotum. The obvious external differences between male and female begin to develop at week 9 when the scrotum and penile shaft (versus the vaginal and urethral openings) become apparent. The transformation is complete by week 14 (Figure 1).

If genital differentiation fails to occur at the fetal stage, the baby will be born a hermaphrodite. Approximately 65% of
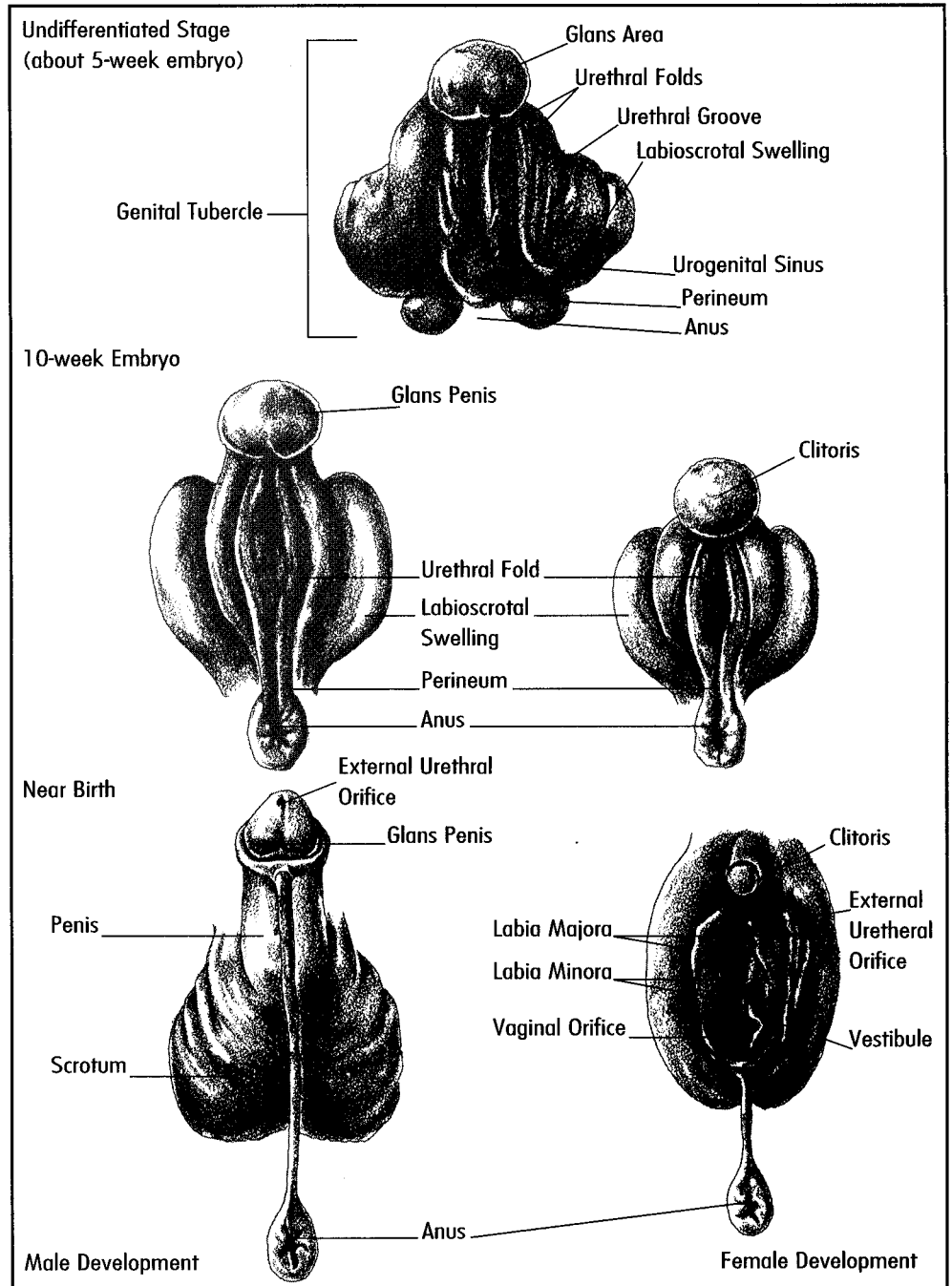

