



1800 Northside Forsyth Drive, Suite 380, Cumming, GA 30041
p: (770) 292-2670 • f: (770) 292-2671

CONDITIONS & TREATMENTS: BIRTH DEFECTS OF THE REPRODUCTIVE SYSTEM

Birth defects of the reproductive system are common, occurring in approximately 1 in 400 women. The reproductive system is derived from two tubes (the Müllerian ducts) which fuse in the midline followed by absorption of the central portion. The upper portion forms the fallopian tubes and uterus. The lower portion forms the vagina. Defects in this developmental process may include absence of the structures (Müllerian agenesis) and abnormalities in fusion and absorption.

Birth defects of the uterus can be diagnosed with ultrasound, sonohysterography, MRI or surgery. An x-ray hysterosalpingogram or hysteroscopy may suggest the presence of an abnormality; however, since neither test gives information regarding the outside of the uterus, other tests must be done to confirm the diagnosis.

Müllerian Agenesis

Müllerian agenesis, or Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, refers to absence of the uterus. It affects one in every 4,000 to 5,000 women. Approximately 15% of women with Müllerian agenesis will have defects of the urinary system and 12% will have abnormalities of the spine. Women with Müllerian agenesis have normal ovaries, normal hormones and may achieve a pregnancy with a gestational carrier. To achieve a pregnancy the woman with Müllerian agenesis undergoes IVF to retrieve the eggs, which are fertilized and transferred to a gestational carrier.

Women with Müllerian agenesis will have a very small, or absent, vagina. Several techniques have been described in order to enlarge, or create, the vagina. Two of the common techniques include dilation and surgery. Both the Frank and Ingram techniques use a series of dilators increasing in size. Alternatively, a type of skin graft known as the McIndoe procedure is used to create a vagina.

Septate Uterus

The septate uterus contains a ridge of tissue that protrudes into the uterine cavity. In a septate uterus the inside is the shape of a heart rather than the normal triangle. The outside of the uterus appears normal. This occurs in approximately 1 in 400 women and is a common cause of miscarriage.

The treatment of a septate uterus is usually an outpatient hysteroscopic surgery in which the septum is cut with a scissor through a hysteroscope (a small scope passed through the vagina into the uterus). The use of ultrasound at the time of surgery may help lead to a more complete surgery and is particularly useful in complicated cases.

Bicornuate uterus

A bicornuate uterus is heart shaped on the inside and on the outside. Surgical correction of a bicornuate uterus is rarely required.

Didelphic uterus

A didelphic, or double uterus results from failure of the two Müllerian ducts to fuse. There is often a double cervix and double vagina. Surgical correction is not usually required.

Unicornuate uterus

A unicornuate uterus occurs when only one of the two Müllerian ducts develops. Surgical correction is not usually required.

Outlet Obstruction

Outlet obstruction occurs when an opening anywhere from the cervix to the vagina is blocked. The most common form of obstruction is imperforate hymen. After puberty, menstrual blood becomes trapped behind the blockage. The collection of blood may become larger with each month, leading to a large painful mass in the pelvis or vagina. These defects are corrected surgically and the complexity depends on the type of defect, imperforate hymen being the simplest.