Mini-Review

Management of Vaginal Agenesis

Rola S. Nakhal MD *, Sarah M. Creighton MD, FRCOG

UCL Institute of Women's Health, University College London Hospitals, London, UK

A B S T R A C T

Rokitansky syndrome and complete androgen insensitivity syndrome are the most common causes of vaginal agenesis. Treatment should be deferred until adolescence to allow informed consent and compliance. The best treatment for vaginal agenesis remains controversial although vaginal dilation therapy is still widely considered the first line treatment because success rates are high and associated risks are low. A variety of surgical options are also available, each with enthusiastic proponents. Long-term outcome studies on most surgical techniques, however, are still lacking and until recently most studies have reported on success rate in terms of anatomical success only, without including sexual function. Moreover, the medical literature lacks prospective comparative outcome studies, meaning that current choice of surgical procedure relies greatly on the surgeon's preference and experience.

Key Words: Vaginal agenesis, Vaginal hypoplasia, Vaginal aplasia, Vaginal dilation, Vaginoplasty, Rokitansky syndrome, Complete androgen insensitivity syndrome

Introduction

Congenital vaginal agenesis is most commonly seen in women with Rokitansky syndrome (also referred to as Mayer-Rokitansky-Kuster-Hauser syndrome or Müllerian aplasia), and complete androgen insensitivity syndrome (CAIS). In addition it can be associated with other rarer complex conditions affecting the urinary and gastrointestinal tracts, such as cloacal and anorectal anomalies. Women with Rokitansky syndrome have a XX karyotype and normal functioning ovaries but an absent or rudimentary uterus and a short vagina resulting from failed embryonic development of the Müllerian duct. It occurs in one in 5,000–10,000 female births; however, the exact etiology is still unknown. Women with CAIS, which affects one in 13,000–40,000 live births, have 46,XY karyotype, testicular gonads, absent Müllerian structures, and a short vagina. This is attributed to a mutation in the androgen receptor gene which renders the body insensitive to testosterone and hence results in a female phenotype with female external genitalia. Since patients with Rokitansky syndrome and CAIS have normal breast development either from normal ovarian hormones in the former condition or from peripheral conversion of testosterone to estradiol in the latter one, these patients typically present in adolescence with primary amenorrhea. Detailed investigation is required to diagnose and differentiate these conditions; however, in a clinical setting the vaginal findings are identical with an absent or short vaginal dimple. Surgical and non-surgical treatments are available to lengthen the vagina and facilitate penetrative sexual intercourse.

Timing of Treatment

In the majority of cases these conditions present in adolescence which means the patient can be fully involved in decisions about the type and timing of treatment. Where presentation is earlier in childhood, it is accepted that both non-surgical and surgical methods of vaginal creation are best deferred until adolescence or even adulthood when the patient has reached physical and psychological maturity.[1,2] This allows for proper decision making and also increases the compliance with vaginal dilation therapy whether used as primary treatment or post-operative adjuvant treatment to prevent vaginal stenosis.

The requirement for vaginal lengthening before intercourse is of course only part of the devastating implications of these conditions. Both conditions result in infertility. In CAIS disclosure of an XY karyotype and the decision about gonadectomy are difficult concepts for an adolescent girl to manage. As the risk of malignancy is found to be low before puberty, the current standard treatment is to delay gonadectomy until puberty. This allows pubertal changes to occur as a result of peripheral conversion of testosterone to estradiol. In addition, delaying gonadectomy will provide the time for the patient to be involved in the decision. All patients with disorders of sex development should be cared for by a multidisciplinary team which includes a psychologist.

Treatment Choice

The best treatment for vaginal agenesis is still controversial. In the USA and UK vaginal dilation therapy is considered the best first-line treatment[2] and surgery is reserved for cases when vaginal dilation therapy fails or when a patient is ineligible for vaginal dilation due to previous perineal surgery. However, in many European

The authors report no conflicts of interest.

* *Address correspondence to:* Rola S. Nakhal, MD, University College London Hospitals, 2nd Floor North, 250 Euston Road, London NW1 2PG.

E-mail address: rolanakhal@homail.com (R.S. Nakhal).

