ORIGINAL ARTICLE

Congenital female lower genital tract abnormailities: two years experience in a teritiary care hospital

Erum Khalid, Sadia Jalil, Tauqir Anwar, Sidrah Nausheen

Abstract

Objective: To report different types of female lower genital tract malformations, their presentations and management, observed in a tertiary care hospital.

Study design: Case series.

Setting and duration: All cases with congenital lower genital tract abnormalities presented and managed at Hamdard University Hospital, Taj Medical Complex Karachi, from January 2009 to December 2010.

Methodology: All cases presented in Gynae clinic of Hamdard University Hospital, Taj Medical Complex Karachi, in the above mentioned period were included.

Results: Total seven cases presented in Gynae clinic with varied presentations. Their age group ranges from 13 years to 38 years. There were two cases of Imperforate Hymen, presented with hematocolpos. One of them had isolated imperforated hymen and one had imperforated hymen with transverse vaginal septum. They were treated with, cruciate incision in hymen and excision of septum. Three cases of transverse vaginal septum identified and among them, two were in lower and one was in mid vagina. These patients presented with dysparunea, inability to consummate and infertility. Excision of septum was the treatment in all three cases. There was one case of vaginal atresia and absent cervix, with recurrent massive hematometra; in this case Hysterectomy with vaginoplasty was performed. There was one case of uterine didelyphis bicollis and hemivagina, presented with purulent discharge and primary infertility, two hemivagina were communicated with each other near cervices but out flow of one hemivagina was patent and obstructed in another. Complete excision of longitudinal septum performed and later she conceived and delivered vaginally. There was a case of Mayer-von Rokitansky-Küster-Hauser's Syndrome (MRKH), presented with primary amenorrhea and primary infertility. Out of 7, three patients were lost to follow up and in the rest there was no further complaint of menstrual irregularity, dysparunea or infertility. Two patients conceived and delivered vaginally. None of our patients had any associated anomaly.

Conclusion: 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.

Keywords: Vagina, Hematocolpos, Hematometra, Imperforate hymen, vaginal septum, urogenital sinus, Cloacal anomalies, Müllerian anomalies

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Introduction:

Any defect in the normal organogenesis involving the urogenital sinus or the Müllerian duct can result in genital tract anomalies. Embryologically; the lower two thirds of the vagina develop from the urogenital sinus. The upper vagina, cervix, uterus and fallopian tubes form from the Müllerian duct system. Failure of vertical fusion

or canalization of the two systems in utero may result in cervical stenosis or atresia, vaginal atresia, or transverse vaginal septa. In addition, hymenal tissue may be imperforate.

There are two kinds of vaginal septa, transverse and longitudinal. A transverse vaginal septum is one of the most common congenital anomalies of the female genital tract. It is thought to be the result of faulty canalization of the embryonic vagina. Transverse septa may be complete, resulting in cryptomenorrhoea and haematocolpometra, or partial, with pinpoint openings allowing menstrual flow. They are usually found in the midvagina, but may occur at any level. When the septum is in the upper vagina it is more likely to be incomplete. If located in the lower part of the vagina it is more likely to be complete.

A longitudinal vaginal septum occurs when the distal ends of the Müllerian ducts fail to fuse properly. A fibrous septum lined with epithelium divides the vagina, and the uterus may be bicornuate, with one or two cervices. Division of the septum should be undertaken if there is dysparunea, if there is obstruction of drainage from one half of the vagina, or if the septum would interfere with vaginal delivery.

The true incidence of these anomalies is unknown but reported between 0.1 and 3.8%[1,2,3]. The incidence of Müllerian agenesis is about one in 4500 women; 30-36% of such malformations are associated with other anomalies as well, particularly anomalies of the kidneys and skeleton[7].

Age at presentation and choice of primary physician may vary as patients present in infancy, adolescent, or as an adult[2,4,5]. This can lead to Incorrect diagnosis and the treatment can be erroneous, and at times disastrous[6].

This study report on varied presentation, diagnostic difficulties, and outcome of 7 cases presented to the Hamdard University Hospital Karachi, in a period of two years.

