An observational study of women with müllerian agenesis and their need for vaginal dilator therapy

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Objective: To assess vaginal development, sexual activity, and the efficacy of vaginal dilator therapy in women with vaginal agenesis (Mayer-Rokitansky-Kuster-Hauser syndrome).
Design: Retrospective case review.
Setting: Tertiary referral university teaching hospital clinic for disorders of sexual development and differentiation (DSDD).
Patient(s): Eighty cases of Rokitansky syndrome.
Intervention(s): None.
Main Outcome Measure(s): Sexual activity and vaginal dimensions.
Result(s): The retrospective case review examined vaginal dimensions and sexual activity at presentation with further evaluation at completion of vaginal dilator therapy. Eleven of 80 patients had undergone surgery in the past; six of these 80 women had received dilator training elsewhere, and four were sexually active. Sixty-three of 80 patients had not undergone any previous treatment. Seventeen were having satisfactory sexual intercourse, 16 were having unsatisfactory sexual intercourse, and 26 had never been sexually active; for four women, no information had been recorded. A total of 32 patients underwent vaginal dilator treatment, and 25 completed the therapy. Their vaginal length increased from 3.2 cm (range: 0 to 7 cm) to 6.1 cm (range: 3 to 9 cm).
Conclusion(s): Diagnosis and management of müllerian agenesis may be achieved without the need for surgery in the majority of cases. Dilator treatment for vaginal agenesis should be offered as first-line treatment, coordinated by a specialist nurse with input from a psychologist. (Fertil Steril® 2011;96:483–6. ©2011 by American Society for Reproductive Medicine.)

Key Words: Rokitansky syndrome, vaginal agenesis, vaginal dilator, vaginoplasty

Müllerian agenesis was described by Mayer in 1829, Rokitansky in 1838, and Kuster in 1910; consequently, the condition was named Mayer-Rokitansky-Kuster-Hauser syndrome (1) but is frequently now shortened to Rokitansky syndrome. There are different grades of severity and spectrums of association with other abnormalities of the renal tract and skeleton. The patients present with primary amenorrhea, normal secondary sexual characteristics due to a normal hypothalamic-pituitary-ovarian axis, and a female karyotype. The external genitalia have a normal appearance, but the vagina is shortened, with usually just a "dimple" or at most a depth of 1 to 2 cm. The incidence of müllerian agenesis is about 1:5,000 female births (2). It is unclear why the müllerian tract fails to develop; although the condition may have genetic origins, not all the genes involved have been elucidated (3). Uterine transplantation is presently being explored, with some success in animals (e.g., sheep), although it is still a long way from being an option for women (4).

Received March 19, 2011; revised May 4, 2011; accepted May 15, 2011; published online June 17, 2011.
F.B. has nothing to disclose. J.M.G. has nothing to disclose. A.H.B. has nothing to disclose.
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Diagnosis may be made via physical examination, hormone profile, and radiologic imaging. Laparoscopy is not required for diagnosis, but it may be indicated if there is abdominal pain.

A neovagina can be created by various vaginoplasty procedures. Several plastic surgery techniques are available. In McIndoe vaginoplasty, a split skin graft is placed over a mould that has been inserted into a space created where the vagina should be; in tissue expansion vaginoplasty, expansion balloons are inserted into the labia and inflated with water over a period of 2 weeks to stretch the labial skin folds sufficiently to fashion a vagina. An artificial vagina can be created from bowel or from peritoneum. The Davydov technique may be performed by minimal access laparoscopic surgery, and Williams vaginoplasty uses the labia to create a pouch, but the result presents an awkward angle for intercourse.

All the surgical procedures have recognized complications and potential long-term side effects. There is a risk of scarring which can cause stenosis, with resultant dyspareunia or apareunia (5). Inadvertent damage to bowel and bladder can occur, and vesicovaginal fistulae have been reported (5–7). Sigmoid colpoplasty can result in excessive, malodorous vaginal mucus (8) or neovaginal prolapse (9), bowel obstruction, leakage of intestinal anastomosis, necrotizing fasciitis (9), rectoneovaginal fistula (10), or carcinoma (11, 12). Additionally, distortion of the pelvic anatomy and the repositioning of the ovaries can make transvaginal retrieval of

oocytes for surrogacy treatment technically more difficult in a patient who has had surgical vaginoplasty (13). Thus, although surgery may appear to be an attractive quick fix to patients, postoperative use of vaginal dilators (sometimes referred to as "trainers") is often still required to maintain patency of the neovagina.

An alternative to surgery is the creation of a neovagina by pressure dilatation alone, as first described by Frank in 1938 (14). This technique allows a vagina to be created from normal vaginal skin, giving it more normal characteristics. Most importantly, this method also allows women to participate in a patient-centered approach to treatment. We present an overview of patients attending our clinic and an audit of the efficacy of this method to achieve vaginal dilatation.

MATERIALS AND METHODS

The Leeds Clinic for Disorders of Sexual Development and Differentiation (DSDD) was established in 1997 as a tertiary referral service for the north of England. A database of all patients has been maintained, amounting to approximately 700 new referrals of which 88 have the diagnosis of müllerian agenesis (Rokitansky syndrome). We performed a retrospective review of 80 cases of Rokitansky syndrome (with eight case notes being unavailable). Institutional review board approval was not required, as the protocol was assessed by the Leeds Research Ethics Committee who considered the study to constitute a retrospective review of standard therapy.

Rokitansky syndrome was defined as the presentation of primary amenorrhea, normal secondary sexual characteristics, a normal hypothalamic-pituitary-ovarian hormone axis, and the absence of a uterus. The pelvic anatomy was assessed by either ultrasound scan, magnetic resonance imaging (MRI), laparoscopy, or a combination of these, most often by the referring clinician before the patient was referred to the DSDD clinic.

