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Primary amenorrhea with transverse vaginal septum and scant hematocolpos: A case report

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ABSTRACT

Background: A genital outflow tract obstruction is an uncommon cause of primary amenorrhea. If ovulation occurs, menstrual bleeding is prevented. Patients typically present with abdominal/pelvic pain due to hematocolpos. Absence of significant hematocolpos could indicate a secondary source of primary amenorrhea and be challenging to the clinical diagnosis. Case: 17 year-old patient with primary amenorrhea, appropriate Tanner staging secondary sex characteristics, and transverse vaginal septum presents with virtual absence of hematocolpos. After vaginal septum resection, the patient began menstruating, although only evidenced by two cycles of vaginal spotting. Conclusion: Significant hematocolpos is an expected sequella of distal outlet obstruction when collated with secondary sexual characteristics. Absence of such along with suboptimal return of menstruation reflects pathophysiology which may be attributed to a coexistent disorder of the hypothalamus or higher central nervous system function.

Keywords: Amenorrhea; Hematocolpos; Distal Obstruction; Hypothalamic Dysfunction

1. INTRODUCTION

Primary amenorrhea is the lack of menses by age 15 with secondary sex characteristics, or at 13 with absence of secondary sex characteristics [1,2]. When evaluating a patient with primary amenorrhea, the pathophysiology can be attributed to numerous sources. Chromosomal abnormalities associated with gonadal dysgenesis are the most common, accounting for 40% of cases [3]. It is then followed by hypothalamic hypogonadism at 30%. A transverse vaginal septum/imperforate hymen represents only 3% - 5% of cases.

The physical exam is abnormal in 15% of cases [2,4]. Laboratory testing for follicle stimulating hormone (FSH), thyroid stimulating hormone (TSH), prolactin levels and a progestin challenge help determine the role of the endocrine system in the pathogenesis [2]. If there is evidence of gonadal failure, a karyotype should be performed. Persistently elevated prolactin could indicate a pituitary tumor. MRI is the most sensitive study to assess the pituitary gland [5]. A pelvic ultrasound would confirm the presence of a uterus and/or ovaries. Lack of adequate reproductive organ development requires specific surgical, medical, psychological and long-term follow-up.

In patients with a distal outlet obstruction, pelvic pain likely secondary to hematocolpos is a common complication. We describe a case of essentially absent hematocolpos in a patient with primary amenorrhea, appropriate secondary sex by Tanner staging and a transverse vaginal septum.

2. CASE

A 17-year-old nulligravid female presented to the adolescent gynecology clinic for primary amenorrhea. She underwent thelarche and pubarche at age 13 - 14, and was Tanner IV breast and pubic hair development. She denied abdominal pain, galactorrhea, headaches, visual disturbances, acne, or hirsutism. There were no changes in weight, or evidence for stress. Her growth chart was notable for a delayed growth spurt. She was an elite athlete, playing multiple sports. Thus, she exercised frequently. Her BMI was 20.51 kg/m^2. However, she attested to healthy eating habits as well as regular exercise. Her prenatal course was uneventful. She delivered at term at 6 l b 13 oz.

On physical exam there was a small vaginal-introital opening, without obvious bulging of the hymen. A Q-tip could be inserted to one centimeter. At her evaluation, her laboratory testing showed normal FSH 6.7 mIU/ml and LH 1.7 mIU/ml. Prolactin, thyroid function studies, and serum androgens were also within normal limits. Her Estradiol level was 35 pg/ml.

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Ultrasound revealed a normal midline uterus with an endometrial lining of 9 mm, and enclosed fluid in the cul-de-sac measuring 75 mm in length. There was a 39 × 36 × 31 mm heterogeneous right adnexal mass. There was no sonographic evidence of hematocolpos (**Figure 1**). Renal ultrasound revealed a junctional parenchymal defect within the right kidney without evidence of renal agenesis or duplication. Magnetic resonance imaging (MRI) confirmed a right adnexal lesion concerning for hemorrhagic ovarian cyst or hematosalpinx, and a dilated vaginal vault suggesting hematocolpos with a distal obstruction.

