

Invited Review

Genitoplasty for the management of 46,XX disorders of sex development and congenital adrenal hyperplasia

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ABSTRACT

46,XX disorders of sex development (DSD) resulting from congenital adrenal hyperplasia (CAH) lead to varying degrees of genital virilization and raise complex questions regarding gender assignment, timing of intervention, and long-term outcomes. Feminizing genitoplasty aims to restore functional anatomy, preserve urinary and sexual function, and support psychosocial well-being within a multidisciplinary and ethically informed framework. While many individuals with CAH retrospectively favor childhood surgery, population data show heterogeneous views, supporting a case-by-case approach rather than a universal moratorium. Ethical and legal standards emphasize transparent discussion of risks, alternatives, and long-term outcomes; consent must be qualified, persistent, written, and reaffirmed over time. Early surgery may be beneficial in selected cases—particularly where functional obstruction or significant virilization is present—yet deferral remains appropriate when uncertainty prevails. Contemporary management integrates nerve-sparing techniques, urogenital sinus reconstruction, and long-term psychosocial support, underscoring the need for individualized, evidence-based care for children with CAH-related DSD.

Keywords: 46,XX disorders of sex development, Congenital adrenal hyperplasia, Feminizing genitoplasty

INTRODUCTION

46,XX disorders of sex development (DSD) secondary to congenital adrenal hyperplasia (CAH) result from 21-hydroxylase deficiency, causing prenatal androgen excess and virilization of external genitalia. Disorders of (or differences in) sex development (DSD) due to CAH are the most common causes of virilized female genitalia. Excess androgen exposure *in utero* leads to varying degrees of urogenital sinus formation, clitoromegaly, and labioscrotal fusion.

The goal of genitoplasty is to restore functional and esthetic anatomy, ensure preservation of sexual and urinary functions, and minimize psychological trauma, while aligning with contemporary principles of shared decision-making and ethical care. Surgical correction—feminizing genitoplasty—aims to achieve normal genital anatomy, preserve urinary and sexual functions, and ensure psychosocial well-being within an ethical, multidisciplinary framework.

GENDER ASSIGNMENT AND SOCIO-CULTURAL INFLUENCES

Gender assignment has traditionally been predominantly influenced by the sex of rearing, and treatment was dependent on the size of the phallus^[1] and it still holds, and in developed countries, as well.^[2] It has now been realized that gender identity is the result of a complex interaction

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between genes and environment. It is impossible to predict what gender any child will come to identify with.

Atypically developed genitalia affect the physical appearance, body image, functions of the urinary tract and gonads, and the psychological and psychosexual development. Therapeutic management of these patients is, therefore, not limited to “cosmetic” surgery. At present, parents make decisions regarding surgical options. Their strong wish to surgically “normalize” their child’s sexual anatomy is based on their view that genital surgery is “obvious” and “necessary” to assure their child’s-positive psychosocial and psychosexual adaptation. Surgery benefits the patients physically and psychosocially, but sometimes harms. Urgent surgery to create unobstructed outlets for urine has to be carefully evaluated, as early clitoral reduction surgery is categorized as cosmetic which may risk genital tissue sensitivity. Surgery is recommended for severe clitoromegaly. It has to be understood that there is also the emergence of activist platforms, advocacy, and human rights groups for children and families with DSD.

The role of healthcare professionals in the management of DSD includes counseling regarding the anatomy and physiology (e.g., hormone production, hormone receptors, and gross anatomy), helping in interpreting various test results, and informing the management team regarding parents’ decisions about gender assignment. Individual cases are referred to the hospital or institutional ethics committee (IEC) where clinicians, ethicists, members from the law as well as from the community get involved in shared decision-making. When IEC finds it difficult to negotiate, the matter is then referred to a family court to decide in the best interest of the child. Thus, there are several concerns:

- Parents seek to act before becoming fully informed about all the options
- Legal and ethical questions – patient autonomy and deferral of decision
- Lack of comparative outcomes associated with performing surgery early vs. later in life.

Current evidence indicates that many individuals with CAH retrospectively support childhood interventions, and a universal moratorium on early genital surgery is not justified. Instead, a case-by-case, individualized decision-making model is recommended, consistent with major ethical analyses.^[3-6]

Decision-making must incorporate improved information on long-term outcomes, transparent discussion of risks and benefits, informed consent and assent processes, and access to peer support groups. When uncertainty is substantial, parents may reasonably choose to postpone elective genital surgery, provided that potential health risks related to urogenital anatomy and function are monitored, and future sexual function and gender identity considerations remain central.

