Congenital malformations of the genital tract

And their management

D. Keith Edmonds FRCOG, FRANZCOG

Consultant Obstetrician and Gynaecologist,

Queen Charlotte's and Chelsea Hospital,

London,

U.K.

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ABSTRACT

Whilst congenital malformations of the genital tract are not common, the sequelae of their presence can be serious. The practising gynaecologist must be aware of the range of congenital abnormalities that may occur and the symptoms that may result from them. Failure to manage these patients correctly may have long-term sequelae for their psychological, sexual and reproductive health. The involvement of a multi-disciplinary team in dealing with these patients is imperative and preparation for surgery, particularly in congenital malformations of the vulva and the vagina are imperative if the long-term sexual function in these patients is to be fulfilled. Surgical correction in adolescence of vulval abnormalities is solely related to sexual function as most of the reconstructive surgery is done in childhood. For the management of MRKH syndrome, the recommendation is now that passive dilatation by Franks' technique is the treatment of first choice and only if that fails should surgical approaches be embarked upon. The results of the surgery are similar in all techniques and the particular surgical centre will have its own preference of which technique it adopts. Congenital absence of the cervix is a complex surgical problem and should be dealt with solely in centres with expertise.

KEY WORDS

Vulval abnormalities; vaginal anomalies; vaginal obstruction; congenital absence of the vagina; cervical atresia; haematocolpos; haematometra.

INTRODUCTION

A number of congenital malformations may not manifest themselves until adolescence and some congenital malformations which have been present at birth or in childhood may remain problems throughout the adolescent years, particularly following the onset of puberty. The genetics and aetiology of these disorders has been referred to extensively in the previous chapter and the management of these disorders in adolescence is a complex, multidisciplinary problem. Not only does the team caring for the adolescent need to address the anatomical difficulties but there are functional, reproductive and psychological issues which also need attention and very often by health professionals with particular expertise in these areas, especially that related to the psychological health of these young women.

In addressing the various malformations and abnormalities, the conditions have been divided into various sections for ease of discussion.

CLINICAL ASSESSMENT

In considering the malformations, some universal principles of clinical management need to be employed. It is imperative that the correct diagnosis of the underlying conditions are made and also that patients are fully investigated not only detailing their genital anatomy and its variance from normal, but any appropriate endocrine and genetic investigations must also be carried out. A large number of these congenital malformations have associated disorders of other organ systems, which also need to be delineated and defined. This may involve a number of forms of imaging depending on the most appropriate to determine the abnormality. Finally, an extremely careful assessment of the psychological status of the patient is imperative. The risks of not involving the psychologists in the management of these patients may cause long-term irreparable damage. Adolescents are very difficult to assess psychologically and intellectually as intellectual development varies from patient to patient. There is a complete discrepancy between physical maturity and intellectual maturity and whilst physical maturity may be achieved by age 15 or 16, intellectual maturity is not reached

until 18 to 20. This discordance presents a major problem for management as a number of the malformations that we will be discussing have physical symptoms which demand surgical attention at a younger age than would be ideally desirable in terms of being able to cope with the difficulties that these young women encounter. The risks of ignoring the psychological aspects of these disorders are long-term sexual dysfunction and the inability to integrate into normal society. So profound are these issues that feelings of heterosexuality may mean that these patients have difficulty sustaining relationships due to the psychological harm that has been ignored in early adolescent years. However, the psychological difficulties of adolescence make assessment much more complex than in childhood or adult life and therefore specially trained psychological counsellors are needed for all units carrying out this type of reproductive surgery.

DISORDERS OF THE VAGINA

Congenital malformations of the vagina fall into 3 categories: remnant cyst, obstructive outflow tract disorders and congenital absence of the vagina.

CONGENITAL ABSENCE OF THE UTERUS (MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME – MRKH).

Complete vaginal agenesis in association with uterine agenesis or hypoplasia is uncommon and known as the Mayer-Rokitansky-Küster-Hauser syndrome (MRKH). The incidence of this syndrome is 1 in 5000 [9]. These patients present as adolescents with primary amenorrhoea in the presence of normal secondary sexual characteristics other than menarche. Their ovarian function is normal and their karyotype is 46XX.