Patients and methods:

All female patients with congenital genital tract abnormalities, presented in Gynae clinic, in last two years (2009-2010) were reviewed. Diagnosis of isolated adrenogenital syndrome and abnormalities of hypothalamus-pituitary axis were ruled out. Data with respect to age at presentation, duration of symptoms, clinical presentations, radiological findings (ultrasound and MRI), diagnosis, surgical procedures, and outcome were retrieved for analysis.

Result:

Total seven presented in Gynae clinic with varied presentations. Their age group ranges from 13 years to 38 years. There were two cases of Imperforate Hymen, presented with hematocolpos. One of them had isolated imperforated hymen and one had imperforated hymen with transverse vaginal septum. They were treated with cruciate incision in hymen and excision of septum. Three cases of transverse vaginal septum identified and among them, two were in lower and one was in mid vagina. These patients presented with dysparunea, inability to consummate and infertility. Excision of septum was the treatment in all three cases. There was one case of vaginal atresia and absent cervix, with recurrent massive hematometra; in this case Hysterectomy with vaginoplasty was performed. There was one case of uterine didelyphis bicollis and hemivagina, presented with purulent discharge and primary infertility, two hemivagina were communicated with each other but out flow of one hemivagina was patent and obstructed in another. Excision of longitudinal septum performed and later she conceived and delivered vaginally. There was case of Mayer-von Rokitansky-Küster-Hauser's Syndrome (MRKH), presented with primary amenorrhea and primary infertility. On follow up, out of 7, three patients were lost to follow up and in the rest there was no further complaint of menstrual irregularity, dysparunea or infertility. Two patients conceived and delivered vaginally. None of our patients had any associated anomaly.

Case 1

Miss A, 13 years old girl presented with cyclical lower abdominal pain and spotting per vaginum. Her menarchy started three months ago.