Shared decision-making should explicitly recognize that a choice is required, weigh evidence objectively, and integrate the family’s values and preferences. This necessitates participation of relevant subspecialists, structured parental involvement, and support for parents in processing emotional burdens—all delivered with unbiased, evidence-based information and avoidance of stigmatizing terminology. To ensure that consent is truly informed, it must be qualified, persistent, written, and repeated over time, with comprehensive disclosure of treatment risks, the option of surgical delay, and the availability of psychological support, consistent with established legal standards.^[7]

The data from 415 respondents (≥ 16 years) with a diagnosis of DSD from 14 specialized clinics in six European countries on the effect of timing of clitoroplasty and associated genital surgeries show that opinions are heterogeneous but there is a trend toward support for early-life intervention.^[8] When asked whether genital surgery should be postponed until the child attained the legal age of consent, 51.2% disagreed with a general postponement, 27% agreed, and 22% were unsure. Regarding the appropriate timing for genital surgery, 46% favored surgery during infancy, whereas 20% preferred the age range of 4–12 years. In a subset of 314 respondents considering the necessity of clitoral reduction, 38.2% agreed that it is necessary in girls with CAH, 14% disagreed, 18.8% were undecided, and 29% did not know. Among 323 respondents evaluating the timing of vaginoplasty, 66% approved performing the procedure in infancy or childhood rather than delaying until adolescence or adulthood.

Shared decision-making has three essential elements and six steps. The three essential elements include the following:

- Explicit acknowledgment that a decision is required
- Evidence concerning the risks and benefits of each option
- Process that takes into account the patient’s/family’s values and preferences.

The six steps of shared decision-making include the following:^[5]

- Inclusion of sub-specialists
- Involvement of parents in decision-making
- Help to parents in addressing their emotional feelings
- Avoidance of terms referring to genitals
- Providers should strive for objectivity which is evidence-based
- Parents should have received unbiased information which may lead to a consensus based on trust and understanding.

TYPES OF SURGERY

CAH due to 21-hydroxylase deficiency constitutes the most common cause of virilization in 46,XX individuals. The

resultant spectrum of genital masculinization—ranging from mild clitoromegaly to severe labioscrotal fusion and a high urogenital sinus—raises complex questions of gender assignment, timing and type of genital surgery, and long-term psychosocial outcomes.

Among the most contentious issues regarding those with CAH, involves XX females. Females with CAH are generally born with an enlarged clitoris and often a vagina that is atypically formed. Questions arise as to whether they should have a genital surgery or not—it is often recommended. Sometimes, dependent on the degree of masculinization of the genitals, there is a question whether they should be reared as males or females. Not infrequently, due to various reasons, such as the disease's late onset, some individuals will be reared as males, while others will request such an assignment. Recommendations as to the management of severely masculinized infants with CAH are controversial. When it was first suggested that severely masculinized females should be raised as males, the idea was generally rejected. Lately, however, the idea is being revived.^[9,10]

Genitoplasty

There are three types of genitoplasty: (1) nerve-sparing clitoroplasty [Figure 1], (2) Follow-up after ileal replacement

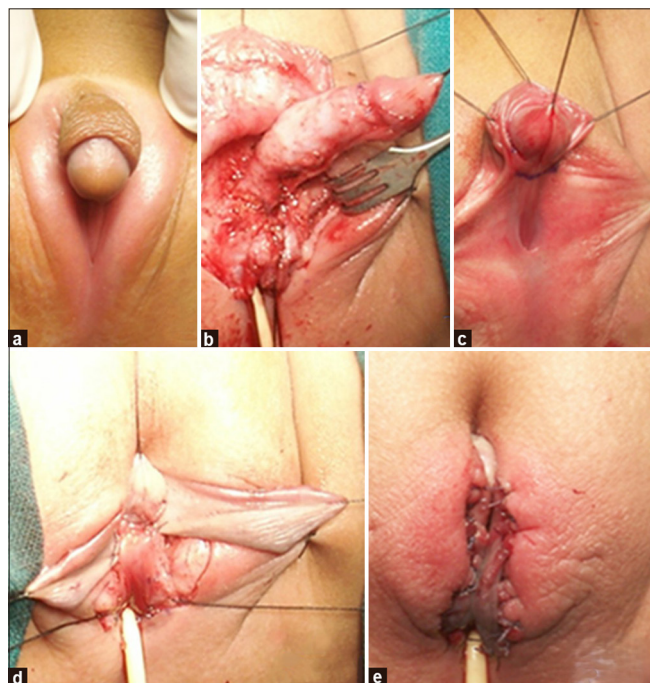


Figure 1: Nerve sparing clitoroplasty in an infant with moderate clitoromegaly; (a) Moderate clitoromegaly; (b) Opening of urogenital sinus; (c) Clitoris released; (d) Labia minora flaps created from preputial hood; (e) Post-operative appearance of recessed clitoris.

of high urogenital sinus. [Figure 2], and (3) Passerini's clitoroplasty [Figure 3]. The principles and ethics of the surgery are listed below:

- Multidisciplinary DSD team: Surgeon, endocrinologist, psychologist, and ethicist
- Individualized timing: Early (6–18 months) if medically indicated, or delayed with patient assent
- Surgical goals: Functional anatomy, preservation of sensation, cosmetic normalcy, and minimal scarring.

