Information for healthcare professionals

ROKITANSKY SYNDROME

INSTITUTO ROKI

Apoio a mulheres com
Síndrome de Rokitansky

To healthcare professionals,

I am a medical doctor and one of the creators of the Roki Institute. Diagnosed with Rokitansky Syndrome at the age of 20, throughout my treatment and up to this day, I frequently encounter the difficulty of obtaining adequate information and finding professionals who are prepared to deal with this little-known syndrome. The challenges are many, from correct diagnosis, finding qualified professionals, to choosing appropriate therapeutic methods for each phase of the syndrome and achieving better results with the least possible physical and emotional suffering. Success in treatment and achieving a healthy life can be achieved mainly with the help of well-prepared doctors in various specialties, healthcare professionals, and integrative medicine practices. This guide was produced to introduce you to the syndrome, inspired by the work of the Center for Young Women's Health, Boston Children's Hospital, Beautiful You MRKH, and the learning acquired in the journey towards health and well-being. We believe that, with shared experiences and constant encouragement of healthcare professionals to update their knowledge, we can build a better story of Rokitansky Syndrome in Brazil.

Welcome,

Claudia Melotti, co-founder of the Roki Institute

The Roki Institute Commitment

- To assist women with Rokitansky Syndrome in various phases - diagnosis, treatment, potential complications, sexuality and maternity related issues;
- To establish continuous medical education programs aimed at increasing diagnostic accuracy and adequate treatment;
- To encourage and connect Brazilian and international universities to conduct studies in genetic, diagnostic, and therapeutic areas;
- To facilitate the acquisition of dilators for those who choose conservative treatment;
- To create a network and connect professionals knowledgeable about the syndrome in the medical field (gynecologists, urologists, pediatricians, cardiologists, gastroenterologists, orthopedists, otolaryngologists), mental health (psychiatrists and psychologists), and integrative medicine (physiotherapists, physical educators, nutritionists, specialists in auxiliary methods such as Mindfulness, Yoga, Reiki, among others);
- To encourage the review and updating of laws related to adoption, surrogacy, and other points related to Rokitansky Syndrome that may arise;
- To organize an annual conference and virtual meetings in a protected chat, promoting the exchange of information and experiences.

The Rokitansky Syndrome

INFORMATION FOR HEALTHCARE PROFESSIONALS

What is Rokitansky Syndrome?

Rokitansky Syndrome, the name used in Brazil for the congenital disease that affects the female reproductive system, with alterations in the development of the Müllerian structures, appears in worldwide literature as MRKH, the initials of all the doctors who described it (Mayer, Rokitansky, Kuster, and Hauser). Maybe one day we can simply call it Roki Syndrome.

Classified into two types:

- Type I, with isolated involvement of the reproductive organs, has an incidence of 1:5,000 women;
- Type II, with associations of systemic alterations, has an incidence of 1:10,000-15,000 women.

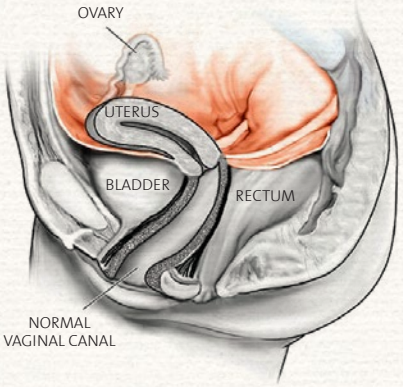
Characteristics of Type I

- Vaginal canal with reduced depth and diameter or complete agenesis;
- Absent or underdeveloped uterus;
- Ovaries and fallopian tubes with normal development;
- External genitalia (clitoris, urethra, hymen, vaginal lips) and anus with normal development.