We reviewed the patients' case notes for information recorded about the method of diagnosis, the outcome of previous treatment, counseling, sexual activity, evaluation of the need for therapy to create a neovagina, and assessment of the outcome of vaginal dilatation treatment. For those who had had vaginal dilator therapy, we collected data on the time taken for the creation of the neovagina, the number of inpatient and outpatient appointments, the changes in vaginal length and width through treatment, the anatomic and functional success, the use of counseling, the complications, the reasons for incomplete treatment, and any requirement for secondary treatments.

The specialist nurse who runs the dilator clinic and instructs and supervises the patients throughout their treatment was their main point of contact. Before commencing therapy, it is standard practice for the patient to see a clinical psychologist, who assesses the patient's readiness to commence therapy and her acceptance of the use of dilators, and determines whether the patient's expectations for the treatment are realistic.

The specialist nurse teaches the patient how to use the dilators, either as an inpatient or an outpatient. Patients are admitted if they live a great distance from the clinic or if they would prefer therapy to start as an inpatient. After this, the patients are seen regularly in the outpatient department at intervals decided by the specialist nurse and the patient, ranging from weekly to every 2 months.

Increasing sizes of handheld Perspex or plastic dilators are placed at the vaginal dimple, and pressure is used to create a vaginal space. Dilators are used three times a day for 15 to 20 minutes each session. Progression onto the next size occurs when the patient and the nurse are satisfied by the present size. Oral analgesia or local anesthetic gel (lidocaine hydrochloride 2%, Instillagel; CliniMed Ltd.) are used as required (15). The nurse monitors the patient's progress by measuring both digitally and with a dilator the width and length of the vagina. Sexual satisfaction is assessed in a qualitative fashion by direct questioning from the specialist nurse. Patients are reminded that their newly created vagina has the same potential for sexually transmitted infections and that they must therefore follow routine precautions.

RESULTS

Between February 1997 and April 2010, 88 patients with Rokitansky syndrome were seen in the clinic (age range: 15 to 48 years). Eight case notes were unavailable, leaving 80 for review. The patients had been referred from both local specialists (gynecology and pediatrics) and also from farther afield, including overseas. The reason for referral included confirmation of the diagnosis (12 women), creation of neovagina (41 women), and surrogacy (15 women).

Eleven patients had undergone previous vaginoplasty surgery. One had a colonic neovagina created 30 years previously; she requested removal due to excessive discharge and subsequently was satisfied with the use of dilators alone. One patient had a Williams vaginoplasty 20 years earlier; she was satisfied but continued to use dilators as part of her daily routine. Two had had a Davydov procedure 15 to 18 years earlier, and both were satisfied. Seven patients had had a tissue expansion vaginoplasty 7 to 12 years earlier; four were satisfied, two had dyspareunia, and one had troublesome hair growth in the vagina. Six of the 80 patients had already received dilator training; four were sexually active, and two were not. Of those who were sexually active, two were satisfied with sexual intercourse, and two were not. Sixty-three patients had not undergone any previous treatment; 17 of these were having satisfactory sexual intercourse, 16 were having unsatisfactory sexual intercourse, and 26 had never been sexually active (four were not recorded).

A total of 32 patients underwent dilator treatment at the specialist clinic. The mean age at the start of treatment was 19.5 years (range: 15.7 to 26.9 years). Fourteen patients had never attempted sexual intercourse, and 18 patients had attempted sexual intercourse before treatment but had found it unsatisfactory due to incomplete penetration or dyspareunia. Twenty-five achieved success, which was defined as either satisfactory sexual intercourse or a normal vaginal length of 7 to 13 cm (16). The average age of finishing treatment was 19.7 years (range: 15.8 to 27.2 years). The mean time taken for completion of treatment was 167.9 days with a range of 1 to 985 days (median: 86.5 days; lower quartile 14.7 days; upper quartile 239.3 days). The patient who had only 1 day of treatment had attended because of deep dyspareunia but only required a single teaching session with the dilators; subsequently she rated her sexual intercourse as satisfactory. The patient who took 985 days had very poor compliance and found it difficult to stay motivated. Other reasons for prolonged duration of treatment included the need for more counseling, failure to make or attend appointments, and personal problems unrelated to the diagnosis. Patients were seen either as inpatients (four women), outpatients (12 women), or as a combination of both (16 women). There was no difference in the outcomes between these three groups. The mean number of times patients were seen by the specialist nurse was 5.6 (range: 2 to 16).

Of the 25 patients with successful treatment, 15 had attempted sexual intercourse but had been unsuccessful or had experienced significant difficulties, and 10 had not yet embarked upon a sexual relationship. The mean vaginal length as measured on digital examination before treatment was 3.2 cm (range: 0 to 7 cm). After treatment, the average length was 6.1 cm (3 to 9 cm), an increase of 3.2 cm (0 to 7 cm) ($P<.001$). The mean initial vaginal length as measured with dilators was 4.7 cm (range: 2 to 8 cm) with post-treatment measurements of 8.2 cm (range: 5 to 10 cm), an increase of 3.5 cm (0 to 7 cm) ($P<.001$). The width of dilators increased from 19.5 mm (range: 15 to 30 mm) before treatment to 28.1 mm (20 to 35 mm), an average increase of 8.5 mm (0 to 15 mm) ($P<.001$). Mild pressure was applied when measuring using the dilator, but no pressure was applied when measuring digitally; all measurements were performed by the same specialist nurse. The pretreatment vaginal dimensions were smaller in those who had not yet attempted