ABSENT UTERUS

Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome (Or Also Just Rkh Or Rkhs); Müllerian Agenesis; Müllerian Dysgenesis, Congenital Absence Of Uterus, Uterovaginal Agenesis, Uterovaginal Aplasia, Absent Endometrium; Genital Renal Ear Syndrome (GRES), Cais Syn

Female disorder in reproductive system at which a woman miss the uterus and thus she is not able to get pregnant and carry a child.

👩‍⚕️ Diagnosis  ♂ Female

About Absent uterus

Absent uterus is a disorder in female reproductive system. The most common reason of absent uterus is its removing during surgical procedure called hysterectomy. Hysterectomy may be made for many reasons. It can be used to treat chronic pain, certain types of cancer, infections, uterine prolapse (when uterus drops through the cervix and protrudes from the vagina), endometriosis (inner lining of the uterus grows outside and cause pain and bleeding) or adenomyosis (inner lining of the uterus grows into uterus muscle).

The extent of hysterectomy depends on the reason. Usually, the entire uterus is removed; moreover, the ovaries and fallopian tubes are removed as well during the surgical procedure. After removing the procedure, it is impossible for a woman to get pregnant.

Apart from those cases, when the uterus had to be removed surgically (hysterectomy) due to the complication (e.g. during the childbirth), the main cause of absent uterus is in incorrect development of Müllerian duct in embryonic stage of life. The Müllerian ducts (or paramesonephric ducts) are paired ducts of mesodermal origin in the embryo. In the female, they will develop to form the uterine tubes, uterus, cervix, and the upper one-third of the vagina; in the male, they regress.

Abnormal development of Müllerian duct

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome

Besides the absence of uterus, MRKH syndrome involves the absence cervix and/or vagina or its part; or the condition causes that the vagina and uterus are underdeveloped. The vagina canal is absent or remarkably shortened (Pic. 1), thus the intercourse is not possible. Women with MRKH syndrome have normal chromosome pattern (46, XX) and normally functioning ovaries. They also have normal external genitalia, breast and pubic hair development. However, renal (kidneys malformation, incorrect position or kidney agenesis), auditory and skeletal abnormalities are common association at MRKH syndrome. Usually, the first noticeable sign of MRKH syndrome is primary amenorrhea (the menstruation doesn’t starts by the age of 16).

This Müllerian agenesis (the failure of an organ to develop during embryonic growth) occurs in 1 out of every 4,000–10,000 females. However the primary cause of Müllerian duct abnormal
development is unknown and most cases develop in people with no disorder history in the family. The common name for this disorder is also Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome.

**Complete androgen insensitivity syndrome (CAIS)**

The second disorder which may become from Müllerian duct abnormal development is called CAIS disorder - complete androgen insensitivity syndrome. Women with CAIS have chromosome pattern 46, XY. The most importantly, due to chromosome pattern XY, the gonads in these women are not ovaries, but the testes instead. During the embryonic stage of development, testes form in an androgen-independent process that occurs due to the influence of the specific genes on the Y chromosome.

These women, however, have normal female external genitalia. They have typically feminine attributes and good breast development, despite they are little bit taller than average. Symptoms of CAIS do not develop until puberty, when no amenorrhea occurs. They also have shorten vagina and absent uterus.

The treatment of CAIS or MRKH syndrome is now limited just on symptomatic treatment. Hormone replacement therapy is recommended in cases with CAIS syndrome; women with CAIS syndrome also usually undergo gonadectomy (testes removing) to mitigate cancer risk. Most women also require vaginal reconstruction which may be surgical or nonsurgical.

If the uterus is absent, there is no chance for a woman to be pregnant. However, when the ovaries have normal development, it is possible to remove the eggs from the ovaries and create the embryo by artificial reproductive technologies (ART) and use the surrogate to give birth to the child, if law allows that.