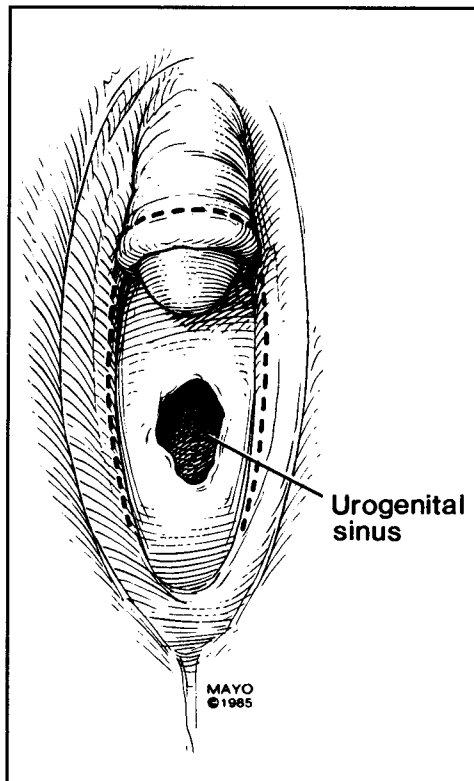**Figure 1.** Development of the external genitals.

Figure 2. *Coronal incision extended bilaterally for shaft resection.*

Figure 3. *For shaft resection, suspensory ligament and corpora cavernosa are exposed.*

hermaphrodites have an ovotestis—a gonad with both testicular and ovarian elements. (They usually have an ovary as well.) When palpated, the ovarian section of the ovotestis feels firm while the testicular section feels soft. The ovotestis is usually found in the inguinal canal or the labioscrotal fold. Generally, 75% of hermaphrodites have enough phallic tissue to develop as a male. Most also have chordee (downward curvature of the erect penis) or hypospadias (urethral meatus on the underside of the penile shaft). At puberty, breast development occurs in as many as 88% of hermaphrodites and nearly 50% experience menstruation in the form of hematuria (blood in the urine).[2,3]

Variations of hermaphroditism are termed "male pseudohermaphroditism" and "female pseudohermaphroditism." The male pseudohermaphrodite has normal to low testosterone levels, normal testes, female ducts, and/or female external genitalia. The testes usually
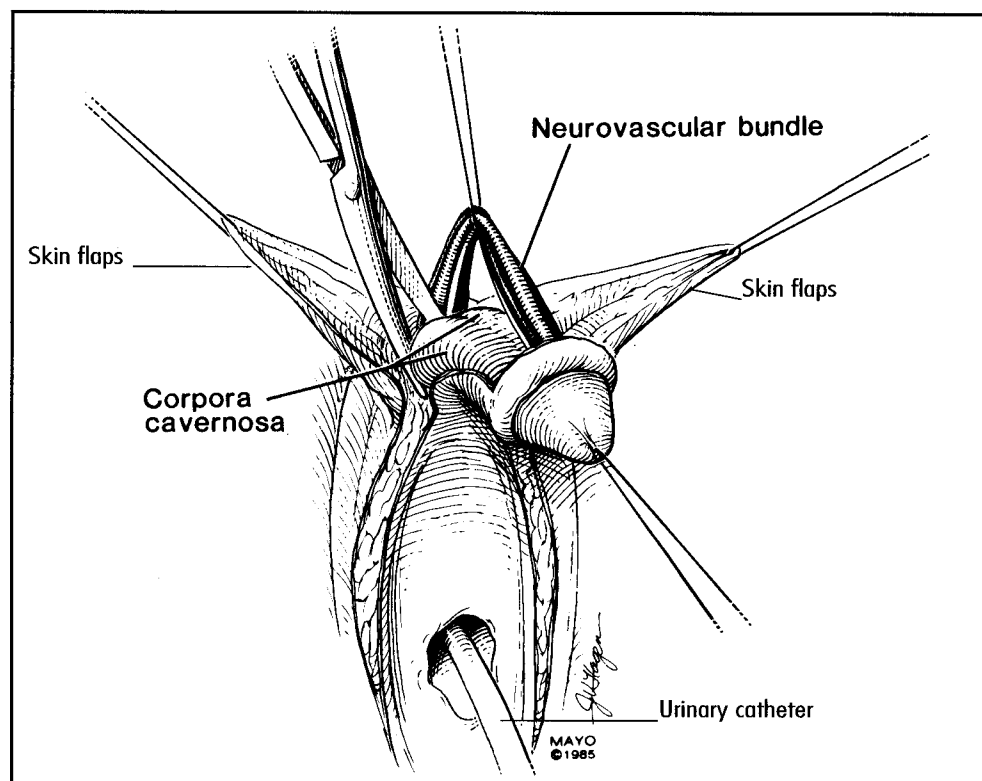
descend by puberty, and voice change may occur, along with scrotal pigmenta-

