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Distal Vaginal Atresia Misdiagnosed as Imperforate Hymen: A Case Managed By Transperineal Vaginal Pull Through

Dr. Kothapally lahari[1], Dr. yannam satya Shilpa[1], Dr. L. Jayanthi reddy[1], Dr. P. Sudhir babu[1]
[1]Department of Obstetrics and Gynaecology, S.V.S Medical College and Hospital, Mahabubnagar, Telangana

Corresponding Author

Dr. P. Sudhir babu

Department of Obstetrics and Gynaecology, S.V.S Medical College and Hospital, Mahabubnagar, Telangana

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Abstract

Three different types of vaginal outlet obstruction are described in the literature: imperforate, transversal vaginal septum and vaginal atresia. Diagnoses were made in different ages of life, from neonatal to the teenage years. Clinical and ultrasound examinations and MRI dominated in establishing the diagnosis. Complex malformations of the female genital tract are not so common. Their correct identification is of paramount importance for appropriate management. A thorough knowledge of embryology, pre-operative imaging with MRI and examination under anaesthesia is essential to identify accurately the constellation of anomalies and to plan appropriate management. This case reports distal vaginal agenesis in an 12 year old girl which was managed by dissecting the lower half of vagina and pull-through vaginoplasty. Rarity and variable presentation of congenital genital tract anomalies can lead to delayed diagnosis and erroneous management. A high index of suspicion and cross-sectional imaging can help in early diagnosis. A comprehensive management is imperative to preserve the reproductive potentials, as significant proportion of patients may experience sexual difficulties, menstrual irregularity, and infertility.

Key words: Imperforate hymen; haematocolpos; mullerian anomalies; vaginal atresia

INTRODUCTION

The vaginal outlet mechanism consists of undisturbed fluid passage from vagina through hymenal opening. The genital tract outflow is important for secretion and menstrual effluxion and as a pathway in reproductive function. Congenital outflow obstruction may occur at different levels and with different clinical presentations (1).

Embryological development of vagina results from lower portion paramesonephric ducts fusion and regression forming the uterovaginal primordium (gives rise to the uterus and superior part of vagina). Contact of the uterovaginal primordium with urogenital sinus induces formation of paired outgrowths named sinovaginal bulbs. The sinovaginal bulbs fuse to form the vaginal plate. The cells of the fused bulbs undergo apoptosis to form the lumen of the vagina. Until late foetal life the lumen of the vagina is separated from the cavity of the urogenital sinus by a membrane – the hymen (2,3).

Abnormal development can result in any of the following three: imperforate hymen (failure of epithelial degeneration), low, mid, or high transverse septum of the vagina (incomplete unit), or atresia of the vagina resulting in persisting of a portion of solid cells cord. Recently interest has focused on expression and function of the mammalian HOX genes as a possible aetiology of these genital developmental abnormalities (4).

Despite different origins of some parts of the vagina and hymen, their obstructed forms are clinically manifested as hydro (metro) colpos in the neonatal period and haemato (metro) colpos at the beginning of puberty.

Vagina originates from two embryonic structures: the upper part from Mullerian duct system and the lower third from the urogenital sinus. Canalization of the vaginal canal is complete by the 20th week. Misdevelopment presented as failure of fusion or canalization of these two systems in vertical plane may be clinically present with a spectrum of Mullerian duct anomalies. Vaginal atresia (VA) is one of them. Missing portion of the vagina is replaced with fibrous tissue (5). According to the American Society for Reproductive Medicine 1998 Classification, vaginal atresia is categorized as Type I (6). Some authors believe that transverse vaginal septum is partial vaginal aplasia (5).

Clinical findings vary depending on the anatomy of the vaginal outlet and the changes in the upper vagina and uterus. The upper vagina becomes enormously distended when the girl starts to menstruate, usually producing a palpable abdominal mass arising from the pelvis. The Fallopian tubes can be normal, although they may be distended allowing escape of the fluid into the peritoneum. Other anomalies are occasionally seen with hydrometrocolpos. Some other combinations of structural anomalies may be present, such as Mayer–Rokitansky–Ku¨ster–Hauser syndrome, Bardet–Biedl syndrome, Fraser syndrome, and Winter syndrome (7–9).

The diagnosis is usually made when symptoms of obstruction are obvious. Ultrasound, abdominal and endorectal (10), and other imaging studies confirm the physical examination. Urogram would demonstrate anterior and superior displacement of bladder and, possibly, hydronephrosis and hydroureter. As Mullerian agenesis can be associated with other anomalies, particularly those of kidneys and skeleton (11,12), further investigations are indicated.
Therapy is directed to relieve the obstruction of the vaginal outlet and provide normal sexual life and reproductive function. Regarding the pathological problem, cultural and religious considerations, it is necessary to inform the parents about the risk of defloration. Vaginal reconstruction is required, sometimes by an abdominoperineal approach, but stenosis and fistula formation could complicate postoperative period.

