

# Laparoscopic evaluation of congenital absence of the uterus.

## A case report

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### Summary

Anatomic gynecologic anomalies, including congenital absence of the uterus, occur with surprising frequency in referral practice of reproductive endocrinology and reproductive surgery. Female individuals with uterus agenesis have normal external genitalia and, therefore, the diagnosis is rarely made in infancy.

We report a case of congenital absence of the uterus in a young woman 25 years old, presenting with primary amenorrhea. The patient had a normal karyotype and normal secondary sex characteristics. Sex hormones were within normal limits, indicating normal function of the hypothalamic-pituitary-ovarian axis.

The diagnosis was set by bimanual gynecologic examination, which was followed by vaginal ultrasound examination.

The patient underwent a laparoscopic investigation, which revealed the absence of the uterus and the presence of small rudimentary uterine horns, connected to normal fallopian tubes. Ovarian morphology was bilaterally normal.

In conclusion we believe that every patient with genital tract abnormalities has to be dealt with individually, depending on the patient's anatomy, desires and age.

**Key words:** Congenital uterine absence; Müllerian agenesis; Primary amenorrhea; Infertility.

### Introduction

Anatomic gynecologic anomalies, including congenital absence of the vagina, defects in lateral and vertical fusion of the Müllerian ducts and disorders of sexual development occur with surprising frequency in referral practices of reproductive endocrinology and reproductive surgery. Sexual differentiation of the reproductive pathway begins early in the fetal period and is attributed to the influence of gonadal hormones – testosterone and Müllerian inhibiting substance (MIS) – on the dual wolffian and Müllerian ducts [1]. During the sixth week of development, a linear invagination occurs in the coelomic epithelium lateral to the mesonephric (wolffian) ducts forming the paramesonephric (Müllerian) ducts. In female patients in absence of MIS, the paramesonephric ducts persist [2]. The paramesonephric ducts approach each other and begin to fuse even before they reach the urogenital sinus. The fused ducts form a tube with a single lumen called “the uterovaginal canal”, which in turn forms the uterus and upper portion of the vagina.

The most serious developmental anomaly of the female reproductive tract is the congenital absence of the uterus and the vagina. In this disorder the Müllerian derivative of the vagina and the remaining Müllerian ducts, destined to become the uterus, fail to develop. The incidence of this disorder among newborn females is approximately one in 2,000-5,000 [3].

### Case Report

We report a case of congenital absence of the uterus with the presence of a dimple representing the urogenital sinus origin of the distal vagina, in a recently married woman 25 years of age, with primary amenorrhea. She did not complain about sexual dysfunctional problems.

The diagnosis was set during bimanual gynecologic examination, where no palpable uterine structure was found. The external genitalia were normal and the urethra opened at the proper site. The absence of the uterus was established by sonographic evaluation (Figure 1).

At admission, serum estradiol and gonadotropins were measured by radioimmunoassay methods. Serum 17-β-estradiol, FSH and LH levels were within normal limits, indicating an intact hormonal axis. The patient had a normal karyotype (46XX) and normal secondary sex characteristics.

A laparoscopic investigation was then performed, which revealed the absence of the uterus and the presence of small, rudimentary, cord-like uterine horns, connected to normal fallopian tubes. These horns were located on the pelvic sidewall, adjacent to the ovary (Figure 2). Ovarian morphology was bilaterally normal (Figure 3).

We noticed neither major abnormalities of the upper urinary tract such as unilateral renal agenesis, unilateral or bilateral pelvic kidney, hydronephrosis, hydroureter and malrotation of the kidney, nor any other minor defect (Figure 4).

Finally we did not note any spine or skeletal abnormalities (Figure 5).

The patient was informed about the importance and the consequences of this certain peculiarity and was advised to follow the “gestational surrogate” method (to use a gestational carrier) in order to have children.