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tion, increased muscle mass, and phallic growth. However, less facial hair is present, and the temporal hairline recedes less than in a normal male. Additionally, pubic and axillary hair may not develop. The labia remain immature in appearance, and the vagina ends in a blind pouch.

Female pseudohermaphrodites often exhibit normal ovaries as well as ambiguous male external genitalia. Excessive adrenal enzyme secretion, which results from a tumor in the adrenal gland or on an ovary in utero, leads to development of the male characteristics exhibited in

female pseudohermaphroditism. Abnormal adrenal secretion of glucocorticoids (hormones that protect against stress and affect protein and carbohydrate metabolism) or mineralocorticoids (which affect the regulation of fluid and electrolytes) may also cause renal dysfunction and hypertension. Treating a pregnant woman with stilbestrol to prevent miscarriage has also been shown to cause female pseudohermaphroditism in the fetus.

Diagnosis
Because normal physical and psychological development requires certainty about sexual identity, diagnosis and surgical treatment of ambiguous genitalia should begin as soon as a problem is suspected. Diagnosis commences with an exploration of family history: Questioning should divulge information on infertility or genital anomalies, unexplained neonatal deaths, and adrenal abnormalities. Palpation of the infant's gonads for normal consistency—firmness for an
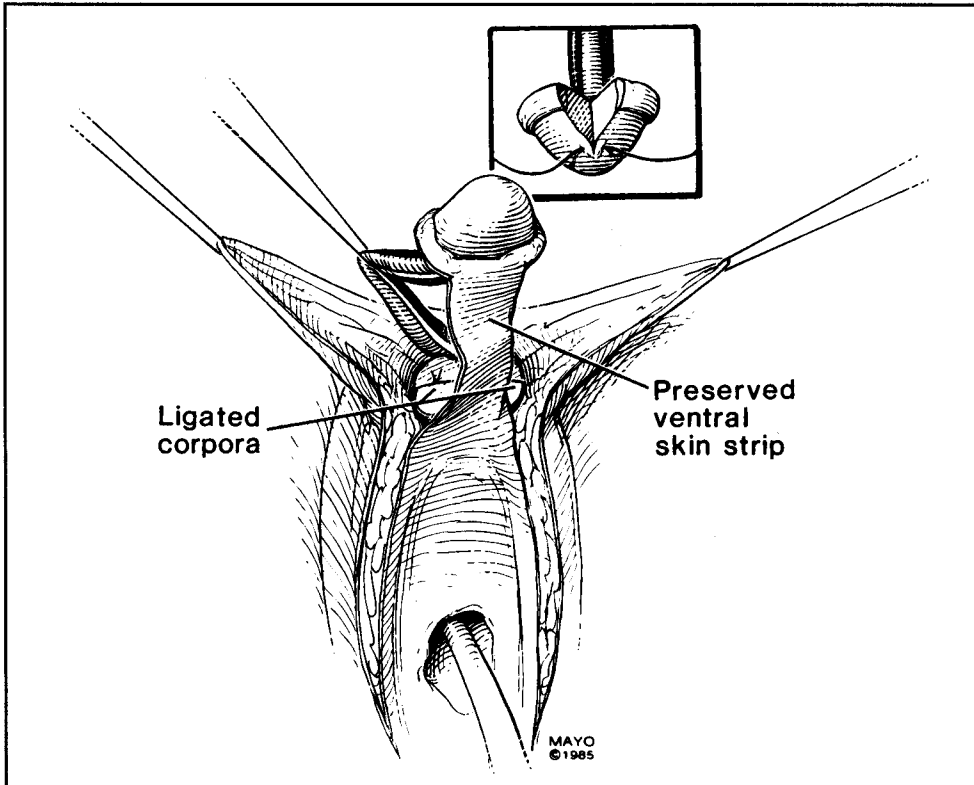
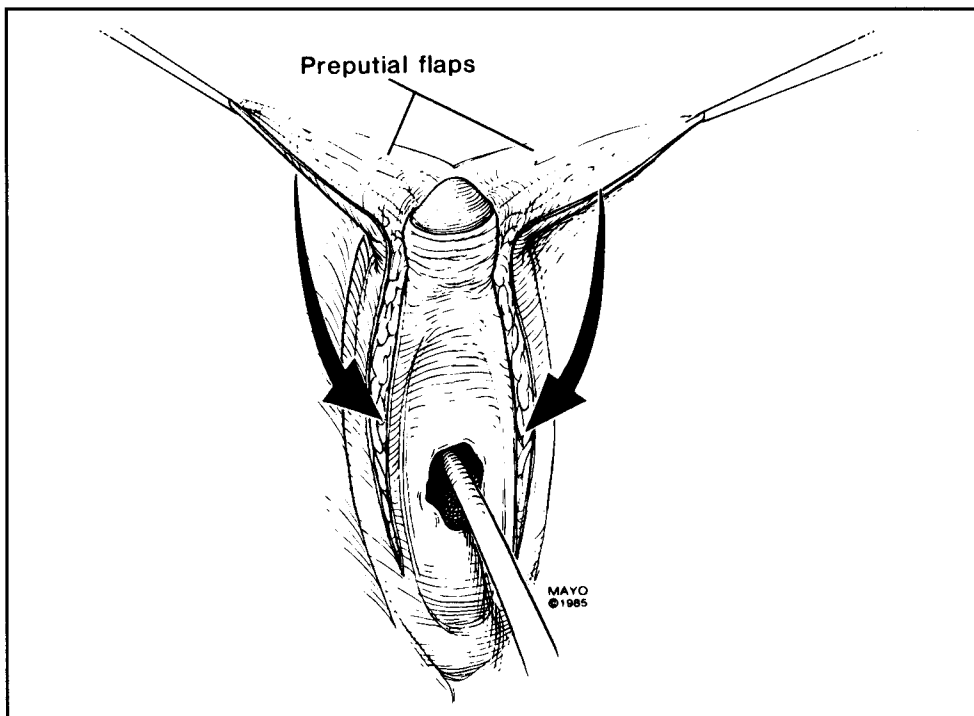**Figure 4.** Removing a wedge of glans tissue to reduce its size.