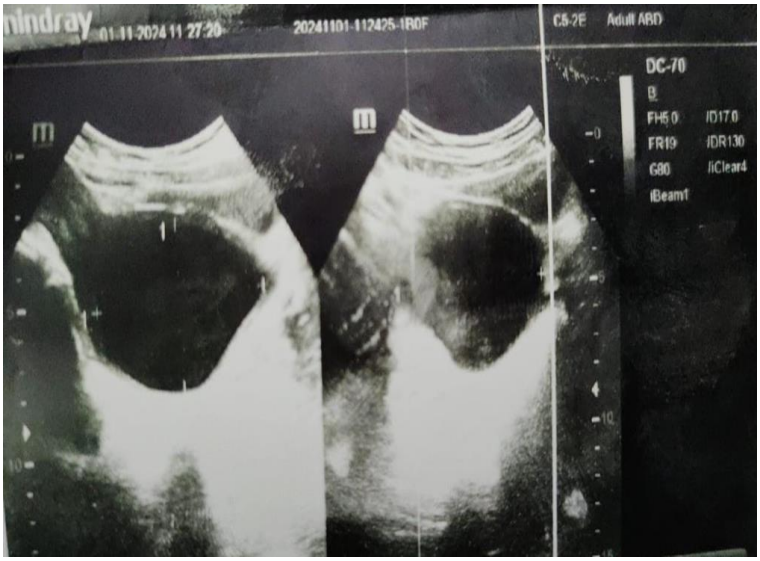

Fig 1 : Transabdominal ultrasound revealing haematocolpos.

CASE REPORT
A 12 year old girl presented with amenorrhea since 2 years and cyclical pain in the abdomen since 2 years every month for the duration of 3days.
Mentrual history- she attained menarche 2years back (Age of menarche-10YEARS) during which there was only spotting per vagina of 2-3 drops per day for the duration of 2 days followed by that there was no spotting or bleeding per vagina until the day of admission. She was not married.
On examination, she was 151 cm in height with secondary sexual characters corresponding to age , abdomen was soft and no lump was palpable.Vagina was replaced by a small 2 dimples. On rectal examination which is highly essential and important, a tense cystic mass suggestive of haematocolpos was felt anteriorly about 3 cm above the anus opening which with no bulging membrane was suspicious of segmental vaginal atresia. Pelvic ultrasound (Fig. 1) illustrated a haematocolpos of 7 cm size but it was difficult to assess the level of obstruction whether imperforate hymen or distal vaginal aplasia; however, MRI pelvis suggested normal uterus with haematocolpos,with its base about 5 cm from anus or introitus suggesting absent segment or segmental vaginal atresia (Fig. 2). No renal tract abnormality was found.

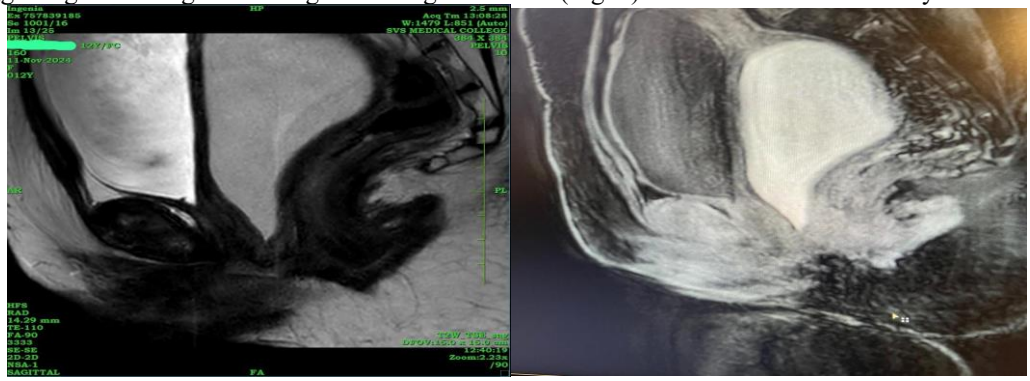
Fig 2 (a) MRI showing fluid collection in the uterus and the proximal vagina with no continuation of vagina down to perineum.

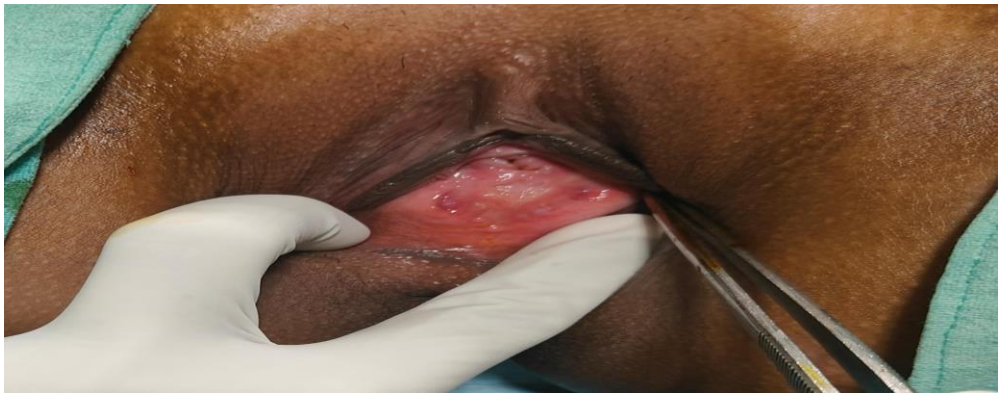

Fig 3: operative view showing no bulging or bluish discoloration and obstruction behind

