**Erratum**

In the Opinions Section (Kives, *Congenital Adrenal Hyperplasia* 57/6: 411), the opinions were omitted. The Opinions Section is reprinted in its entirety below.


**Opinions Section**

**Early versus Late Intervention of Congenital Adrenal Hyperplasia**

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At the annual NASPAG meeting in San Diego, there was much discussion about the appropriate timing for treating young girls with congenital adrenal hyperplasia. Traditionally a reduction clitoroplasty and cutback vulvoplasty are often performed in the neonatal period. When surgery is performed in the neonatal period, a second surgery is often required at the time of puberty to improve vaginal caliber. More recently, however, some experts are suggesting delaying corrective surgery until adolescence to possibly improve the surgical results. This allows the patient to participate in the decision for surgery.

Females who are deficient in 21-hydroxylase will experience clitoral hypertrophy, labioscrotal fusion, and displacement of the urethral orifice. The degree of virilization varies among individuals depending on the degree of androgen exposure. Immediate treatment involves identifying those individuals with the sodium wasting form of 21-hydroxylase deficiency. Sodium wasting may result in hyponatremia, hyperkalemia, dehydration, and even death. Surgery to correct masculinization of the external genitalia, however, is not an emergency. It is therefore not surprising that a delay in the surgery until adolescence is being suggested as an alternative approach.

To further address the issue of appropriate timing for surgery, I have obtained two expert opinions on the subject, one in favor of delaying surgery and the other favoring a more traditional approach.


**Feminizing Genital Surgery: What Should Be Done and When?**

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**Introduction**

The role of genital surgery in the treatment of intersex conditions is under intense scrutiny. There is significant consumer dissatisfaction, with intersex peer support

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groups around the world expressing condemnation of infant genital surgery performed for cosmetic reasons.\textsuperscript{1-5} Challenges to the current practice of infant feminizing genital surgery are being made at every level including that of alleged Human Rights abuses.\textsuperscript{4} Clinicians working in this area are unlikely to escape question and need to be able to demonstrate that if they recommend genital surgery, there is an acceptable evidence base to justify it as being in the best interest of the child.

\textbf{Indications for Surgery}

In Europe and North America, current standard practice advocates feminizing genital surgery for all children with ambiguous genitalia assigned to a female sex of rearing.\textsuperscript{5,6} This practice is relatively recent and became popular in the last half of the 20\textsuperscript{th} century partly because of the availability of appropriate surgical expertise. From that time, feminizing genital surgery was rapidly accepted as the appropriate treatment and was not questioned by the medical profession until very recently, after prompting by adult intersex patients.

A standard infant feminizing genitoplasty consists of removing some of the clitoris to reduce its size, constructing a vagina or opening the vaginal introitus, and sometimes refashioning of the labia. Proponents of feminizing genitoplasty in infancy cite both psychological reasons to operate—such as a more stable gender identity development and a better psychosexual and psychosocial outcome—and physical reasons such as menstruation and subsequent intercourse in adulthood. To justify such a serious intervention as irreversible genital surgery, it is important that there should be good evidence of the benefits of such surgery.

\textbf{Quality of Evidence}

Unfortunately there is little available objective long-term outcome data from representative groups of intersex adults. Collection of data has been hampered by widespread policies of non-disclosure of diagnosis. This leaves patients unaware of their diagnosis, unable to access medical or peer support, and unavailable for recruitment into outcome studies. In addition, surgery for ambiguous genitalia is so widespread that it is difficult to recruit those few patients who have not had surgery. Most published studies are collections of small series with variable outcome measures and indicators. Patient opinion largely remains absent and despite calls for more equitable doctor-patient collaboration, no such studies yet exist.