Figure 5. Skin flaps are brought down and sutured in place to create labia.
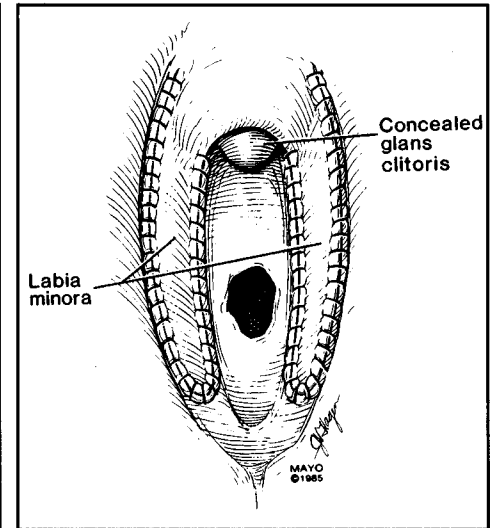

Figure 6. Postoperative clitoral shaft resection.

ovary or softness for a testicle—is also crucial to initial diagnosis.

Investigation should also include performing a karyotype and abdominal ultrasound, and testing for adrenal abnormalities by checking for serum levels of 17-hydroxyprogesterone and 17-hydroxypregnenolone. Karyotypes reveal the genetic makeup of the cell—XX for female or XY for male. They may be

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performed on bone marrow aspirate or preferably, on cultured lymphocytes. The abdominal ultrasound will demonstrate the presence of a uterus and/or testes in the newborn's inguinal canal. If physical examination, karyotyping, ultrasound, and serum-level testing fail to provide a definitive diagnosis, the remaining diagnostic approaches include laparoscopy (exploring the abdomen with an endoscope) or exploratory laparotomy to examine structures and biopsy the ovotestis.

