# Rare Diagnosis of Vaginal Atresia in a Patient with VATER Syndrome and Primary Amenorrhea

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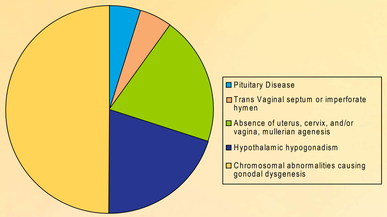
## Objective

This case presents the evaluation of amenorrhea in an adolescent patient with VATER/VACTERL syndrome.

## Background

Congenital abnormalities of the female reproductive organs account for approximately 20% of cases of primary amenorrhea.

**Most Common Etiologies of Primary Amenorrhea**




***A 14 year old female presents for evaluation of primary amenorrhea***

- PMH:
  - VATER syndrome
  - Severe GERD
  - Chronic headaches
- PGyn:
  - Thelarche/pubarche age 10.5
  - Denied cyclical abdominal pain
- PSHx:
  - Repair of tracheo-esophagal fistula
  - Surgical correction of VSD and PDA
  - Multiple esophageal dilitations
  - Correction of anal atresia
  - Laparoscopic right oophorectomy for teratoma
- Medications: Reglan TID
- Physical Exam
  - BMI 30
  - Acanthosis nigricans
  - Acne
  - Hursutism
  - Tanner Stage IV breast and genital development
  - Vaginal introitus not clearly visualized. Patient did not tolerate exam

## Laboratory and Diagnostic Evaluation

- **Prolactin 104.8 ng/ml**
- **Testosterone 93ng/dl**
- LH - 7.6 mIU/ml
- FSH - 5.7 mIU/ml
- TSH - 2.67 uU/ml
- Estradiol - 73 pg/ml
- DHEAS - 221 mg/dL
- Brain MRI - no intracranial lesions
- Renal US - within normal limits
- Pelvic US:
  - uterus 4.4 x1.9x4.2cm

## Initial Diagnosis and Management

- Primary amenorrhea secondary to:
  - Hyperprolactinemia from chornic use of Reglan
  - Polycystic ovarian syndrome
- Discontinue Reglan
- Recheck prolactin level after two months
  - 6.5 ng/ml

## 6 weeks later...

Patient presented to the Emergency Department with severe lower abdominal pain, worse in the right lower quadrant. A CT scan of the abdomen and pelvis showed the uterus and vagina to be distended with fluid suggestive of hydrometrocolpos. Left ovarian follicles were also visualized. The patient was then taken to O.R. for an exam under anesthesia.

**Exam Under Anesthesia**

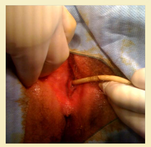


*Figure 1.* Intraoperative findings from exam under anesthesia. A Foley catheter is placed in the urethra, which was placed lower than expected. Findings showed a normal clitoris and clitoral hood, but there was no vaginal opening between the urethra and anus. The anal orifice was enlarged. Recto-vaginal exam revealed slightly enlarged uterus with no apparent vaginal mass.

**Pelvic MRI**

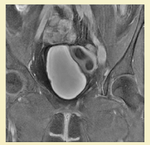


*Figure 2.* MRI showing hematocolpos with right-sided hematosalpinx and an atretic or hypoplastic vagina with fluid distending the cervix and uterine canal.

## Final Diagnosis and Management

### Vaginal Atresia

- Occurs 1 in 5,000-10,000 females
- Frequently associated with Mayer -Rokitansky -Kuster-Hauser (MRKH) syndrome: absent uterus, a deformed or missing vagina, normal ovaries and normal external genitalia
- Can be associated with Winter syndrome, which is characterized by renal agenesis and deafness
- Identified during adolescence when cyclic pain and a lack of menstrual flow indicates the condition.

### Referral to university-based fertility center

- Surgical correction of obstruction
- Operative findings
  - Blind vaginal introitus with occluded and dilated upper vagina approximately 6-8cm from the introitus;
  - Normal uterus, left ovary and tube;
  - Surgically absent right ovary;
  - Dilated right tube; and
  - Endometriosis

## Management

A patent and functional vagina was formed by creating a space between the bladder and rectum. The distended upper vagina was mobilized and attached to the introitus. The patient was subsequently instructed on the use of vaginal dilators and placed on a continuous Orth-Evra patch.

## Discussion

Primary amenorrhea in an adolescent can be a diagnostic challenge. The differential diagnosis is long and includes disorders of the hypothalamus, pituitary, thyroid and adrenal glands, ovarian failure, PCOS, and pregnancy. This case shows that a complete and timely physical examination is even more pertinent in the evaluation of primary amenorrhea, particularly when there is a history of congenital anomalies or birth defects. We would also recommend that pediatricians continue to include external genital examinations as a routine part of the pediatric physical assessment.