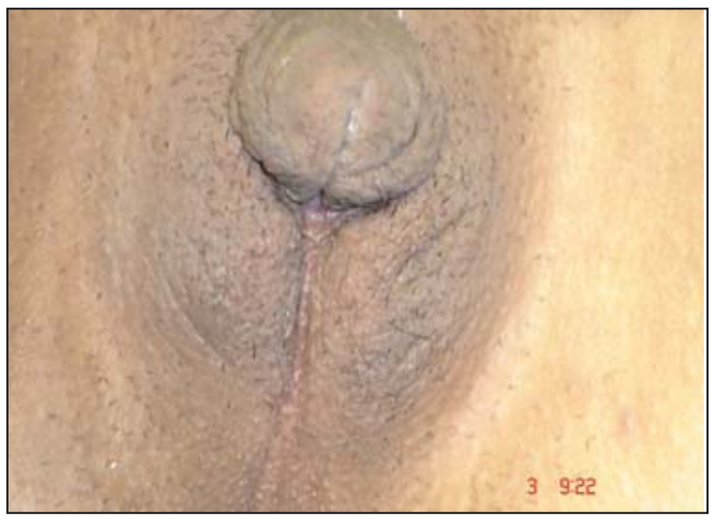

Figure 1: *Clitromegaly labila fusion*

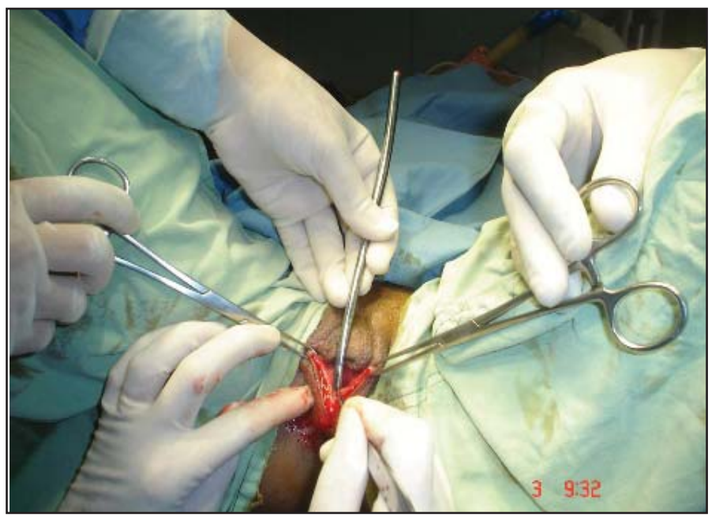

Figure 2: *Separating labial fusion*

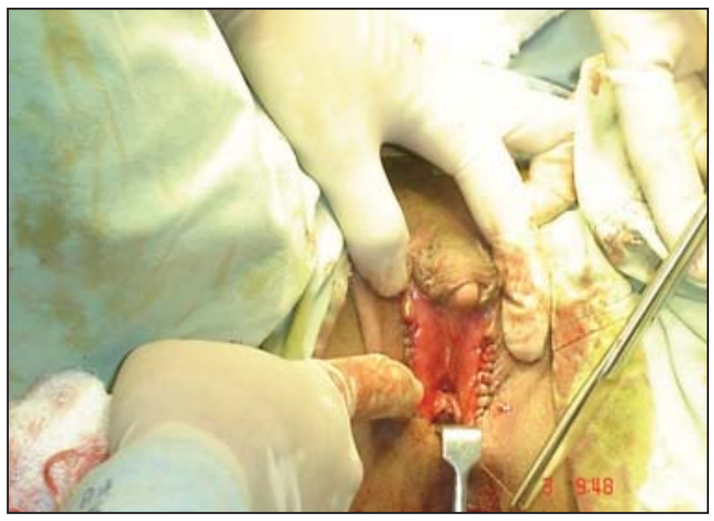

Figure 3: *Reconstrcution (vagina visible)*

She was a known case of Congenital Adrenal Hyperplasia with ambiguous genitalia and was on steroids orally. Her serum 17OH progesterone, electrolytes, urea and creatinine were in normal range. Her secondary sexual characteristics were well developed but there was genital ambiguity (clitromegaly) thick and labias fused together with no vaginal orifice. There was a small hole below the clitoris at the upper end of fusion (Figure 1). Her ultrasound revealed normal looking uterus and ovaries. Sinogram was performed through that hole in the fusion line which revealed well developed vagina. She was taken to operating room, a probe was inserted in the hole and both labias were separated in the centre about 3-4cms in length, urethra and vagina visualized (Figure 2). Haemostatic sutures were taken in the line of incision (Figure 3). On follow up; she is menstruating regularly, no problems so far.

Case 2

Miss B, 24 years old girl, unmarried and was getting married in next two months. She was diagnosed to have imperforate hymen and had drainage of hematocolpos at the age of 15 years (Figure 4,5), since then no further workup was done and she was menstruating regularly. She came for gynecological assessment before her marriage. Her secondary sexual characteristics were well developed but it was difficult to assess her vagina so Transperineal scan performed which revealed normal uterus and ovaries with a small bulge seen, that was Lower transverse vaginal septum. Excision of the septum was performed. Later she got married and there is no complaint of dysparunea.

Case 3

Mrs AB, 24 years old, married since 6 months, presented with non consummation of marriage. Her menstrual cycles were regular. Examination revealed lower transverse vaginal septum. Trans abdominal Ultrasound showed normal uterus and ovaries. Transperineal scan showed thick transverse vaginal septum near the hymen in the vagina. Excision of lower transverse vaginal septum performed. Later she conceived and delivered a live baby girl vaginally.

Case 4

Mrs.ABC, 22 years old girl, married for last four months presented with dysparunea. Examination revealed shortened vagina, possibly vaginal septum in the middle third. Transperineal scan showed a thick transverse vaginal septum in the middle third of vagina with normal, uterus, cervix and ovaries. Excision of septum was performed. She was followed till three months and there was no dysparunea but since then she is lost to follow up.