Surgical Conversion

Once testing has yielded conclusive results, the surgical conversion process may begin. Surgical repair should be completed before sexual awareness develops, which typically occurs at 3 years of age. Beyond this age, a sex change would be contraindicated psychologically. However, the child's awareness of the reason for surgery is generally not a concern because the surgery is often performed on newborns to alleviate parental anxieties.

When determining surgical approach, preserving fertility is considered less important than addressing cosmetic and social factors. In most cases of ambiguous genitalia, surgical conversion to female genitalia offers the best cosmetic and social results. Clitoroplasty and vaginoplasty (each performed as a separate procedure) also prove less complicated than creating male genitalia.[4] (Hermaphrodites born with a uterus and ovaries are generally able to bear children.)

Preoperatively, endoscopic examination and x-ray films confirm the configuration of genitourinary structures. Cystoscopy (examination of the bladder) and vaginoscopy reveal the existence of the urethra and vagina as well as their relative position.

Clitoroplasty

Three major procedures fall under the heading of clitoroplasty: clitorectomy, clitoral recession, and clitoral shaft resection. Clitoral shaft resection is the procedure of choice in more than 90% of ambiguous-genitalia cases because it results in better cosmetic and sexual function than either clitorectomy or clitoral recession.[6] Clitoral recession is performed in only 10% of the cases, and clitorectomies are rarely performed. All three procedures are described below; however, clitoral shaft resection is described in greater detail because it results in the most successful outcome.

Described in 1966 by R.E. Gross, MD, clitorectomies involve modifying the phallic stump, which may be accom-

plished as early as 3 weeks of age without significant complications. While clitorectomies generally yield good cosmetic results, they often result in diminished sexual function and sensation because the dorsal neurovascular bundle and the ventral mucosal plate of the corpus spongiosum cannot be preserved. Additionally, clitoromegaly—leaving the phallic stump unresected—may lead to painful erections at puberty.[5]

A technique described in 1970 by J.G. Randolph, MD, and W. Hung, MD, involves clitoral recession, which preserves the erectile tissue and the nerve block. During this procedure, the erectile

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tissue is buried beneath the mons pubis and secured with sutures in the corporal fascia. However, the glans is left whole so it may serve as the sexual organ. This procedure is preferred when the phallic shaft is short (less than 1 cm).

In 1981, K.I. Glassberg, MD, and G. Laungani, MD, described clitoral shaft resection, which begins by placing the patient in the lithotomy (dorsosacral) position and inserting a urinary drainage catheter. If the child's diminutive size renders conventional stirrups ineffective for positioning, "frog-legging" (putting the legs in a lithotomy-like position) will allow access to the perineal area. Safety precautions must, however, be taken when securing the legs to prevent nerve damage, which can result from improper body positioning. Patients with adrenogenital syndrome will require steroid administration during surgery, which should be continued postoperatively while tapering the dosage over a 3-day period.

Once the patient is prepped, a circumferential incision is made that extends bilaterally and vertically on the coronal margin of the shaft (Figure 2 on p. 11). (The ventral skin is left intact to provide

blood circulation to the glans.) Dissection continues until the suspensory ligament and the corpora cavernosa have been exposed so they can be ligated and divided (Figure 3 on p. 11). Once the corpora cavernosa are removed, the glans is sutured against the pubic bone with nonabsorbable sutures just below the origin of the dorsal suspensor ligament. If the glans is too large, a small wedge of tissue may be removed from the dorsal aspect to achieve a better appearance (Figure 4). Should the urethra need to be shortened, some of the ventral skin may be resected, leaving the dorsal skin to supplement circulation. The preputial skin that has been dissected off—but not resected from the phallus—is used to create the labia minora. The skin is split in the midline, and the flaps are brought to either side of the ventral skin strip and sutured with absorbable suture (Figure 5).

Following surgery, the glans should be completely enveloped by the labia. If not, a reduction glansplasty is performed. Complications include tissue sloughing of the glans or in rare cases, glans atrophy. However, the newly created clitoris typically appears normal and responds normally to sensation (Figure 6). Figures 7 and 8, on p. 14, are examples of preoperative and postoperative cases of clitoral shaft resection.

