

genetic, chromosomal, and anatomic abnormalities). All causes of secondary amenorrhea also can cause primary amenorrhea.

Disorders of the Uterus or Outflow Tract Abnormalities of the uterus and outflow tract typically present as primary amenorrhea. In patients with normal pubertal development and a blind vagina, the differential diagnosis includes *obstruction* by a transverse vaginal septum or imperforate hymen; *müllerian agenesis* (Mayer-Rokitansky-Kuster-Hauser syndrome), which can be caused by mutations in the *WNT4* gene, and *androgen insensitivity syndrome* (AIS), which is an X-linked recessive disorder that accounts for ~10% of all cases of primary amenorrhea (**Chap. 391**). Patients with AIS have a 46,XY karyotype, but because of the lack of androgen receptor responsiveness, those with complete AIS lack features of androgenization and have female external genitalia. The absence of pubic and axillary hair distinguishes them clinically from patients with müllerian agenesis, as does a testosterone level in the male range. The rare patient with 5 α reductase type 2 enzyme deficiency has a similar presentation but undergoes virilization at the time of puberty. *Asherman's syndrome* presents as secondary amenorrhea or hypomenorrhea and results from partial or complete obliteration of the uterine cavity by adhesions that prevent normal growth and shedding of the endometrium. Curettage performed for pregnancy complications accounts for >90% of cases; genital tuberculosis is an important cause in regions where it is endemic.

TREATMENT

Disorders of the Uterus or Outflow Tract

Obstruction of the outflow tract usually presents as dysmenorrhea or lower abdominal cyclic pain with no menses. Evaluation of the patient includes a medical history, physical examination including a perineal examination, and ultrasound imaging. In some cases, an MRI can more accurately identify the reproductive tract anomaly prior to surgery. It is important that surgery be performed as soon as the diagnosis is made as the risk of endometriosis is increased with retrograde menstrual flow. *Müllerian agenesis* may require surgical