Genetics of MRKH

MRKH is not usually found in a familial pattern but it is found quite commonly in conjunction with other malformation syndromes [10]. Although

affected siblings have been documented with vaginal agenesis, discordant monozygotic twins have also been reported [11]. This indicates that a single autosomal recessive gene cannot be the explanation for this condition. Although Shokeir [12] proposed an autosomal dominant inheritance from a study of 16 families in Saskatchewan, subsequent studies have failed to confirm this [13,14]. It would therefore seem that the most logical explanation is a polygenic multifactorial inheritance and these inherited abnormalities normally have a recurrence risk of between 1 and 2%. In the offspring reported by Petrozza et al [14], none of the female offspring have exhibited Mullerian aplasia.

Molecular genetic studies have so far failed to attract any candidate genes to explain this phenomenon. Whilst a number of studies have looked at the possibility of defects in the homeobox series, as yet no abnormality has been found.

Diagnosis

As alluded to above, these patients present with primary amenorrhoea and investigation by imaging reveals that there are two forms of the MRKH syndrome. In the first group, the only abnormality is congenital absence of the vagina and uterus and in the second group congenital absence of the vagina and uterus is also associated with renal, ear and skeletal abnormalities [15]. With regard to skeletal abnormalities, these are well known to occur in some 10-12% of patients [16,17]. The incidence of hearing difficulties in MRKH patients has been reported by Strubbe et al [18] and a hearing loss of some degree was attributed to a congenital origin in 10% of MRKH patients. These

findings illustrate the need to evaluate MRKH patients a little more thoroughly for associated abnormalities than perhaps has been previously practised.

Management

The management of these patients falls into two distinct areas. The management of their congenital anatomical abnormality with the need to be sexually active and the psychological impact of the knowledge that these individuals have no vagina and no uterus.

Psychological aspects of Mullerian aplasia

It is difficult to quantify the emotional trauma which is associated with the knowledge that the patient has Mullerian aplasia. There are 3 parties at least involved in this process. The patient herself, who may be a young adolescent aged 14 or 15, and her parents. The shock is considerable to all and the first reference to the need for psychotherapy is cited in 1968 by Kaplan based on 9 patients with MRKH syndrome [19]. He describes the importance of the impact of a physician's lack of appreciation of the difficulties that may ensue and how this inability to communicate may make the psychological effects much worse. He further develops his hypothesis of the need for psychological input in 1970 when he describes a series of cases outlining the great difficulties that these patients have [20]. Poland and Evans [21] studied 54 patients and described the emotional reactions to the diagnosis and treatment which varied with the age of the patient and her relationship both with her parents and with a heterosexual partner. Many patients were initially depressed, questioned themselves over their gender

and their ability to fulfil the female role in the future as an adult. However, rather than the expected difficulty of sexual intercourse, it was infertility that was the most difficult part for these young women to accept. In having instituted a policy of prolonged counselling, they showed that emotional reaction and reinforcement was a vital part of the medical management of these patients but the input of psychological services was fundamental in improving outcome of therapy. In the last 10 years, there has been an evolution in the multidisciplinary approach to the management of MRKH syndrome and the importance of psychological support has been emphasised to an even greater extent [22]. The evaluation of group therapy which has been ongoing in a number of centres for some years was first reported by Weijenborg [23] and demonstrated the invaluable use of group programmes for helping MRKH syndrome patients deal with their psychologically stress. It is impossible to emphasise too greatly the importance of psychological input into the preparation of patients, not only for the adaptation to their congential abnormality and the ability to deal with this currently and in later life, but also in the preparation of the patient for whichever therapy is chosen to help her to achieve a functional vagina. Without adequate psychological assessment and the involvement of the psychological or the clinical nurse specialist in the timing of treatment, then the chances of success are vastly reduced.

Non-surgical management of Mullerian agenesis

Whilst as yet it is not possible to create a new uterus for these individuals, a number of techniques have been described for the creation of a vagina. A recent policy statement by the American College of Obstetricians

and Gynaecologists emphasises the primary role of the non-surgical approach with vaginal dilators as being the treatment of first choice [24]. The use of vaginal dilators was first reported by Amussat in 1835 [25]. Amussat's technique was to use strong digital pressure on the vaginal dimple over a series of sessions but in 1938 Frank modified this method to use pyrex tubes [26]. Frank described 6 cases in his paper, only one of which was not successful but, interestingly, for almost 40 years this technique was only infrequently used. Rock et al reported success with the use of dilators in only 40% of patients [8] and this report further reinforced the lack of enthusiasm for the use of this technique. However, organisation of the therapy at that time was poor, had little psychological input and the patients were given almost no ongoing support from clinical nurse specialists with expertise in these areas. Once this had been introduced into normal practice, a number of reports supported the idea of a non-surgical approach.