In addition to basic pediatric soft-tissue instruments, special instruments for clitoroplasty include: skin hooks, Jones scissors, curved Halsted clamps, curved Iris scissors, Adson tissue forceps with teeth, Adson tissue forceps without teeth, narrow Allis clamps, and Stille scissors. Sutures include: 3-0, 4-0, 5-0, and 7-0 absorbable; 3-0 silk ties; and 2-0, 3-0, and 4-0 nonabsorbable.

Vaginoplasty

Following clitoral modification, vaginoplasty is performed to complete the transformation to female genitalia. To prevent vaginal orifice stricture, surgeons prefer to perform vaginoplasty when the patient is close to puberty. However, in some cases, the procedure is performed earlier in life. The three major types of
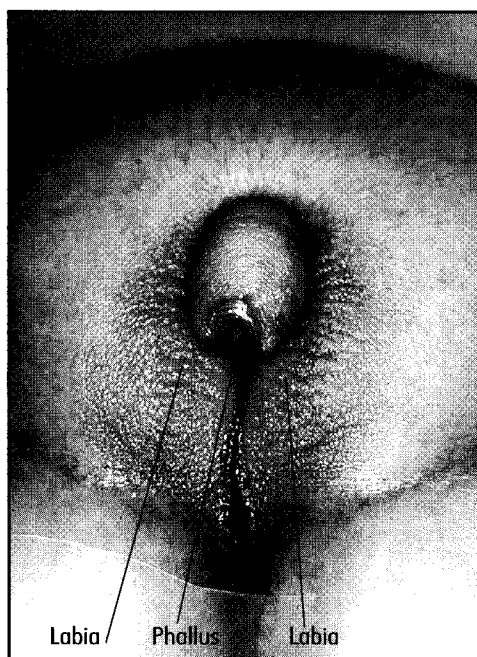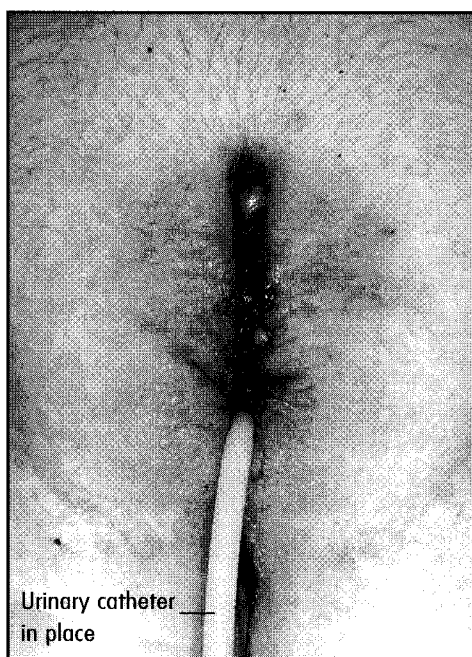
Figure 7. Preoperative clitoral shaft resection.

Labia / Phallus / Labia

Figure 8. Postoperative clitoral shaft resection.

