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Common congenital anomalies of the female genital tract

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Abstract

The impact of common congenital anomalies of the female genital tract is hugely variable. Some anomalies are asymptomatic chance findings requiring no intervention. Others have a major impact on the potential for sexual activity and fertility. A good knowledge of basic embryology is important to understand the pathogenesis and clinical features of these anomalies. All gynaecologists should be aware of these conditions and possible clinical presentations. Whilst some conditions, such as imperforate hymen require a simple surgical intervention, other more complex anomalies need careful assessment and accurate pre-operative assessment to optimise the long-term outcomes. The contribution of uterine anomalies to subfertility is poorly understood and the role of uterine surgery needs further research.

Keywords: Genital anomaly; Müllerian anomaly; Imperforate hymen; Vaginal agenesis; Rokitansky syndrome; Vaginal septum; Uterine septum

1. Introduction

Congenital anomalies of the female genital tract are relatively common. Many are asymptomatic, found by chance and require no intervention. However, other anomalies may have a devastating impact on the potential for sexual activity and fertility. Anomalies may be isolated to the reproductive tract or may be part of a complex syndrome affecting other organs within the body. Some patients may be diagnosed at birth or during childhood whilst others may not present until later in adult life. A careful and detailed assessment is needed to reach the appropriate diagnosis and ensure treatment maximises future potential for sexual function and reproduction.

2. Embryology

It is important to have a good grasp of the embryology of the female genital tract to understand the nature of congenital anomalies and the symptoms which may ensue.

Primordial gonads (genital ridges) appear during the sixth week of embryogenesis. These develop in association with the mullerian and wollfian ducts. The fate of these ducts depends upon the genetic sex of the embryo. In XY embryos, the SRY gene (sex determining region of the Y chromosome) stimulates testicular differentiation. The developing testes produce androgens and anti-mullerian hormone (AMH), which cause virilisation and regression of mullerian structures. In XX individuals, absence of the SRY gene allows the gonads to develop into ovaries whilst the subsequent lack of AMH allows the mullerian ducts to develop into a uterus, fallopian tubes and vagina.

The mullerian ducts grow in a caudal and medial direction and fuse in the midline to form the primitive uterus. These ducts form the right and left fallopian tubes and midline fusion of these structures produces the uterus, cervix and proximal two-thirds of the vagina. This rudimentary vagina fuses with the posterior urethra at week 7 to form the urogenital sinus. Fusion of the mullerian ducts also brings together the lateral peritoneal folds that form the broad ligaments. The vagina develops from a combination of the mullerian tubercles and the urogenital sinus. Cells proliferate from the upper portion of the urogenital sinus to

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form structures called the sinovaginal bulbs. These fuse to form the vaginal plate which extends from the mullerian ducts to the urogenital sinus. This plate begins to canalise, starting at the hymen and proceeds upwards to the cervix. This process is not complete until 21 weeks of gestation.

The external genitalia in females consist of the genital tubercle, the urogenital sinus and the urethral and labioscrotal folds. The genital tubercle becomes the clitoris and the urethral folds develop into the labia minora, and the labioscrotal folds become the labia majora. By week 12 of development these structures are recognisably female. These structures are sensitive to androgens and form the penis and scrotum in normal male development. Defects in development can occur at any of the stages described above.

3. Vaginal anomalies

3.1. Abnormal appearance

3.1.1. Polyps

Hymenal polyps and tags are not uncommon after birth. They are usually very small – less than 5 mm – and are probably due to oestrogen stimulation. Most resolve spontaneously and they are rarely seen after the age of 3 years. Very rarely they can persist and become more polypoid. In this case, they can cause an inflammatory and sometimes bloody discharge and in this situation will need excision.

3.2. Vaginal obstruction

3.2.1. Imperforate hymen

An imperforate hymen can occasionally be detected on prenatal ultrasound scan or present at birth if associated with a hydrocolpos. A pelvic mass may be palpable clinically or visible on ultrasound. In this case, excision of the hymen is needed in the neonatal period. A cruciform incision is made in the bulging hymen and the redundant skin excised. Occasionally pre-pubertal girls can be referred with no obvious hymenal opening. In most cases, the hymen is likely to have a poorly visible microperforation. Even if the hymen is not perforate, in the absence of a collection, definitive surgery should be deferred until the child is peripubertal and the hymen oestrogenised but before build up of a haematocolpos.

Most commonly, an imperforate hymen causes menstrual obstruction and prevents the escape of menstrual flow. This will cause worsening cyclical pain in the absence of menstruation and a pelvic mass due to a haematocolpos. The obstruction usually occurs at the junction of the lower third of the vagina, at the level of the hymen. It is sometimes possible to visualise a bulging, blueish hymenal membrane, once the labia are gently parted. An ultrasound scan will confirm the diagnosis. A simple resection of the obstructing membrane will release the menstrual flow, and allow further normal menstruation.

It is essential that imperforate hymen is distinguished from transverse vaginal septum and cervical agenesis. Both of these condition are less common but can present with very similar symptoms of cyclical pain and amenorrhoea. However, the surgical approach and long-term outcomes are very different and it is essential that surgical reconstruction is not attempted without being aware of the correct diagnosis.

3.2.2. Transverse vaginal septum

Less commonly, vaginal obstruction may be due to a transverse vaginal septum. It is essential to differentiate this from an imperforate hymen. This may be an isolated anomaly although has been described as part of McKusick–Kaufman syndrome [1]. Presentation will be as for an imperforate hymen with cyclical pelvic pain and a pelvic mass in the absence of menstruation. However, on parting the labia the typical bulging membrane is not seen and the vaginal introitus usually looks normal. The diagnosis can be suggested on ultrasound scan although an MRI scan is usually necessary to determine accurately the width of the septum and the amount of normal vagina above it. If the septum is thin and low it may be possible to resect from below although care must be taken to ensure the whole septum is removed to prevent contracture. If the septum is thicker and higher in the vagina, a combined abdominal and perineal approach is needed. It may be possible to remove the septum and reanastamose the proximal and distal vaginal segments. However, if in some cases the distance is too great and the gap must be bridged with a skin graft or section of bowel.

3.2.3. Longitudinal vaginal septae

Most women with a longitudinal vaginal septum and double vagina will be asymptomatic. A few will report difficulty with inserting tampons or dyspareunia, in which case surgical excision of the septum may be indicated [2]. Occasionally, patients present with a double uterus and unilateral haematocolpos secondary to an obstructed hemivagina. Such patients characteristically have ipsilateral renal agenesis. In this situation, excision of the vaginal septum is the preferred surgical option.

3.3. Absent vagina

3.3.1. Labial adhesions

Labial adhesions are acquired and not congenital. However, they can be mistaken for an absent vagina causing significant distress for families until the correct diagnosis is reached. It is very important to distinguish common labial adhesions from a congenital anomaly. Labial adhesions or fusion is estimated to occur in up to 3.3% of pre-pubertal girls [3]. The peak incidence is in the first year of life and it is never present at birth. The appearance is typical with fusion of the labial skin extending from the posterior fourchette towards the urethral opening. There is a clearly visible thin membranous line in the midline where the tissues fuse. The urethra may be just a pinhole opening in