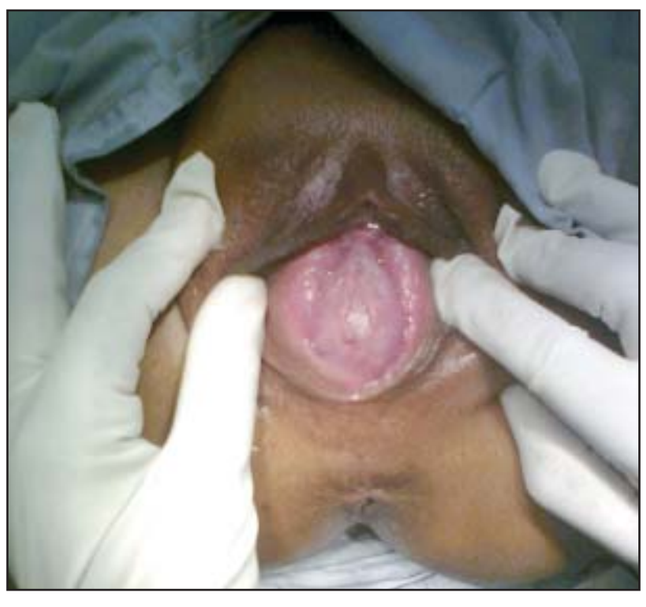

Figure 4: *Bluish bulging membrane of imperforate hymen*

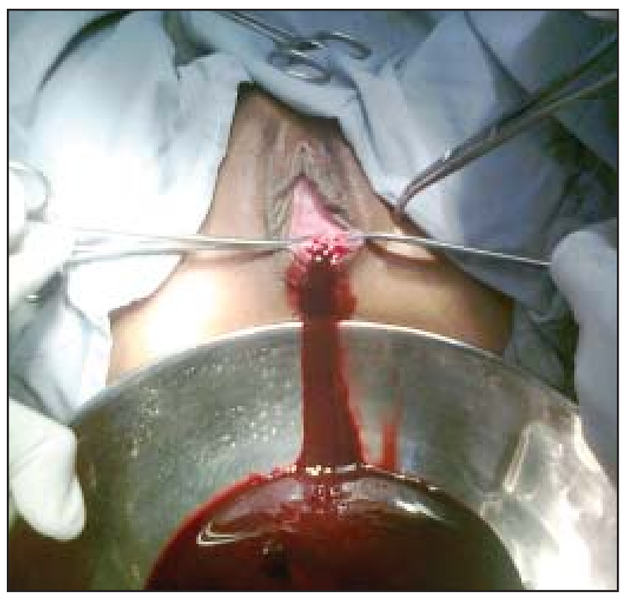

Figure 5: *After cruciate incision haematocolpose drained*

Case 5

Miss XY, 18 years old girl, presented with acute abdominal pain. She had the history of cyclical abdominal pain for which she was taken to a doctor, hematometra was identified and only the drainage of hematometra was performed abdominally, about four months ago. She had primary amenorrhea but used to have cyclical abdominal pain persistently. On examination, her secondary sexual characteristics were well developed, on perineal exam, there was no vagina. Rectally large tender mass felt most likely massive hematometra. Her abdominal ultrasound revealed hematometra, absent cervix and absent vagina. Her laparotomy was performed initially hematometra was drained then tried to identified the communicating tract between uterus and vagina but it was totally occluded with thick and dense tissue. So hysterectomy was performed followed by vaginoplasty. She was doing well for next six months but then she is lost form follow up.