Urinary catheter in place

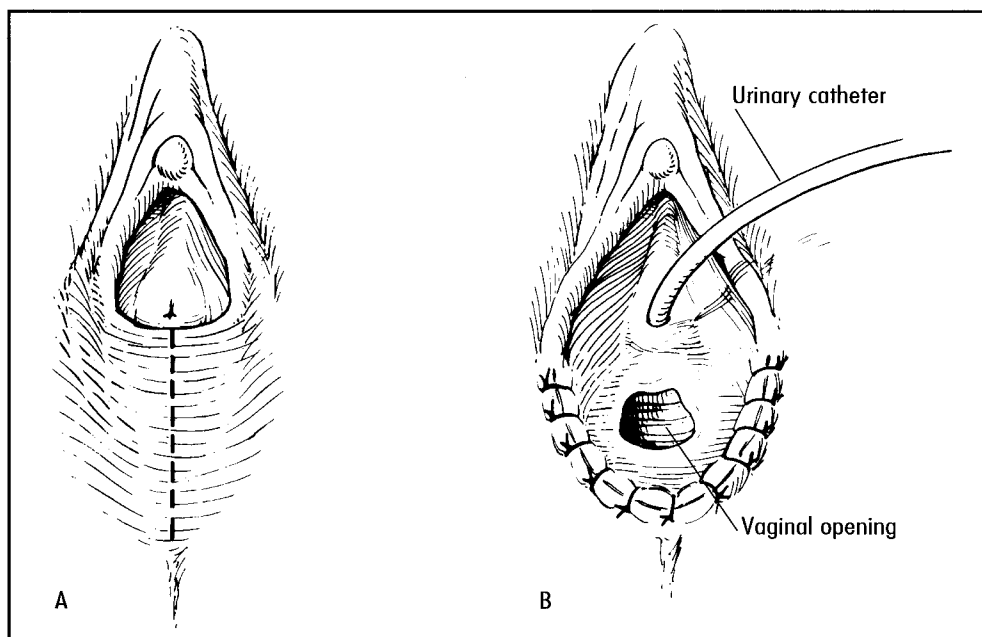Urinary catheter

Vaginal opening

A

B

Figure 9. A—Incision of skin over vaginal opening (cut-back vaginoplasty). B—Vaginal opening exposed and excess skin removed.

vaginoplasty include: cut-back vagino-plasty, flap vaginoplasty, and pull-through vaginoplasty. The location of the vaginal orifice relative to the urethra will dictate the extent and type of reconstruction necessary; therefore, before scheduling surgery, the exact nature of the vaginal defect must be determined through radiography and cystoscopy.

Cut-back vaginoplasty is the least complex procedure and may be performed when the child exhibits minimal virility in the genital structures. Incision of a thin layer of skin over the vaginal opening creates an adequate entrance (Figure 9).

Flap vaginoplasty is performed when the vaginal orifice and the urethral

meatus are buried deep in the perineum. An inverted V or U incision is made in the perineum in order to create two thick vascular flaps. The lower portion of the flaps becomes the inferior aspect of the labia minora while the superior portion is sutured to the vaginal orifice. The perineal incision is extended toward the ischial tuberosities, taking care not to injure the rectum. The urogenital sinus is then incised on the posterior wall until the urethra and vagina are visible. Finally, the apex of the perineal flap is sutured into the vaginal opening 1 cm to 2 cm in length using absorbable sutures (Figure 10).

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Postoperatively, the urinary catheter is removed 24 to 48 hours following surgery, and a vaginal pack is left in place for 48 hours. Two to three weeks postoperatively, careful and extensive dilatation of the new vaginal opening is begun in order to prevent skin contracture and vaginal stenosis. Dilatation is performed daily for up to 6 months using a properly sized Hegar dilator.

Pull-through vaginoplasty—the most complex vaginoplasty procedure—is indicated when the vaginal opening converges with the urethra in the area of the verumontanum or in cases of vaginal atresia (the absence of a vagina). In both cases, a vagina must be created in order for genitalia to appear normal. Though somewhat similar to flap vaginoplasty, pull-through vaginoplasty requires more extensive perineal dissection. Additionally, full-thickness flaps must be sutured in place around plastic stents to create the shape of the neovagina.

