COMMENTARY

Congenital abnormalities of the genital tract: Management or mismanagement?

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Summary

Congenital malformations of the genital tract are uncommon and their management requires skill that is only available in a very few centres worldwide. The management requires sensitivity both in terms of the psychological welfare of the patient and the physical welfare and these must be combined together to provide such support. Management of patients outside of these environments is less than optimal and this article seeks to exemplify a principle of management excellence.

Introduction

Gynaecologists dealing with patients with congenital abnormalities are charged with three fundamental responsibilities.

1. For those who have pain, the relief of that pain is imperative. In obstructive outflow disorders, this pain can often be very acute and debilitating.
2. The restoration of reproductive potential if possible.
3. The management of the psychological sequelleae that arise in patients who have this type of problem.

Congenital abnormalities of the genital tract are uncommon. Congenital absence of the uterus and vagina (Meyer-Rokitansky-Koster-Hauser Syndrome – MRKH syndrome) has an incidence of approximately 1 in 5,000 female births based on the best demographic data that is available (Aittomaki et al. 2001). Obstructive outflow disorders with a septum transversely across the vagina has an incidence of only 1 in 20 – 70,000 female births (Mattingly and Thompson 1985). Congenital absence of the cervix is so rare that only case reports and small series exist in the world literature with numbers of reported cases being in the region of 80 in total (Edmonds 1999). It is instantly apparent that these congenital abnormalities are so rare that the management of them requires a multidisciplinary approach. It is inappropriate for their management, whether surgical, medical or psychological to be delivered outside specialist centres.

Presentation

The clinical assessment of any adolescent who presents with either primary amenorrhoea or secondary amenorrhoea can be carried out by any gynaecologist with clinical acumen. History and examination are extremely important as they are in all clinical circumstances. Investigation should be appropriate; for example, ultrasound scanning of the pelvis for congenital abnormalities is appropriate so long as the operator is skilled at imaging congenital abnormalities. As we have said already, these abnormalities are rare and therefore there are very few individuals who have this skill. Sadly, as a result of this lack of experience, ultrasound findings can be very misleading and reports written by inexperienced individuals may completely mislead the clinician in terms of management. An example of this would be an adolescent with primary amenorrhoea presenting with a pelvic mass. All too often, this cystic area is diagnosed as being an ovarian cyst and adolescent girls may be subjected to laparotomy on these ultrasound findings only to discover that there is no ovarian cyst but a haematocolpos. The surgeon is often ill-equipped to deal with that problem and the patient has then been subjected to an unnecessary laparotomy. Imaging by MRI in centres where there are no experienced ultrasonographers is a more reliable form of investigation but gives no more information that an accurate ultrasound. In circumstances where a diagnosis is expected to be unusual, clinicians are recommended to attend the ultrasound and guide the ultrasonographer over thought processes around congenital abnormalities. This way the combined experience of a gynaecologist and an ultrasonographer can be brought together to reach the correct diagnosis. To illustrate the range of abnormalities which should be referred to centres with a specialist interest in this area, we will look at several examples of congenital abnormalities.

Imperforate hymen

Teenage girls may well present with primary amenorrhoea and dysmenorrhoea and in these circumstances clinical
examination is all that is required to make the diagnosis. Abdominal palpation may reveal a mass arising from the pelvis if the haematocolpos is large enough. Examination of the vulva will reveal a bulging membrane that is obviously blue in colour. This blue colouration reflects the fact that the hymen is very thin and behind it is menstrual blood that is black in colour and gives the hymen the appearance of being blue. There is no need in these circumstances for any imaging to make the diagnosis and management of this condition is extremely straightforward. A cruciate incision can be made in the hymen and the haematocolpos drained. This procedure may be carried out by any competent gynaecologist knowing that the diagnosis is correct. These patients do not need referral to a specialist centre.