**Associated diseases**

- uterine prolapse (uterus drops through the cervix and protrudes from the vagina)
- hysterectomy (absent uterus due to surgical procedure)
- endometriosis (inner linig of the uterus grows outside and cause pain and bleeding)
- adenomyosis (inner lining of the uterus grows into uterus muscle)
- uterine fibroids (benign growths of the uterus)
- Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome
- complete androgen insensitivity syndrome (CAIS)

**Complications**

- infertility
- amenorrhoea
- vaginal reconstruction (following surgical treatment to provide the normal sexual life)

**Risk factors**

- urinary incontinence
- vaginal prolapse (part of the vagina coming out of body)
- chronic pain after surgical procedure of hysterectomy
- infections
- injury of surrounding organs
- following hormonal therapy (because of removed organs, which produced hormones such as estrogen and progesterone)

All risk factors are connected with surgical removing of uterus, there are no risk factors in cases of MRKH syndrome and CAIS; the absent uterus in these cases is due to chromosomal abnormalities.
Impact on fertility

Uterus is main part of female reproductive system and place in the body, where the embryo and baby develop after the conceiving. Without uterus, the eggs from ovaries cannot be fertilized naturally and woman cannot conceive the baby and be pregnant.

Prevention

A hormonal birth control or an intrauterine device may successfully treat endometriosis and heavy vaginal bleeding the causes of possible hysterectomy in future. These options might also allow controlling the symptoms of uterine fibroids (causes of heavy bleeding)

There is no prevention, however, in case of absent uterus due to MRKH syndrome or CAIS.

Symptoms

At MRKH syndrome and CAIS, there are no symptoms until the age of puberty. The first symptom then is the absence of menses. Lately, at the beginning of sexual life, problems at intercourse appear, as the vagina is shortened or completely absent.

Symptoms may be connected to associated diseases, mostly in case, when hysterectomy is later executed. For hysterectomy, the most common reason is uterine fibroids, benign growths of the uterus. Uterine fibroids may cause excessive size of uterus, pressure or pain, bleeding or even severe anemia. Pelvic relaxation is another condition which may lead until hysterectomy. Mild relaxation may cause first degree prolapse when the cervix is on the halfway down to vagina. A woman experiences a loosening of the support muscles and tissues in the pelvic floor area.

Therapies

Self therapy

There are several causes for hysterectomy (surgical way of uterus removing) procedure, from which heavy bleeding or pain, are the most common ones. However, there are ways how to reduce the causes and thus to avoid hysterectomy.

Heavy bleeding and painful periods may be influenced by diet. For a woman with these problems, reducing the meat and dairy products consumption is recommended as well as avoiding to drink coffee and alcohol. Besides, stress management techniques are a plus.

In addition, dietary supplements with essential fatty acids, acidophilus and bioflavonoids are seen beneficial. These supplements acts as weak natural estrogens and are able to block stronger estrogens; the estrogen dominance is the primary cause of uterine fibroids development and thus pains and heavy bleeding. Natural progesterone is also the way to correct estrogen dominance and return hormonal balance to the body. Homeopathic medication and herbal remedies has shown to be beneficial as well to treat the problems with pains and heavy bleedings.
Correcting hormonal imbalance in general may be accomplished through a purification program, stress release, eating well and changing other lifestyle factors.

As regard to MRKH syndrome and CAIS, there is no possible self therapy or alternative treatment.

**Conventional medicine**

In case of uterus removing or in case of CAIS, the hormonal therapy is recommended to remain the hormone levels or as the prevention to later mineral deficiencies which may occur. Surgical therapy is good solution for women with MRKH syndrome or CAIS as vaginal reconstruction is often required.

**Pharmacotherapy**

**Hormone replacement therapy**

When removing uterus surgically, a woman looses the organs producing typical feminine hormones, which are needed to replace pharmacologically. For women with CAIS syndrome, the estrogen replacement therapy is critical to minimize bone mineral deficiencies later during the life.

**Surgical therapy**

**Gonadectomy**

Part of women with the CAIS diagnosis decides to undergo the operation of gonadectomy when the testes (at the place of ovaries) are removed as the prevention to later risk of tumors.

**Vaginal reconstruction**

As the vagina is shortened or completely absent in most cases, and thus the sexual intercourse very painful or not possible, the vaginal reconstruction is required. It may be provided by surgical or nonsurgical way.