1083-3188/$ - see front matter
doi:10.1016/j.jpag.2011.06.003

countries, surgical vaginoplasty such as a laparoscopic Vecchietti or Davydov procedure is the first line treatment and vaginal dilation is only used post-operatively. There is a lack of long term outcome data on surgical and non-surgical options. In addition there are no comparative studies of different techniques. It must be remembered that the main objective of surgery is not just to create a passageway for penetration but to facilitate enjoyable sexual intercourse. Only recently has sexual function data been included in outcome studies. At present the choice of the method depends on the genital configuration, previous surgical attempts, and, above all, the surgeon's preference and expertise.[3]

Nonsurgical Methods

Vaginal Dilation

Vaginal dilation therapy for vaginal agenesis was first described by Frank in 1938.[4] It involves the use of vaginal molds, initially made from pyrex and currently made from plastic, of increasing width and length. The patient is asked to apply gentle pressure on the vaginal dimple for at least 30 minutes daily for several months with the aim to achieve a vaginal length of 7–8 cm. This therapy works by the pressure effect which acts to progressively stretch the vagina with time. Thus the prerequisite for this treatment is first a healthy non-scarred vaginal dimple which is amenable to stretching and second, but more important, a motivated patient who is ready to adhere to such a lengthy treatment.

Because vaginal dilation is a non-invasive and inexpensive method with a high success rate, of up to 90%,[5] it has been recommended as first line treatment for vaginal agenesis by several medical bodies including the American College of Obstetricians and Gynecology Gynecologists (ACOG).[2] However, the definition of "success" in earlier studies has been criticized[6] as it was either vaguely defined or mainly focused on the "anatomical success".[5,7,8] Later studies included sexual function as an integral element of success, which highlighted the difficulties encountered with this type of therapy. These include low compliance and patient dissatisfaction[9] as the regime was described by patients as distasteful,[10] painful and a constant reminder of their abnormality.[6] Despite the fact that vaginal dilator therapy is mainly patient driven, psychological support and supervised programs are necessary to maintain patients' motivation. The obstacles to dilation therapy can be overcome when treatment is delivered by a multi-disciplinary team which acts to boost patient's confidence as described by Ismail-Pratt et al[11] who reported non-painful sexual intercourse in 81% of participants with a background anatomical success rate of 86%. Another study also concluded that despite the difficulties encountered with lubrication, pain, and orgasm, the degree of satisfaction with the overall sexual life was comparable to the control population.[12] However, the condition itself does have a negative impact on sexual and emotional wellness and anxiety levels are higher in women with Rokitansky syndrome who have undergone vaginal dilation treatment compared to those who did not require vaginal dilation.[13]

The timing to commence dilation therapy is elective and is best planned when the patient is emotionally mature.[2] The question remains whether the patient should be given the option to choose between vaginal dilation or first line surgical option, bearing in mind that vaginal dilation might still be needed as adjuvant treatment after surgical reconstruction.

In fact the merits of vaginal dilation therapy extend beyond its noninvasive nature to other important aspects. First of all, even when dilation therapy fails, Routh et al[14] have proven that initial progressive perineal dilation followed by vaginoplasty would still be more cost-effective than initial vaginoplasty in 99.99% of simulations. In addition, until now the key focus of clinical management has been to increase vaginal size and to achieve penetrative sexual intercourse. However, the functional success of vaginoplasty is not necessarily proportional to the length of the new vagina. Initiating vaginal dilation therapy can prompt concomitant psychological support which should not only improve the outcomes of any treatment but would also orient patients towards realistic expectations even when surgery is still needed.

Surgical Methods

Surgical vaginoplasties can be subdivided into the following categories:

1. Creation of a perineal pouch. This includes the Williams vaginoplasty and subsequent modifications.
2. Lining a neovaginal space. This includes procedures based on the McIndoe technique where a neovaginal space is dissected between the bladder and rectum, then lined with different types of tissue.
3. Laparoscopic procedures. These methods include the Vecchietti procedure and Davydov procedures.
4. Intestinal vaginoplasty.

Creation of a Perineal Pouch

Initially described by Williams in 1964, this technique fell into general disuse until modified by Creatsas et al in 2001.[15] It involves a U-shaped incision extending from the medial side of the labia at the level of the external urethral meatus down across the perineal body to form a skin flap. The tissues are then mobilized and sutured in layers to form a pocket in the perineum to allow coitus.[3] In a study on 178 Rokitansky patients, anatomical success was achieved in 96% while 94% reported sexual satisfaction in a non-validated follow-up assessment.[16] Possible complications of the Creatsas vaginoplasty are hematoma, wound opening, and infection in addition to excessive hair growth from the skin flap. It is difficult to explain the success of the Creatsas modification as the Williams procedure fell into disuse because the perineal pouch was often too short and superficial for comfortable penetration. It is likely that the