A transverse incision was given between the 2 vaginal dimples (Fig.3) and dissection was done carefully for approximately 5 cm to reach the tense bulge of the haematocolpos. An incision was given on the bulge and chocolate coloured blood was drained. On inspecting the upper vagina, no abnormality was detected.
Mucosal margins of the upper vagina were pulled down till the introitus and sutured there with intermittent sutures (Fig.4.a). vaginal mould was inserted (Fig.4.b) The family was informed that there could be bleeding on defloration or not.

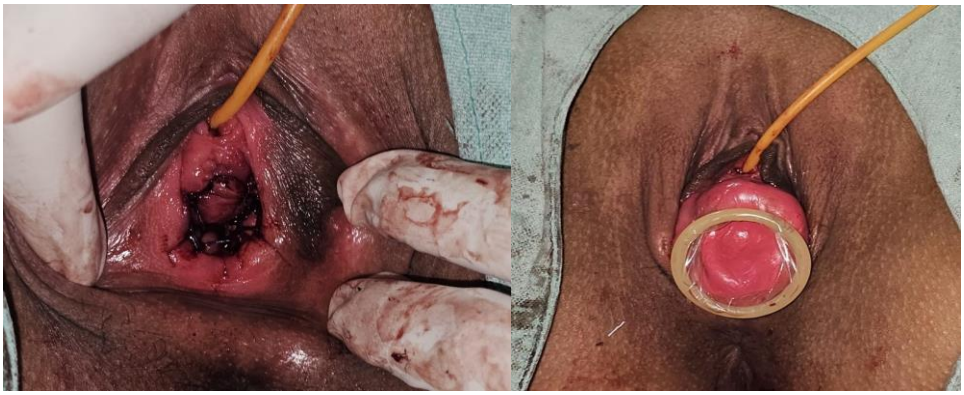

Fig.4: (a)Operative view at the end of operation.
(b)Vaginal mould was inserted.

DISCUSSION

Female reproductive tract abnormalities are generally encountered in 2–3% of women (13). Various combinations have been described in the literature. Imperforate hymen is the most common defect. Incidence of isolated vaginal agenesis is 1:5000 women and that of uterus didelphys in fertile women is 0.16%, but the combination of the two is not known(14,15). However, incidence of Mullerian anomalies is higher in population with impaired fertility, with 8% having uterus didelphys (16,17).
Uterus and vagina are formed embryologically by the dynamic process of differentiation, migration, fusion and canalization. A wide variety of abnormalities of uterus and vagina are caused by the disruption of one or more of the above processes. Selective agenesis of lower vagina, segmental agenesis and vaginal atresia is a separate entity and is usually associated with normal Mu¨llerian development. Although incidence of Mu¨llerian abnormalities is not low and cases of isolated defects of various types have been reported previously but the combination of anomalies as in present patient is unique. This suggests the need of careful pre-operative evaluation of the anomalies and individualization of the therapeutic approach to restore functional anatomy and reproductive potential (18,19).
The present case depicts an unusual case with a common presentation and yet another different mode of treatment. It can be a technical challenge to restore the function and anatomy of that anomaly, and imaging modalities such as ultrasonography and MRI can be of great help in making a surgical blueprint.

CONCLUSION

The genital tract's expulsion function is important especially from the beginning of puberty and menstruation. The outflow obstruction may occur at different levels with variations in clinical presentation. Examination of genitalia is still essential. Suspicion of absence of vaginal outlet can arise during this simple evaluation which may indicate further investigation. Ultrasound is essential in diagnostics but in some cases MRI is necessary prior to surgery. Surgery of vaginal outlet obstruction depends on local anatomy. Simple incision/excision or vaginal reconstruction can be performed. Postoperative follow-up is necessary until the normal menstrual cycle is established. The complex combination of various Mullerian

anomalies can be a challenge to the gynaecologist and there may be a difficulty in diagnosing the type of malformations. A thorough knowledge of embryology, pre-operative imaging with MRI and examination under anaesthesia is essential to identify accurately the constellation of anomalies and to plan appropriate management.

Conflict of interests, grant support and financial disclosures
None.

Ethical approval
Written informed consent was obtained from the patients for publication of this case report and accompanying images.

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