The technique of nonsurgical vaginal reconstruction is based on using dilators and it is the appropriate first-line approach in most patients. Self-dilation requires patients to manually place the dilator on the vaginal dimple for approximately 30 minutes to 2 hours per day. The patients have to be aware that it will take several months to achieve their goal; however patients are able to achieve anatomic and functional success by vaginal dilation. Mentors (other patients with vaginal agenesis who have successfully dilated) are the big support to young women attempting dilation.

Surgical treatment is the option for female patients who were unsuccessful with dilators or for those who prefers this way of solving the problem. However, surgical creation of a vagina requires ongoing postoperative dilation or vaginal intercourse to maintain adequate vaginal length and diameter.

**Assisted reproduction**

If all treatments used by conventional medicine fail, the approaches of artificial
reproductive techniques are used.

If the ovaries show the normal development and normal function, there is possibility of egg retrieval and following in vitro fertilization (IVF). IVF and ART generally start with stimulating the ovaries to increase egg production. Most fertility medications are agents that stimulate the development of follicles in the ovary. Examples are gonadotropins and gonadotropin releasing hormone. After stimulation, the physician surgically extracts one or more eggs from the ovary, and unites them with sperm in a laboratory setting, with the intent of producing one or more embryos. Fertilization takes place outside the body.

The fertilized eggs (embryos) are cultivated under very stringent conditions and examined every day by the embryologist to evaluate their progress. The embryos are usually cultured for 3 to 5 days, before the best one(s) are selected to be put (transferred) in to the womb.

Morphological assessment of embryo appearance at the proper, distinct time points during development is a routine procedure in embryo selection. Moreover, time-lapse technology improvements has been evaluated as an aid to identify the embryo(s) with the highest implantation potential that enable to objectively select the embryo(s) for transfer. Time-lapse embryo monitoring allows continuous, non-invasive embryo observation without the need to remove the embryo from optimal culturing conditions.

The technique of selecting only one embryo to transfer to the woman is called elective-Single Embryo Transfer (e-SET) or, when embryos are at the blastocyst stage, it can also be called elective single blastocyst transfer (eSBT). It significantly lowers the risk of multiple pregnancies, compared with e.g. Double Embryo Transfer (DET) or double blastocyst transfer (2BT).

Infertile couples may also resort to egg donation or embryo donation when the female partner cannot have genetic children because her own eggs cannot generate a viable pregnancy.

However if the uterus is absent, there is necessary to place the fertilized egg into surrogate mother, if the law of the country allows to do so. Surrogacy via a gestational carrier is also an option when a patient's medical condition prevents a safe pregnancy, when a patient has ovaries but no uterus due to congenital absence or previous surgical removal, and where a patient has no ovaries and is also unable to carry a pregnancy to full term.

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**Therapies**

- **Egg donation**
  Process by which a woman donates eggs for purposes of assisted reproduction or biomedical research.
  Learn more at: [www.fertilitypedia.org/edu/therapies/egg-donation](http://www.fertilitypedia.org/edu/therapies/egg-donation)

- **ICSI**
  A micromanipulative fertilization technique in which a single sperm is injected directly into an egg.
  Learn more at: [www.fertilitypedia.org/edu/therapies/icsi](http://www.fertilitypedia.org/edu/therapies/icsi)

- **Sperm donation**
  The procedure in which a man (sperm donor) provides his sperm for fertility treatment.
  Learn more at: [www.fertilitypedia.org/edu/therapies/sperm-donation](http://www.fertilitypedia.org/edu/therapies/sperm-donation)
Standard IVF
A process in which an egg is fertilised by sperm outside the body: in vitro. Own or donated gametes may be used.
Learn more at: www.fertilitypedia.org/edu/therapies/standard-ivf

Gallery

Pic
Absent vagina in woman with Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome.

Sources
“Agenesia_de_vagina2” https://en.wikipedia.org/wiki/M%C3%BCllerian_agenesis#/media/File:Agenesia_de_vagina2.png”
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