Laparoscopy was performed revealing a hemorrhagic ovarian cyst. Both fallopian tubes appeared normal and no endometriosis was visualized. Exam under anesthesia and needle aspiration of the vaginal vault revealed 0.1 ml "old blood" (hematocolpos). Although the MRI suggested hematocolpos, the laparoscopy findings were more aligned with the ultrasound, which demonstrated no evidence of hematocolpos. A transverse vaginal septum was resected. Six weeks postoperatively, a small Pederson speculum could be admitted with a depth of 7 cm. Eight weeks postoperatively, she had two episodes of vaginal spotting, each lasting 3 - 4 days, 28 days apart. She denied any premenstrual molimina. Additionally, she reported a clear, odorless vaginal discharge that was likely physiologic.

3. DISCUSSION

Distal obstruction(s) often result in the accumulation of menstrual fluid in the vagina or uterus described as hematocolpos or hematometra, respectively. Typically these patients present with cyclic pelvic pain and amenorrhea. As demonstrated by the literature review in **Table 1**, it is rare to present with a distal outlet obstruction, without abdominal pain and hematocolpos. Location of the sep-

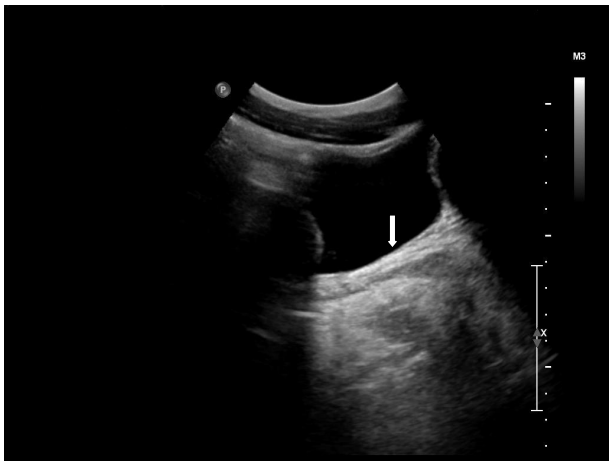

Figure 1. Absence of hematocolpos demonstrated by collapsed vaginal cavity (white arrow) on ultrasound.

Table 1. Literature review of hematacolpos/hematametra in setting of genital tract outlet obstruction.

Source	Type of Obstruction	Subjects	Mean Age	Abdominal/Back Pain	Hematacolpos/Hematametra
Amortegui, A.J., 1979 [12]	IH	1	13	Yes	200 ml
Sailer, J.F., 1979 [13]	IH	1	15	Yes	500 ml
Wenof, M., 1979 [14]	TVS	1	18	Yes	No, 15 ml PC
Rock, J.A., 1982 [15]	IH, TVS	22 IH, 26 TVS	IH 14.7, TVS 15.9	IH: 8 C, 14 P TVS: 11 C, 22 P	IH: 18, TVS: 23, MC-IH: 2, TVS:3
Shaw, L., 1983 [16]	IH, VA	14	9 - 15	11 patients	13 HC, 4 HM
Letts, M., 1990 [17]	IH	4	13	Yes	400 - 800 ml
McIvor, R.,1990 [18]	IH	1	14	Yes	Yes
Loscalzo, I.L., 1995 [19]	IH	1	13	Yes	1200 ml
Polasek, P., 1995 [20]	TVS	1	17	Yes	100 ml
Peterson-Sweeney, K.L., 1996 [21]	IH	1	13	Yes	800 ml
Kushnir, O., 1997 [22]	IH	1	13	Yes	Yes
Ahmed, S., 1999 [23]	IH, TVS	1	12.5	Yes	Large Amount, MC: 100 ml
Bakos, O., 1999 [24]	IH	1	13	Yes	400 ml
Buick, R.G., 1999 [25]	IH	1	14	Yes	1000 ml
Hall, D.J., 1999 [26]	IH	1	15	Yes	1500 ml
Rana, A., 2002 [27]	TVS	1	17	Yes	No, MC 500 ml
Ali, A., 2003 [28]	IH	15	14.3	All	15 HC, 9 HM