The surgical objectives include the following:

- Clitoral size reduction with neurovascular preservation
- Reconstruction of separate urethral and vaginal openings
- Functional and esthetic perineum and introitus
- Avoidance of stenosis and loss of sensation.

The operative techniques commonly employed are (1) nerve-sparing clitoroplasty which reduces hypertrophy while preserving the dorsal neurovascular bundle and glans sensitivity and (2) urogenital sinus mobilization which creates separate urethral and vaginal outlets with optimal orientation and depth [Figures 4 and 5].

Postoperative care includes the following:

- Hormonal optimization: Glucocorticoid/mineralocorticoid balance
- Catheter care and dilatation: Gentle, timed regimens
- Psychological counseling: Long-term adaptation support
- Follow-up: Assessment of function, cosmesis, and satisfaction through adolescence.

The long-term outcomes include (1) improved genital appearance and psychosocial adjustment and (2) preservation of clitoral sensitivity with nerve-sparing methods and



Figure 2: Follow-up after ileal replacement of high urogenital sinus.

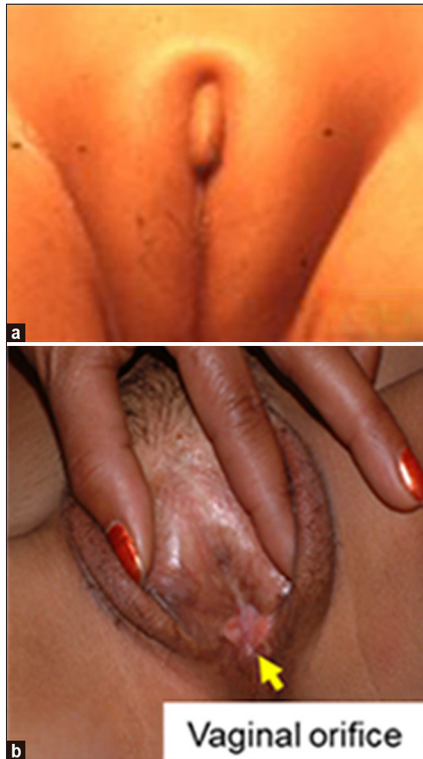


Figure 3: Long-term results “Passerini’s technique” of clitoroplasty; (a) clitoromegaly; (b) Follow-up around puberty.

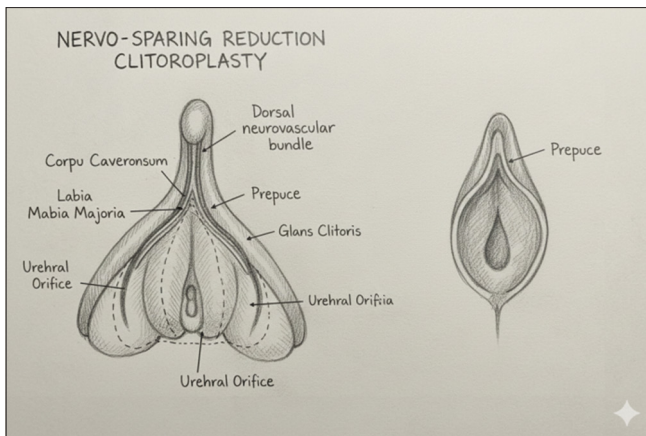


Figure 4: Nerve-sparing clitoroplasty.

minimization of recurrent stenosis or fistula with meticulous technique and dilatation compliance.

Future perspectives include (1) 3D anatomical mapping and custom surgical simulation, (2) minimally invasive, endoscopic-assisted sinus mobilization, (3) tissue-engineered vaginal grafts and biointegrative repair, and (4) global registry-based outcome tracking and patient-reported metrics.

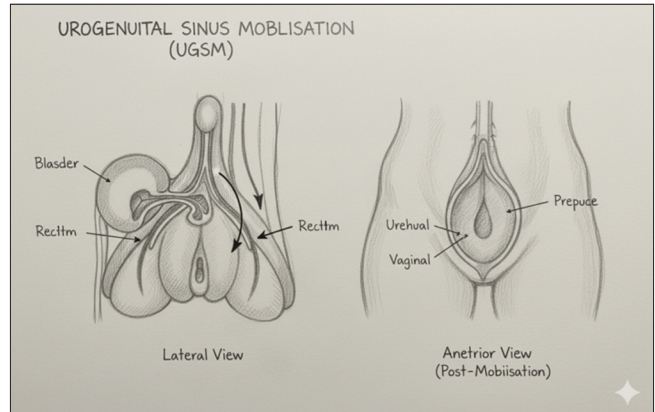


Figure 5: Urogenital sinus mobilization.