Pull-through vaginoplasty carries the risk of urinary stress incontinence if the
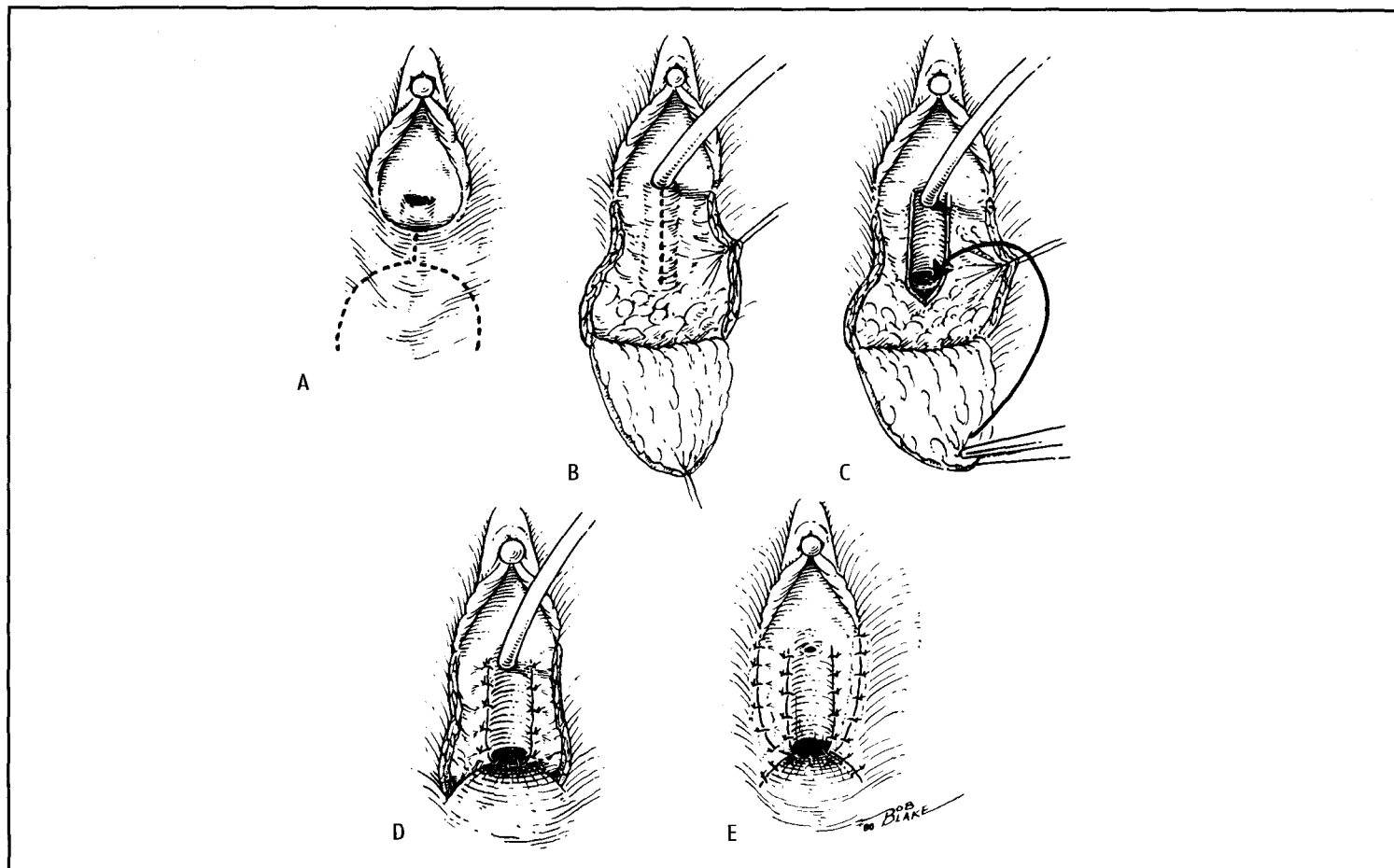
Figure 10. *Flap vaginoplasty. A——Incision. B——Flap retracted. C——Urethral and vaginal openings exposed. D——Apex of flap is sutured into vaginal opening. E——Lower portion of flap is sutured in place to create labia.*

external urethral sphincter is transected. To avoid injury to the bladder neck, it is advisable to delay surgery until the child reaches at least 2 years of age. Urinary tract infections caused by vaginal drainage are rare even though the vaginal opening is proximal to the urethral meatus.

Postoperatively, both dilatation of the vaginal opening and a treatment regimen consisting of female hormone therapy are necessary. In addition to the instruments used during clitoroplasty, the following instruments are required during vaginoplasty: nasal speculum, bi-valved vaginal speculum, Jewett sounds, ruler, and a silver probe. Suture selection depends on the child's size. Δ

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References
1. Jirasek JE. *Development of the Genital System and Male Pseudohermaphroditism.* Baltimore, Md: The Johns Hopkins Press; 1971.
2. van Niekerk WA (ed). *True Hermaphroditism: Clinical Morphologic and Cytogenic Aspects.* Hagerstown, Md: Harper & Row; 1974.
3. van Niekerk WA. *True Hermaphroditism: The Intersex Child.* 1981;8:80.
4. Kramer SA, Weinerth JL. Treatment of Ambiguous Genitalia. In: *Pediatric Plastic Surgery.* St Louis, Mo: CV Mosby Company; 1984;3:917.
5. Gross RE, Randolph J, Crigler JF Jr. Clitorectomy for Sexual Abnormalities. In: *Surgery.* 59:300-308.
6. Barrett TM, Gonzales ET Jr. Reconstruction of the female external genitalia. *Urol Clin N Am.* 1980;7:455.

Additional Reference
Tortora GJ. *Principles of Anatomy and Physiology.* 8th ed. New York, NY: Harper Collins College Publishers; 1996.