Case 6

Mrs. XYZ, 24 years old married for 2 years and presented with primary infertility and purulent dirty vaginal discharge for last 4 years. She was actually seen by many gynecologists but couldn't diagnose properly so she presented very late to us. Her menstrual cycles were regular and secondary sexual characteristics were very well developed but on per vaginal examination, purulent copious discharge was present in vagina. A bulge was also felt in the right lateral wall of vagina. Cervix and uterus couldn't visualize because of discharge. Transvaginal ultrasound showed, uterine didelyphis, bicollis (Figure 6-A) and two hemivagina which were communicated with each other at the level of cervices . Right vagina was obstructed at the interoitus and it was filled with purulent fluid (Figure 6-C) but the left side vagina was patent from where discharge of right sided hemivagina was pouring out. Both ovaries were normal (Figure 6-B). Detailed abdominal and transrectal scan were performed but no other associated anomaly was identified. She received surgical treatment in two stages. In first stage, right sided hemivagina was drained by an incision in the septum, about 100 -150 ml purulent discharge drained out. Second stage was after 6 weeks and in which the entire longitudinal vaginal septum excised. It was uterine didelyphis bicollis and longitudinal vaginal septum dividing vagina in to two longitudinal hemivagina. After surgery uterine didelyphis with bicollis connected with single vagina. Later, she conceived and delivered a baby girl vaginally.

Case 7

Mrs ABC, 38 years old, married for 10 years presented with primary amenorrhea. She was sexually active with no complaints and there was no cyclical abdominal pain. Her examination revealed, well developed secondary sexual characteristics. Per vaginum, vaginal pouch of 3 cms ending blindly. Uterus and cervix were felt absent. Her ultrasound showed normal ovaries with absent uterus, cervix and upper part of vagina.

Her Serum estrogen and Day 21 Progesterone were normal. Couple counseled in detail regarding her problem and they refused for further workup and any treatment.. She is doing well except that she is amenorrhic. Her follow up was not possible as she was from northern part of Pakistan.

Discussion:

The exact etiology of such genital tract anoma-

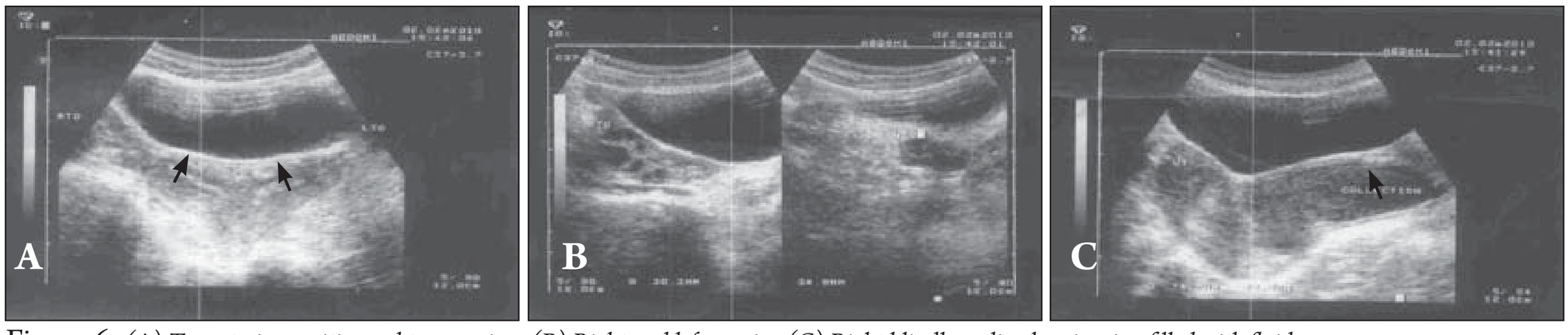

Figure 6: *(A) Two uterine cavities and two cervices, (B) Right and left ovaries, (C) Right blindly ending hemivagina filled with fluid*

lies is unclear. Any defect in the normal organogenesis involving the urogenital sinus or the Müllerian duct can result in genital tract anomalies. The nature of the Obstruction and embryogenesis can vary[2 4,5,8]. A Vaginal septum is a result of failure in canalization of Vaginal plate at various levels. Hymen represents a remnant of urogenital membrane, whereas cloaca and common urogenital sinus result from interruption of normal differentiation of hindgut. Associated congenital anomalies involving urogenital (incidence 30–50%), gastrointestinal, skeletal, and Cardiovascular system are common in patients with Complex anomalies[3,6,9–12].

