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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 include:

- Physical Exam
- Medications: Reglan TID
- Primary amenorrhea secondary to:
  - Hyperprolactinemia from chronic use of Reglan
  - Polycystic ovarian syndrome

Initial Diagnosis and Management
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.

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 uIU/ml

- Estradiol - 73 pg/ml
- DHEAS - 221 mg/dl
- Brain MRI - no intracranial lesions
- Pelvic US: - uterus 4.4 x 1.9 x 4.2 cm

- Renal US - within normal limits
- Testosterone 93ng/dl
- DHEAS - 221 mg/dl
- Prolactin 104.8 ng/ml
- LH - 7.6 mIU/ml
- FSH - 5.7 mIU/ml
- TSH - 2.67 uIU/ml

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.

Initial Diagnosis and Management

- Discontinue Reglan
- Recheck prolactin level after two months - 6.5 ng/ml

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.

Exam Under Anesthesia

- Identified during adolescence
- Recto-vaginal exam revealed slightly enlarged urethra and anus. The anal orifice was enlarged.
- 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.

Pelvic MRI

- MRI showing hematocolpos with right-sided hematometra and an atrial or hypoplastic vagina with fluid distending the cervix and uterine canal.

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 oral hood, but there was no vaginal opening between them. A hydrometrocolpos was noted. Figure 2. MRI showing hematocolpos with right-sided hematometra and an atrial or hypoplastic vagina with fluid distending the cervix and uterine canal.