The technique involved requires passive dilatation of the vaginal dimple using graduated dilators. The dilators that are used come in a number of formats and also are made of a number of different materials. None of the particular designs is any better than the others and the results are significant in that a well-motivated, well-supported patient can achieve a vaginal length which is totally satisfactory for intercourse in 85% of cases [22] (Table 2). It is, however, important that whichever technique is used to try to create a vagina, that a proper assessment is made of the results of therapy. This has, in fact, been extremely poorly addressed over many years and apart from sporadic reports of small numbers, there has been no comparative study until recently. In a study carried out by Nadarajah et al (personal communication) we have

shown that in 60 patients followed up for up to 5 years, over 90% of the girls had a totally satisfactory sexual experience. 25% of patients complained that they either had poor lubrication or dyspareunia but this did not interfere with their enjoyment of sexual intercourse and this was extremely gratifying. We believe that this technique has now been shown to be highly successful and as stated by the American College of Obstetrics and Gynecology, is the procedure of first choice.

In an attempt to try to improve patient co-operation with this type of passive dilatation, Ingram developed a method whereby a dilator was placed through a bicycle seat stool so that the patient could lower themselves onto the dilator and use her body weight to create the pressure [27]. Ingram reported the use of his technique on 24 women of whom 50% had primary vaginal agenesis. Twenty out of the 24 patients had successful treatment and the only 4 who failed to have a successful result had had previous neovaginal surgery. This technique is identical to Frank's procedure in terms of its principle and the outcomes are equally gratifying.

Surgical management of Mullerian agenesis

In those patients who fail to achieve a functional vagina with passive dilatation, a surgical approach may be required. It is extremely important to realise that whatever techniques are used, these patients will need to use vaginal dilatation post-operatively in almost all cases. As a result of this, in just the same way as it is important to prepare the girls for passive dilatation primary treatment, the fact that they have failed primary treatment at least means that they are familiar with the vaginal dilatation technique and it is

more likely they will persevere post-operatively and have a good result. However, failure of the use of dilators sometimes brings a negative approach from the patient who hopes the surgery will be a one-off event that will then mean they do not need to use vaginal dilatation at all. It is important, therefore, that an appropriate psychological assessment is carried out to ensure the patients are ready for this type of procedure and that they also get appropriate support post-operatively.

There are a numerous number of procedures that have been described to try to create a vagina that is functional. They fall into a number of categories (Table 3).

Surgical creation of the neovaginal space

For all techniques in this group, the initial approach is identical in that a transverse incision is made at the apex of the dimple and a digital dissection of the space between the urethra and bladder anteriorly and the rectum posteriorly is carried out. It is essential that the depth of the neovaginal space reaches the peritoneum which lines the pelvic cavity as failure to do this results in excessive contracture post-operatively. A mould is then placed in the space and this mould may be lined by a number of materials. In the United Kingdom the most widely used material is amnion but in the United States it is more common to use the McIndoe Reed technique with the use of split thickness skin grafts taken from buttock. The complications of this type of surgical approach involve intra-operative damage to either the rectum, urethra or bladder with subsequent fistula formation with rates ranging from 0 to 7.6% [8,28]. A number of authors have used different mould types, both soft, semi-

rigid or rigid, and there seems to be good evidence that the use of soft moulds reduces the risk of fistula formation, presumably from lack of avascular necrosis from pressure. An alternative to the use of these moulds is the use of the inflatable soft stent which it is claimed reduces the risk of haematoma formation without comprising healing and the risk of fistula formation [28]. In the largest reported series by Alessandrescu et al [29] with the treatment of 201 patients, the surgeons used a rigid mould throughout their experience and had a fistula rate of <1% and therefore it may well be that it is not the type of mould that is important but the skill and experience of the surgeon. The functional outcome of the McIndoe type of procedure is summarised in Table 4. Although the functional outcome is reported anecdotally with success rates ranging between 80 and 100%, there has not been a specific study surveying sexual satisfaction to ensure that these results are indeed true.

The use of amnion has been described by Ashworth et al. [30] and subsequent experience of this technique has shown it to have extremely similar success rates to the McIndoe procedure. It has the advantage that no graft site is required, thereby leaving no external scars for the patient to have to tolerate. However, it is important that the use of this material is properly governed and that the donors are suitably screened for HIV and CJD.