Congenital abnormalities of genital tract are uncommon. Congenital absence of uterus and vagina (MRKH syndrome) has an incidence of 1 in 5000 female births[13]. Obstructive outflow disorder with a transverse vaginal septum has an incidence of 1 in 20 – 70000 female births[14]congenital absence of cervix is extremely rare that only case reports exists in the literature so far[15]. Incidence of uterine didelyphis with two hemivagina along with outflow obstruction is probably less than 1 in 100, 000 female births.[16]

Unawareness among physicians and relative rarity of these malformations has resulted in errors in diagnosis, needless abdominal exploration, and hysterectomies with mortality up to 50%[4,6,17]. Late onset of sexual development, ability of vagina to expand, normal looking vaginal orifice in mid and high transverse vaginal septum, and limited access to expert Medical facilities in the developing world can also de-lay the presentation as seen in the reported patients[18]. These anomalies usually manifest in neonates and in young girls of 9–15 years of age who have good secondary sexual development.

Lower abdominal mass and tense bulging bluish membrane at introitus is the most common presentation[2,4]. About 15% of abdominal mass in newborn girls are due to hydrometrocolpos[1]. Over distended vagina compresses the adjacent organs and can cause abdominal pain, intestinal obstruction, urethral obstruction, hydronephrosis, bladder perforation, and venous stasis in the lower extremities[4–6,19]. Urinary tract symptoms such as enuresis, urinary retention, overflow incontinence, and recurrent urinary tract infections are reported in more than 50% of patients[17,19].

A high index of suspicion and focused clinical examination can avoid incorrect diagnosis. Ultrasonography and magnetic resonance imaging (MRI) are diagnostic modalities of choice and can reveal associated renal anomalies and anatomy of complex lesions[1,2,5,8,10,12]. Further evaluation may require voiding cystourethrography (VCUG), intravenous pyelogram (IVP), renal scintigraphy, and endoscopy. Imperforate hymen and low vaginal septum need the incision in the membrane after the identification and catheterization of the urethra. The cut edges of hymeneal ring are sutured to vaginal mucosa with absorbable sutures[2,4]. However, in order to avoid injury to adjacent organs, combined abdominoperineal approach is preferable in high transverse vaginal septum and low vaginal atresia[10,13,20]. After abdominal exposure and drainage of vagina, a finger in lumen guides further dissection to perineum. An inverted U flap in the perineum is a valuable adjuvant to avoid anastomotic tension and stenosis[20].

Patients in whom the greater part of vagina and cervix is absent are difficult to manage. Fertility is doubtful in such circumstances even after extensive reconstructions[4]. It may be prudent to consider hysterectomy and reconstruction of vagina.

Management of common urogenital sinus and cloacal malformation is complex and a preliminary colostomy and urinary diversion may be required. Perineal procedure may result in sepsis and scarring making subsequent repairs difficult[2]. In order to get best results, a team of surgeons experienced in management of such lesions should perform genitourinary and bowel reconstruction simultaneously.

In cases of uterine didelyphis, the common presentation is septate vagina but rarely one of the hemivagina is obstructed, which presents with

hemi- hematocolpos or hemi- pyocolpos. Such cases are difficult to diagnose clinically because patient is menstruating with patent hemivagina and outflow obstruction from obstructed hemivagina is missed, which happened in one of our case and because of the same reason she was diagnosed very late when presented with hemi-pyocolpos. Exision of entire septum is the treatment.

Long-term follow-up after the reconstruction is crucial, as many patients may experience sexual difficulties, menstrual irregularities, infertility, spontaneous abortions, and premature delivery[4,8]. Infertility is probably due to endometriosis and pelvic adhesion in patients with outflow obstructions[4]. Rock et al. reported pregnancy rate of 47% in cases of lower obstruction, 43% in middle third, and only 25% in upper third obstructions[21].

Conclusion:

Rarity and variable presentation of congenital obstructing lesions of vagina can lead to delayed diagnosis and erroneous management. A high index of suspicion focused clinical examination and imaging help in early diagnosis. Centralization of care to clinicians with a special interest, watchful follow up and use of assisted reproductive techniques may preserve reproductive potential in these patients.

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