

## Sexual infantilism accompanied by congenital absence of the uterus and vagina: Case report

JEROME H. CHECK, M.D.  
MARTIN WEISBERG, M.D.  
JANE LAEGER, M.D.

*Division of Reproductive Endocrinology and Infertility,  
Department of Obstetrics and Gynecology, The Jefferson  
Medical College of Thomas Jefferson University, Philadelphia,  
Pennsylvania*

THE ETIOLOGY of primary amenorrhea usually falls into one of three classifications: (1) hypothalamic-pituitary, (2) ovarian, or (3) müllerian. The first two categories frequently have a similar presentation, with sexual infantilism accompanying normal, albeit hypoplastic, internal and external genitalia (excluding ovaries). The third category is distinguished by normal secondary sexual development along with congenital absence of the uterus and/or upper two thirds of the vagina.<sup>1</sup>

We wish to describe the first reported case of sexual infantilism accompanied by congenital absence of the uterus and vagina.

The patient, a 22-year-old, white woman, presented with primary amenorrhea and sexual infantilism. She was of normal intelligence. She was 58 inches tall and weighed 89 pounds. General physical examination was normal. Breasts were Tanner Stage I and pubic hair was Tanner Stage II. The external genitalia were female and normal in appearance but a speculum could not be inserted. Similarly, bimanual vaginal

examination could not be performed and rectal examination failed to reveal a palpable uterus.

Pelvic ultrasonography showed neither uterus nor ovaries. Tomograms of the sella turcica were normal. Laboratory studies included serum luteinizing hormone (LH), 1.6 mIU/ml (normal, 2 to 10 mIU/ml); serum follicle-stimulating hormone (FSH), 2.1 mIU/ml (normal, 2 to 10 mIU/ml); serum prolactin, 2.5 ng/ml (normal, 0.0 to 25 ng/ml); thyroxine, 6.6 ng/100 ml (normal); and triiodothyronine resin uptake, 37% (normal). An 8 A.M. cortisol examination was normal at 12.8 µg/100 ml. Vaginal cytology showed 100% parabasal cells, indicating marked estrogen deficiency. Chromosomal analysis showed a normal 46,XX karyotype. An intravenous pyelogram was normal.

Examination of the patient under anesthesia similarly failed to demonstrate a midline pelvic structure and there was no vagina proximal to the fourchette. Diagnostic laparoscopy revealed normal-appearing ovaries and tubes bilaterally. The tubes ended in a uterine remnant on either side, which did not meet in the midline. There was no cervix and the proximal vagina was absent.

Treatment with estrogen and progesterone replacement and creation of a functional vagina by the Frank procedure were initiated.

Primary amenorrhea caused by failure of the hypothalamic-pituitary-ovarian axis is characterized by abnormalities of FSH and LH (low in the case of hypothalamic-pituitary failure and high in the case of ovarian failure). The resulting hypoestrogenism is manifest as sexual infantilism, but in both cases the uterus and vagina are present and normal. Hormonal replacement therapy is appropriate and indicated. In müllerian agenesis, on the other hand, the hypothalamic-pituitary-ovarian axis is normal and the patient is euestrogenic, presenting with normal secondary sexual development. Congenital hypoplasia or absence of the uterus and vagina is responsible for the amenorrhea.

It is frequently difficult to perform an adequate vag-

inal or rectovaginal examination on a hypoestrogenic patient. However, continuity between the uterus and the vagina must be documented in these patients prior to hormonal replacement therapy. Otherwise, an unexpected association with vaginal agenesis may result in hematometra or retrograde menstruation once cycles are established. This concern over estrogen replacement has heretofore been more theoretical than actual because there have been no previous case reports of müllerian agenesis coexisting with hypogonadotropic hypogonadism. In this case, a nonfunctional uterine remnant was found and estrogen therapy was given with only vaginal dilatation. However, congenital absence of the vagina may exist with a functional uterus, and, if this association is found, surgical intervention would be necessary.

#### REFERENCE

1. Griffin, J. E., Edwards, C., Madden, J. D., Harrod, M. J., and Wilson, J. D.: Congenital absence of the vagina (the Mayer-Rokitansky-Kuster-Hauser syndrome), *Ann. Intern. Med.* **85**:224, 1976.