Continued

Chircop, R., 2003 [29]	IH	1	13	Yes	1000 ml
Liang, C.C., 2003 [30]	IH	15	13.2	11	No, 13 HM
Joki-Erkkilä, M., 2003 [31]	IH, TVS, LVS	16 TO, 10 LO	TO 14.3, LO 18.6	7 TO, 2 LO	300 - 1000 ml (both)
Stone, S., 2004 [32]	IH	1	11	Yes	300 ml
Sakalkale, R., 2005 [33]	IH	2	13.5	Yes	400 - 600 ml
Nazir, Z., 2006 [34]	IH, TVS	13 IH, 5 TVS	11-14	18 Abd, 7 Back	All
Walsh, B., 2006 [35]	IH	1	14	Yes	1000 ml
Ambegaonkar, G., 2007 [36]	IH	1	14	Yes	Yes
Acar, A., 2007 [37]	IH	65	13.9	All	All
Dane, C., 2007 [38]	IH	2	15	Yes	500 ml (1), Yes (1)
Mattern, M., 2007 [39]	TVS	1	14	-	Large Amount
Buyukbayrak, E.E., 2008 [40]	IH	1	13	Yes	300 ml
Kumar, K., 2008 [41]	IH	1	11	Yes	Yes
Adali, E., 2009 [42]	IH	1	12	Yes	1000 ml
Basaran, M., 2009 [43]	IH	2	13.5	All	All
Gyimadu, A., 2009 [44]	IH	1	12	Yes	450 ml
Mou, J.W., 2009 [45]	IH	3	12.5	All	500 - 600 ml
Saks, E.K., 2009 [46]	TVS	1	16	Yes	1000 ml
Dennie, J., 2010 [47]	TVS	3	13.7	All	All
Homa, L., 2011 (present case)	TVS	1	17	No	0.1 ml

IH: Imperforate Hymen; TVS: Transverse Vaginal Septum; VA: Vaginal Atresia; HTVS: High Transverse Vaginal Septum; TO: Transverse Obstruction; LO: Longitudinal Obstruction; C: Cyclic; P: Persistent; HC: Hematacolpos; HM: Hematometra; MC: Mucocolpos; PC: Pyocolpos.

tum can affect the timing of presentation. Septa in the lower third of the vagina, such as in this case, occur less frequently (15%), allowing for greater vaginal distension and later presentations [6]. Given that this patient did not present with the typical hematocolpos or lower abdominal pain that is associated with outlet obstructions, and had a suboptimal return of menstruation, she was evaluated for a secondary cause of primary amenorrhea, such as hypothalamic dysfunction.

Hypothalamic amenorrhea is typically characterized by amenorrhea in the absence of anatomic, chromosomal or organic abnormalities. Often a diagnosis of exclusion, it is one of the most common causes of secondary amenorrhea [7]. Hypothalamic dysfunction is associated with abnormal GnRH secretion secondary to a physical/psychological life event or an idiopathic process. Patients are characterized by low-normal gonadotropins, normal prolactin levels, normal sella turnica imaging and failure to demonstrate withdrawal bleeding [8]. Studies indicate that leptin influences the regulation of hypothalamic function, and its administration can induce GnRH pulsatility and menstruation [9]. Because estrogen deficiency is often seen in these patients, the effect of this condition on peak bone mass in young women is concerning. Treatment strategies should aim to address weight gain, exercise reduction, psychosocial factors, bone loss prevention, and the treatment of infertility [10]. In this patient, recognizing some features of the “female athlete triad:” low energy availability, amenorrhea, and osteoporosis, helped establish hypothalamic dysfunction as a possible secondary diagnosis [11].

In conclusion, determining the source of amenorrhea in an adolescent population can be difficult. Transverse vaginal septum in this case was not associated with hematocolpos to the degree that would be in accord with Tanner staging. Clinicians must be aware of atypical presentations and potential multifactorial etiologies of amenorrhea. Ensuring a thorough evaluation is essential in order to mitigate long-term effects of a misdiagnosis.

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