\textbf{Genital Surgery and Psychological Outcomes}

Research in the 1950s introduced the view that gender development was mainly a function of postnatal environmental factors. It was postulated that babies are psychosexually neutral at birth and so have the potential to develop a male or female gender, depending on the way their parents treat them, on the appearance of their genitalia as male or female, and on the way society perceives their gender and reacts to it.\textsuperscript{7} On the basis of this work, the optimal gender policy recommended that sex should be assigned solely on the basis of the external genital appearance as being suitable for male or female sexual function, and that all should then undergo early genital cosmetic surgery. As the majority of intersex cases were felt to have inadequate male genitalia, infant feminizing surgery became standard treatment. It is clearer now that gender identity differentiation is a complex and multifactorial process, involving prenatal influences, and postnatal hormonal, social and psychological determinants.\textsuperscript{8,9} The relative importance of external genital appearance in this process is unknown but may be minimal. The idea that one can stabilize gender development with infant cosmetic surgery remains unconfirmed, with a lack of any evidence for the role of genital appearance in gender identity development.

In addition to gender stability, there is an expectation that infant feminizing genitoplasty improves psychological outcomes. Most medical literature assumes that psychological outcomes are better after cosmetic surgery and often recommend that gender assignment and surgery should be at an early age so that the child never needs to be aware of having been born with an intersex condition.\textsuperscript{10} However there is simply no available evidence at the moment showing that genital surgery improves psychological outcome. It has been suggested that all intersex children are at risk of psychological problems, irrespective of genital appearance and surgery\textsuperscript{11} and if this is the case, then surely appropriate psychological support is the remedy rather than irreversible genital surgery.

Relief of parental anxiety has also been cited numerous times as one of the prime indications for feminizing surgery in childhood. However, this relief does depend upon the clinician’s reassurance that surgery will ensure good outcomes. It may be that if parents are aware of the risks of surgery and the lack of evidence of good long-term outcomes, that they will not choose surgery. Their understandable anxiety may still be relieved in many ways, including full information, contact with other families, and expert clinical psychological input.
Vaginoplasty

Although it is true that most of those assigned female gender will wish to have vaginal intercourse, and treatment to produce a vagina will be necessary at some stage, the appropriate timing of such surgery is also under dispute. In children with ambiguous genitalia assigned female, vaginoplasty is an integral part of feminizing genitoplasty and is commonly performed during the first year of life. This is despite the fact that the child will not menstruate for a further 10 or so years (if she has a uterus) and is unlikely to be sexually active until after puberty. Early infant vaginoplasty might be justified if there were good evidence that it produced better long-term anatomical, cosmetic, and functional outcomes than later delayed surgery; however, this does not seem to be the case. Most of the follow-up studies of vaginoplasty have looked at the exteriorization of the vagina in congenital adrenal hyperplasia (CAH). Many recent studies have demonstrated high rates (40–100%) of introital stenosis and frequent requirements for repeat reconstructive surgery in adolescence before tampon use or intercourse.\(^{12,13}\) Other vaginoplasty techniques, such as using an intestinal segment, involve major surgery and long-term problems such as persistent vaginal bleeding and mucus discharge are not uncommon.\(^{14}\)

Delaying vaginoplasty until puberty may lead to better vaginal healing when the skin is fully estrogenized. Adjunctive treatments to prevent stenosis such as vaginal dilators may be helpful, but cannot be used in the young child. Vaginal dilators alone may be sufficient to create a vagina in some conditions, thus replacing the need for surgery altogether. In those girls with a uterus, menstrual drainage usually occurs without complication via the urogenital sinus. If in doubt this can be confirmed by an endoscopy assessment. If there is an obstruction to menstruation, then vaginoplasty can be performed in early puberty. In those without a uterus, surgery can be deferred as long as preferred. Deferring vaginoplasty until adolescence may reduce the number of procedures required to achieve a satisfactory vagina. Certainly, it would avoid young children undergoing a painful operation to produce a vagina which is unnecessary in childhood and may well need further revision surgery. Most important, it allows the patient to be fully informed as to the benefits and risk of surgery and to be provided appropriate psychological support.