These data suggest that CAH persons predominantly favor interventions in childhood;^[8] a moratorium on one-for-all solutions is not justified and case-by-case decision-making is better suited.^[3-6] Efforts in improving information on long-term outcomes, informed consent, and assent should continue and contact should be made with support groups. If considerable uncertainty exists, the parents should be motivated to postpone elective genital surgery.

On the medical and surgical management of CAH, the European Society for Paediatric urology and the Society of Pediatric Urology of the United States standpoint also aim at (1) avoiding potential health hazards – anatomy and function of urogenital tracts, (2) meeting parents’ expectations, and (3) helping individual future satisfactory sexual function which is consistent with their gender identity.^[11]

Regarding the management of infants with DSD conditions and ambiguous genitalia in a survey, the pediatric urologists overwhelmingly favored female gender assignment for females with CAH, even if they were extensively masculinized.^[1,12] They recommended feminizing surgery – reducing the size of an enlarged clitoris – and considered that preservation of female fertility was of foremost importance and the masculinization of behaviors or inclinations was of lesser importance. There was a great difference of opinion regarding the optimal age for the surgery. There are case reports where patients have reported dissatisfaction with early genital surgery. However, the magnitude and extent of this satisfaction need further verification.^[13] On reaching adulthood, some individuals see themselves as distinctly non-male/non-male.^[14] This has given an advocacy for another category to be recognized as the “third gender.” Some authors advocate the consideration of a third gender or of even more gender categories.^[15,16] Australia has become the first industrialized country known to have removed the legal barrier to such gender assignment by allowing an X, signifying unspecified sex or intersex, in the sex field of passports, with the State of Victoria issuing a corresponding

birth certificate that lists sex as “indeterminate – also known as intersex.”^[9]

ETHICAL AND LEGAL CONSIDERATIONS

Ethical decision-making in the management of children with DSD requires a careful balance between medical indications, potential long-term outcomes, and the evolving autonomy of the child. Clinicians must recognize that parents often make decisions under conditions of emotional distress and uncertainty and therefore need structured, unbiased counseling that clearly outlines the risks, benefits, and alternatives to early genital surgery, including the option to defer intervention until the child can participate meaningfully in decision-making. Ethical practice mandates that information should be presented in a manner that supports parental understanding, avoids stigmatizing language, and respects the child’s future gender identity, bodily integrity, and sexual function.

From a legal standpoint, consent for elective genital surgery in infancy must meet a higher threshold of scrutiny [Box 1]. Jurisprudence has established that valid consent must be qualified and persistent, documented in writing, and based on complete disclosure of treatment risks, the possibility of postponing surgery, and the availability of psychological support. Importantly, authorization should be obtained on multiple occasions over a reasonable time, rather than through a single consent event, to ensure deliberation and to minimize coercion. These principles, articulated in rulings such as the Constitutional Court of Colombia,^[7] underscore the obligation of clinicians and institutions to ensure that surgical decisions are ethically defensible, legally robust, and firmly grounded in the best interests of the child.

CONCLUSION

46,XX DSD due to CAH presents a broad spectrum of genital virilization that affects anatomy, function, body image, and long-term psychosexual development. Feminizing genitoplasty can restore functional and esthetic anatomy and improve psychosocial outcomes, but its timing and

scope remain ethically complex and clinically individualized. Current evidence does not support a universal moratorium on early genital surgery; instead, a case-by-case approach, grounded in multidisciplinary evaluation, is most appropriate. Parents require comprehensive, unbiased information on risks, benefits, alternatives, and long-term outcomes, recognizing that gender identity cannot be predicted with certainty and that some cases warrant deferral of elective surgery.

Survey data indicate heterogeneous views but there is a general trend toward support for early intervention in selected cases. Nonetheless, consent for such procedures must meet heightened ethical and legal standards – qualified, persistent, written, and reaffirmed over time – to ensure voluntariness and protect the child’s best interests. Advances in nerve-sparing techniques and urogenital reconstruction have improved functional outcomes, but long-term follow-up, psychosocial support, and transparency about uncertainties remain essential. Overall, management of CAH-related 46,XX DSD requires individualized, evidence-based care that integrates surgical expertise, ethical safeguards, parental partnership, and sustained, multidisciplinary support.

Ethical approval: Institutional Review Board approval is not required.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patients have given their consent for their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Box 1. Legal issues in elective genital surgery

- To ensure that the consent is truly informed, it has to be “qualified and persistent”
- The consent must be in writing
- The information provided must be complete
- The parents must be informed about the dangers of current treatments
- The possibility of delaying surgeries and giving adequate psychological support to the child
- The authorization must be given on several occasions over a reasonable time period.^[7]

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