The use of adhesion barriers has been reported, although only in small numbers of patients, but in the report of Jackson and Rosenblatt [31] the four patients treated had 100% success in creating a neovagina after six months.

The use of peritoneum to line the neovaginal space has been popularised in Russia by Davydov [32].

Bowel vaginoplasty

The use of a segment of the intestine to act as a vagina has been in use since 1892 by Sneguireff [33]. He used the rectum as a substitute for a vagina and created a colostomy and as such did not become popular. The first time that ileum was used was in 1907 when Baldwin reported the use in a patient [34] and this became known as Baldwin's procedure. The advantage of using ileum is that the calibre of the bowel remains constant and there is good lubrication but there are disadvantages as the small intestinal mucosa is very easily traumatised by intercourse and bleeding often occurs. Also, there is chronic secretion of mucous from the loop and in the end this technique has not remained popular [35]. The use of an isolated segment of sigmoid colon was first reported in 1914 by Ruge [36]. He used lower sigmoid colon and although initially a number of these procedures ended with bowel necrosis and fistulae, over the ensuing 80 years, the technique has gradually become modified and improved and is now known as a colocolpopoiesis [37]. The most recent results of the use of bowel are summarised in Table 5 where although the success rate overall is between 77 and 90%, the complication rates are not inconsiderable. This technique of using the bowel therefore is generally reserved for more difficult cases where simple vaginoplasties have failed and the post-surgical situation is one that results in excessive scarring. In these circumstances, in order to create a neovagina of some function, the whole neovaginal area needs to be excised and replaced by a loop of bowel and it is recommended that sigmoid colon is the best segment to use.

Syed et al. [38] report the outcome of 18 children who had undergone colovaginoplasty when aged between 1.5 and 8 years. They used sigmoid

colon in the hope that this might abolish the problems of the emotional difficulty of reconstructive surgery in adolescence by creating a functional vagina in childhood. Although there were no major complications in the early follow-up period, 3 patients developed severe vaginal discharge problems within 2-7 years and the histology confirmed diversion colitis. This is a reasonably serious complication and one which has made these authors suggest that childhood bowel vaginoplasty should not be continued.

Finally, the long-term sequelae of this procedure are unknown but a recent report from Hiroi et al [39] reports a mucinous adenocarcinoma arising in a neovagina using the sigmoid colon.

Neovaginoplasty using peritoneum

Although it had been described earlier, the use of peritoneum to line the neovagina was first reported in a series by Davydov et al. in 1974 [32] and has since been known as the Davydov operation. In his procedure, Davydov performs a laparotomy having created a neovagina in the normal way and then by mobilising the peritoneum from the peritoneal cavity including the rectum, he uses this to line the neovaginal cavity. The advantage of this technique as claimed by Davydov and co-workers is the lack of granulation and scar formation. However, as can be seen in Table 6 the success rate is not universally 100% although the results are remarkably good in terms of vaginal intercourse success rates.

Recently, a laparoscopic approach for the use of peritoneum has been reported by Soong et al. [40]. In this series they claim 100% success rate and

suggest that it is unnecessary for this procedure now to be performed through a laparotomy. Similar experience has been reported by other authors [41-43].

Vecchetti's operation

This procedure involves the creation of a neovagina using dilatation of the vaginal dimple with a traction device attached to the abdomen. In the conventional operation, a laparotomy is performed and a suture is passed through the perineal membrane from above. This suture is threaded through a plastic olive and the suture is then passed back through the vault and up through the abdomen. The abdomen is then closed, the ligature attached then to a traction device which is strapped to the patient's abdomen. Traction on the suture is then increased on a daily basis to pull the olive into the neovagina and stretching the vaginal skin to create a vagina. This mimics the technique of Frank but does not rely on the woman herself to use the dilators. After 7-9 days Vecchetti claims to have a vagina of some 10-12 cm in length and at that stage he introduces the use of vaginal dilators to ensure that the skin remains stretched. In Vecchetti's personal series of 522 procedures he claims 100% success rate and only 9 complications, which included one rectal and one bladder fistula. Table 7 outlines the conventional Vecchetti technique and its results.

Recently, a number of series have reported a laparoscopic approach to this procedure, thereby avoiding the laparotomy to insert the suture. The rest of the procedure is as in the conventional technique and the results are seen in Table 8.