Clitoral Surgery

Clitoral reduction is the commonest clitoral procedure performed nowadays, although clitorectomy is still the procedure of choice in some centers in Europe.\(^{15}\) Clitoral surgery aims to make the clitoris smaller and more “feminine” looking. The clitoris is an erotically important sensory organ, and its only known function is its contribution to female sexual pleasure. However, sexual response is multifactorial, and the exact contribution of the clitoral glans, clitoral hood, and clitoral corpora to sexual function is not well understood. Recent work on the neuroanatomy of the human fetal clitoris has demonstrated an extensive network of nerves completely around the tunica with multiple perforating branches entering the dorsal aspect of the corporeal body and glans.\(^{16}\) Any incision to the clitoral glans, corpora, or hood may risk damage to the innervation. Other surgical risks are loss of the clitoral glans from vascular insufficiency, and pain during sexual arousal in the remaining corporal tissue following either amputation of the corporal bodies at the pubic bifurcation or recession of the corporal tissue. Additionally, the effect on future sexual function of removing the paired clitoral corpora that make up the clitoral body is unknown.

Only a few studies look at the impact of clitoral surgery on subsequent sexual function in adult life. Two studies looked at psychosexual function in women after genital surgery for congenital adrenal hyperplasia.\(^{17,18}\) In both studies, women with CAH had significant sexual dysfunction when compared to control groups and attributed their difficulties to their surgery. A third study compared a group of women with ambiguous genitalia due to various different intersex diagnoses, some of whom had undergone genital surgery and some of whom had avoided surgery. While both groups had significant sexual dysfunction, specific difficulties with orgasm were much more frequent in the group who had undergone clitoral surgery.\(^{19}\) Most recently, genital sensation has been shown to be impaired in a small group of women with CAH who had undergone feminizing genitoplasty in childhood.\(^{20}\) These studies confirm that cosmetic surgery to the clitoris does not ensure improved adult sexual function and in fact damages sexual function.

Conclusion

Feminizing genital surgery has been regarded as the cornerstone of treatment and parents and clinicians view it to a great extent as a “cure.” However, review of the current literature does not support the use of feminizing infant genitoplasty as an effective treatment for stable gender or psychosexual development. While immediate cosmetic results can be good, the evidence for satisfactory post-pubertal anatomical and cosmetic outcomes is poor. A functional vagina is not necessary for a young girl prior to menstruation or sexual intercourse and vaginal surgery can be deferred.
until later in life in the majority of cases. This should limit the total number of operations an individual will undergo, reduce the substantial risk of fibrotic stenosis, provide patients with greater choice of vaginal enlargement interventions at adolescence and allow them to be involved in the decisions.

In addition, there is now emerging evidence of damage to future adult sexual function caused by clitoral reduction. In the absence of firm evidence that infant feminizing genital surgery benefits psychological outcome, the option of not performing infant genital surgery must be discussed with the family. Truthful information must be given to the patient and their family from the outset on the aims of the surgery and the risks to their daughter’s future sexual function. It is time to consign the policy of routine feminizing genital surgery for all to the history books. The option of specialized psychological and peer support as a realistic alternative to surgery must be made available to all families.

References

1. Intersex Initiative (IPDX): Available: www.ipdx.org

Early Intervention of CAH Surgical Management

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Introduction

Our management protocol of 46XX newborns with congenital adrenal hyperplasia (CAH) is to first establish a definitive diagnosis and then to treat this disorder as quickly and efficiently as possible. Through a multi-disciplinary approach, we involve genetic counselling, psychology, and social work to help with the evaluation and care of these patients. If they have ambiguous genitalia, we perform a perineal reconstruction as early as possible to minimize the period of gender uncertainty. The surgical goals of perineal reconstruction are primarily to create an opening for the vagina, to create a normal looking vulva, and to separate the vagina from the urinary tract, thus resulting in a female-appearing perineum.

Early reconstructive surgery allows us to use all available tissue, resulting in an enhanced healing response. By performing a one-stage procedure, one avoids discarding the redundant skin of the enlarged clitoris, which occurs in a two-stage surgery.

This surgery on the masculinized external genitalia facilitates gender rearing as a female, which can prevent