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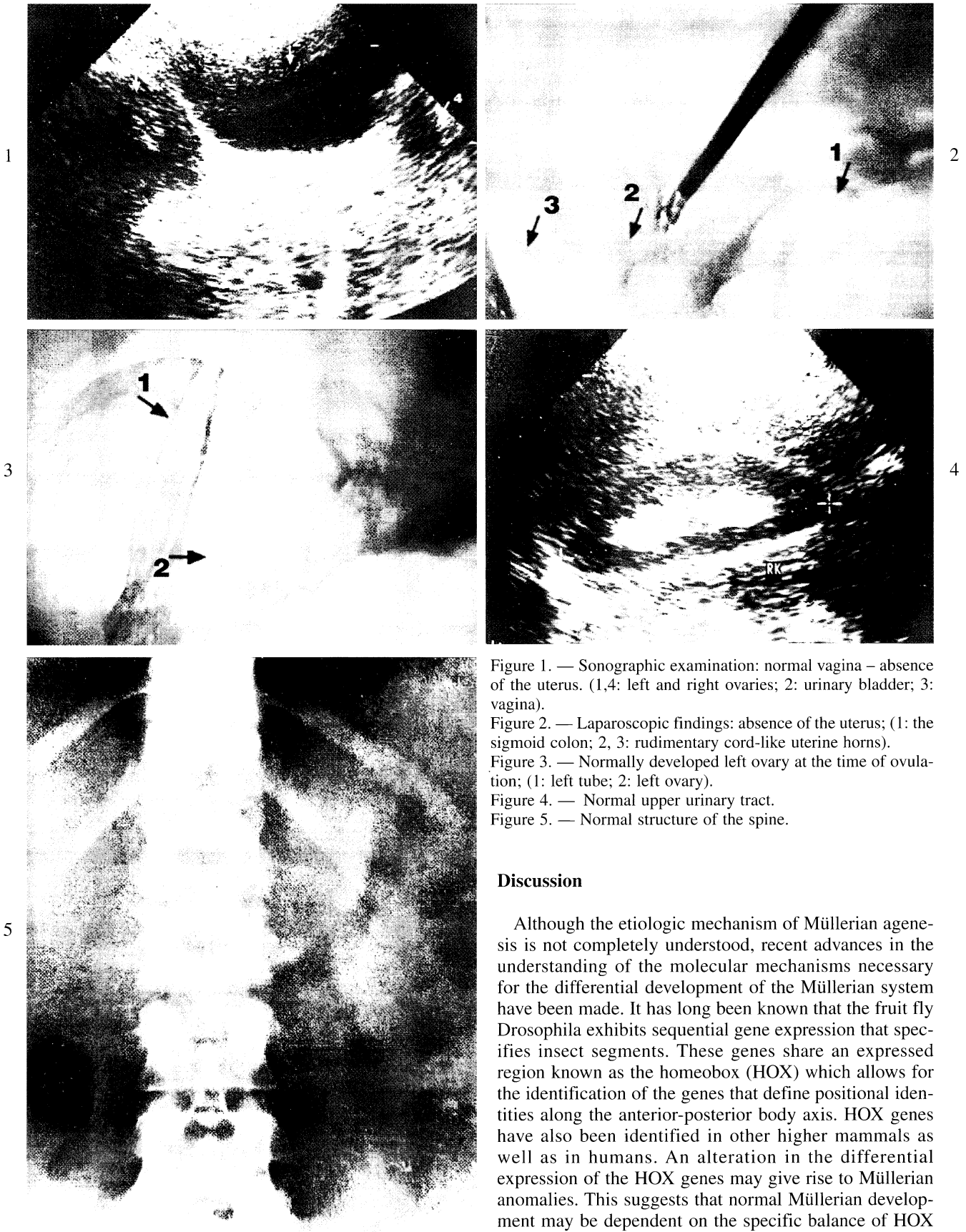


Figure 1. — Sonographic examination: normal vagina – absence of the uterus. (1,4: left and right ovaries; 2: urinary bladder; 3: vagina).

Figure 2. — Laparoscopic findings: absence of the uterus; (1: the sigmoid colon; 2, 3: rudimentary cord-like uterine horns).

Figure 3. — Normally developed left ovary at the time of ovulation; (1: left tube; 2: left ovary).

Figure 4. — Normal upper urinary tract.

Figure 5. — Normal structure of the spine.

## Discussion

Although the etiologic mechanism of Müllerian agenesis is not completely understood, recent advances in the understanding of the molecular mechanisms necessary for the differential development of the Müllerian system have been made. It has long been known that the fruit fly *Drosophila* exhibits sequential gene expression that specifies insect segments. These genes share an expressed region known as the homeobox (HOX) which allows for the identification of the genes that define positional identities along the anterior-posterior body axis. HOX genes have also been identified in other higher mammals as well as in humans. An alteration in the differential expression of the HOX genes may give rise to Müllerian anomalies. This suggests that normal Müllerian development may be dependent on the specific balance of HOX gene expression [4-7].

Müllerian agenesis may also be confused with androgen resistance syndrome, in which there may also be a shallow vaginal pouch and no uterus. Complete androgen resistance syndrome is more likely in patients with scant pubic and axillary hair. The definitive method for distinguishing between Müllerian agenesis and complete androgen resistance syndrome, however, is to determine the karyotype of the patient. Patients with Müllerian agenesis have a 46XX karyotype whereas those with complete androgen resistance have a 46XY karyotype.

The young patient presented in that report had a normal 46XX female karyotype. She complained only of primary amenorrhea and lack of fertility. We did not notice a familial occurrence of the syndrome. The normal ovarian function led to normal breast development and female body habitus.

In conclusion, successful management of congenital anomalies of the genital tract demands both intense psychological support and a high degree of surgical skill. In-depth consultation should be obtained or patients should be referred to centers where such expertise exists and where long-term follow-up can be provided. Finally each case has to be dealt with individually, depending on the patient's anatomy, desires, and age [8, 9].

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