Transverse vaginal septum

Patients may present in an identical way to the imperforate hymen when there is a transverse vaginal septum preventing menstrual loss. These septae may be at any level within the vagina although the most common are in the lower-third. Here, as in the imperforate hymen, menstrual blood is unable to escape and as a result of this over time, there is increasing cyclical abdominal pain. The vagina has an enormous capacity to contain blood and up to 2.5 or 3 l of menstrual blood have been seen on occasions. Abdominal palpation will again reveal a mass arising from the pelvis in circumstances where menstrual obstruction has been sufficient but examination of the vulva and vagina will reveal a blind-ending vagina and, significantly different from the imperforate hymen, the appearances of the septum are clearly pink. This is because the septum is considerably thicker than an imperforate hymen and therefore there is no transillumination from the menstrual blood behind. The septae that occur within the vagina are of variable thickness but the aetiology of the obstruction is completely different from the imperforate hymen. Here, the septum has to be removed in its entirety and an end-to-end anastomosis performed of the two vaginal margins. The maintenance of the vaginal passage during the healing period is crucial to the long-term functional capacity of the vagina. Failure to achieve this will mean that the patient will present later with a vaginal stenosis. Problems of vaginal stenosis are enormous and reconstructive surgery in this area is extremely difficult. The primary results in successful first-time surgery are excellent if the appropriate post-operative care is given. This requires the use of vaginal dilators and this care needs to be very carefully supervised by an individual who is trained and experienced in this type of therapy. There are psychological problems to be overcome in the early teenage years if this is to be achieved and sadly all too often surgeons do not have this type of multidisciplinary approach. Surgery done in inappropriate circumstances will result in long-term damage to sexual function, a problem that is completely avoidable when the patients are operated on in specialist centres.

Congenital absence of the cervix

This extremely rare abnormality arises when the lower end of the uterus fails to develop. Vaginal agenesis may also be present and the presenting symptoms are of dysmenorrhoea and primary amenorrhoea. Occasionally, a haematometra may occur although retrograde menstruation tends to avoid this. Until the mid-1980s and still in most countries worldwide, the treatment of this condition was hysterectomy. Rock et al. (1984) and Edmonds (2003) have shown that in some cases, reconstructive surgery can be carried out with the salvage of the uterus for reproductive purposes and isolated pregnancies have been reported. Again, this type of surgery should not be carried out outside of very specialist centres with an expertise in the management of this condition.

Uterine anomalies

Dysmenorrhoea is a difficult symptom in adolescents. It is often associated with no congenital abnormalities whatsoever and management advice is around the use of non-steroidal anti-inflammatoryatories or the oral contraceptive pill, which will indeed resolve the problem for the majority of teenagers. Any teenager who fails to respond to this type of therapy should have imaging of her pelvis. Congenital abnormalities of the uterus, which may range from arcuate uteri through unicorneate, bicornuate, uterus didelphys and obstructed uterine horns can only be diagnosed on
imaging. Ultrasound is more than capable of making this diagnosis although many patients are subjected to an MRI as the expertise is not necessarily locally available. This is completely appropriate. The management of congenital abnormalities of the uterus again requires expert knowledge. Many of these women are quite understandably concerned about their reproductive function and need to be appropriately and accurately informed as to the difficulties that pregnancy and childbirth might bring. Patients who have obstructed uterine horns need to have the horn surgically removed and the uterus reconstructed. Again, successful surgery will mean that the uterus will remain intact and the patient able to tolerate pregnancy and labour without difficulty. Reconstruction of the uterus should again be carried out by surgeons with an expertise in this area. Failure to apply the appropriate surgical technique will leave the uterine wall defective and the risks associated with pregnancy will be apparent.

**Congenital absence of the uterus (MRKH) syndrome**

Congenital absence of the uterus is the second most common cause of primary amenorrhoea. The impact of learning that you have been born without a uterus or a vagina is considerable. The sequelae from a psychological point of view are devastating not only on body image and personal demeanour but also the impact of not being able to fulfil your biological potential of reproduction. The vast majority of these patients do not require a surgical approach in order to achieve a functional vagina and vaginal dilator therapy is extremely successful. However, success in terms of physically being able to have intercourse is only a part of the care that these patients require. No other congenital abnormality requires such a multidisciplinary approach as does this condition. Here, the support of psychologists, gynaecologists, self-help groups, clinical nurse specialists and nurses are all mandatory parts of the management of the patient and without these success is extremely limited. Recently, the results of this multidisciplinary approach were published by Nadarajah et al. (2005) where results are presented over the long term. This again demonstrates the success that can be brought about from specialist centres and why isolated management of these conditions is not in the best interest of patients.

**Conclusion**

Congenital abnormalities of the genital tract are rare. Gynaecologists are likely to see one case every 10 years during their consultant years in practice. This type of incidence does not allow them to gain expertise in managing these patients, either medically, surgically or psychologically. Specialist expertise is available at the National Centre and patients must be referred to specialist centres if their outcome is going to be in each individual patient’s best interests. In patients with MRKH, the long-term needs also need to be addressed and these needs can only be provided by centres with long-term expertise. The complications that can arise from inappropriate management are all avoidable and gynaecologists should be encouraged to deal with those problems for which they have the expertise and to refer those cases that are appropriate. We owe this duty of care to our patients and to their long-term reproductive and sexual life.

**References**