Vaginoplasty using skin flaps

The first reported use of skin flaps from the labia minora was in 1921 by Graves [44] and Frank and Geist in 1927 suggested that a tube graft from the inner aspect of the thigh could be used for vaginoplasty [45]. The principle behind using skin flaps is the advantage of full thickness grafting. The disadvantage of the McIndoe technique which uses a split thickness graft technique is the problem of contraction of the vagina post-operatively and in the full thickness grafts this ought to be avoided. Over the ensuing years a number of procedures have been suggested using gracilis myocutaneous flaps [46] and rectus abdominis myocutaneous flaps [47]. Wee and Joseph in 1989 described a technique of a pudendal thigh flap vaginoplasty [48] and in the follow up study of 12 patients [49] they found this technique to be extremely useful particularly in patients who had vulva anomalies secondary to congenital adrenal hyperplasia. A further report of using pudendal thigh flaps in 8 patients with MRKH syndrome showed this to be successful in 100% of cases in achieving a functional vagina [50]. However, the disadvantage of this technique and any technique which uses hair-bearing skin as the donor site inevitably means that there is some hair growth in the created vagina and this can be a problem, both in terms of dyspareunia and discharge.

The use of a free flap graft from the scapula was first reported by Johnson et al. in 1991 [51]. This technique was carried out on 3 women and although they achieved a good functional length vagina at the end of the procedure, the surgical undertaking was enormous and this technique has not subsequently become popular. Giraldo et al. in 1994 reported the use of a

vulvoperineal fasciocutaneous flap (the Malaga flap) in the treatment of MRKH [52]. They report the results of 6 cases, the results of which are very encouraging.

Finally, the use of tissue expanders to create excessive vulval skin to act as the donor site was first reported by Lilford et al. in 1988 [53] and they reported their experience of 17 cases subsequently [54]. They report vaginal length was satisfactory in 16 of the 17 women although again they had 2 patients who complained of vaginal discharge and a number of patients who subsequently complained of hair in the vagina causing dyspareunia. Serra et al. [55] reported their experience in 1993 of 6 patients with 100% success with only one patient complaining of dyspareunia and Belloli et al. [56] reported their experience in 1997 of 2 patients with a successful result. Whilst this only constitutes 25 patients altogether the results in creating a vagina seem to be very good. However, the complications of hair-bearing skin makes this more difficult although Belloli's use of labial skin may have some promise.

Williams' vulvoplasty

A vulvovaginoplasty was described by Williams in 1964 which involved the creation of a vulval pouch using the labia majora and minora [57] but as this procedure creates an abnormal angle for intercourse and may in fact create dyspareunia in the long-term, this procedure does not have a role in routine vaginoplasty. In those patients in whom a neovagina cannot be created, this may provide some form of pouch into which intercourse can occur and can be considered in these rare circumstances.

MRKH AND SURROGACY

In MRKH syndrome the ovarian function is normal. Therefore, the possibility of IVF surrogacy is one that has come to fruition in terms of success. In studies initially looking at the ovarian response to gonadotrophins, the studies showed that there was an absolutely normal response rate in these patients in IVF programmes [58,59]. Petrozza et al. in 1997 [14] reported on 162 IVF cycles for MRKH syndrome patients with 34 live children born and the fact that no congenital abnormalities were found in these offspring was very encouraging and strongly suggests that congenital absence of the vagina is not inherited in a dominant fashion. Similarly, Beski et al. reported their experience with similar outcome figures and no congenital abnormalities [60] and the experience of Goldfarb et al. [61] in their surrogacy programme gave equally encouraging results. These encouraging reports should encourage clinicians to consider IVF surrogacy as an option for MRKH patients who wish to attempt pregnancy.

UTERINE ANOMALIES

The only uterine anomaly in the menstruating teenager that may be a problem are rudimentary horns. These cause increasingly severe dysmenorrhoea and in all teenagers who fail to respond to the normal medication for primary dysmenorrhoea, an ultrasound scan should be performed to identify the presence of a rudimentary horn. Occasionally, these may be communicating in which case no haematometra will be seen but if they are non-communicating a haematometra is apparent and retrograde

menstruation may well lead to a haematosalpinx and/or an endometrioma. Treatment of this problem is excision of the rudimentary horn and reconstruction of the uterus and reproductive performance in these women is normal if the horn is non-communicating. If they have a communicating horn, care must be taken at the time of surgery to reconstruct the uterine cavity to give it sufficient strength to withstand the physiological changes of pregnancy and a decision should be made as to whether or not delivery should be by elective Caesarean section.

Occasionally, in the MRKH syndrome the uterine anlage may have functional endometrium and here excision of the anlage resolves the problem.