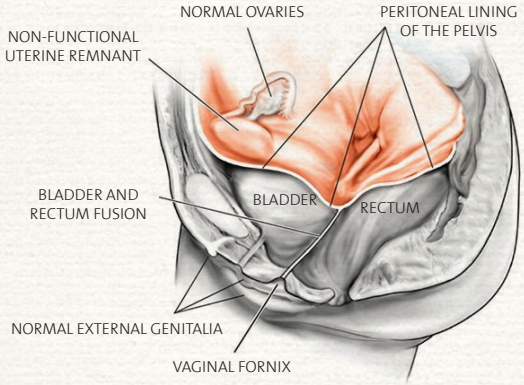
Characteristics of Type II

- Renal alterations with unilateral agenesis, horseshoe kidney, hydronephrosis, pelvic kidney, ectopic ureter; in approximately 40% of women with Type II;
- Bone abnormalities such as scoliosis, spina bifida, syndactyly, polydactyly or ectrodactyly, in approximately 30-40% of women with type II;
- Hearing impairments, with sensorineural hearing loss in 10-25% of women with type II;
- Cardiac abnormalities, with atrial septal defect or conotruncal defects in 2-3% of women with type II;
- Gastrointestinal abnormalities, with imperforate anus or other anorectal malformations;
- Abdominal abnormalities with single or multiple abdominal hernias.

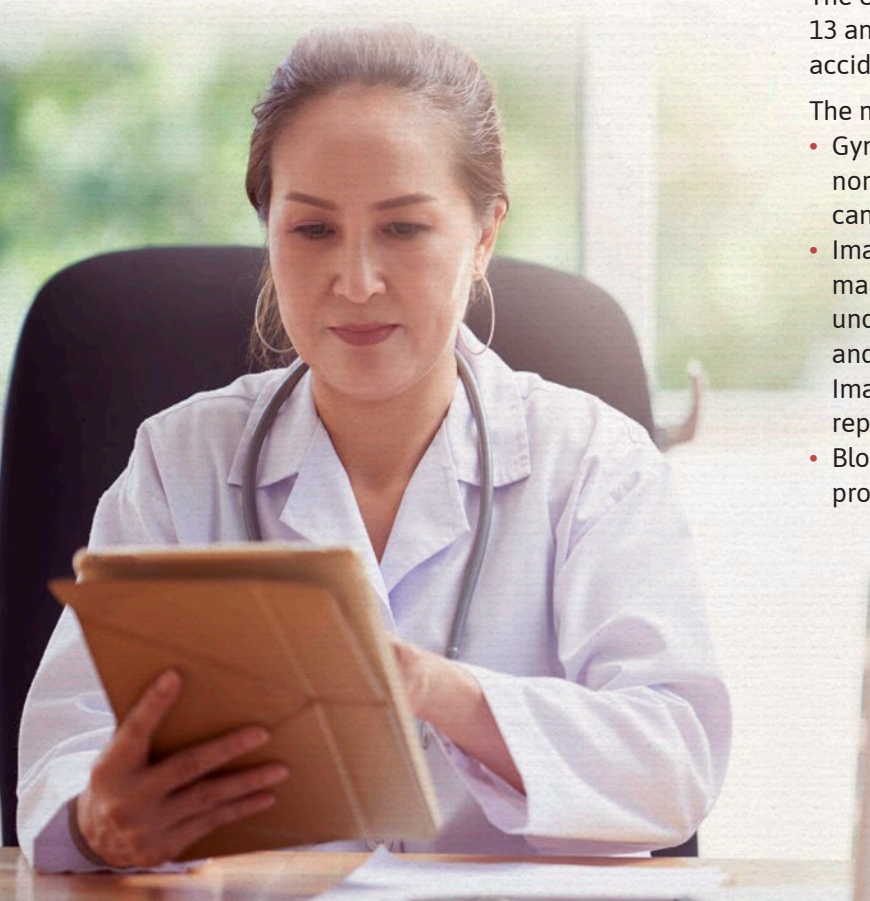
WITH ROKITANSKY SYNDROME



WITHOUT ROKITANSKY SYNDROME



Clinical history and diagnostic routine



Causes

The cause of Rokitansky Syndrome is still unknown.

The presence of cases within the same family and recent research suggest possible genetic causes, involving the LHX1, SHOX, and TBX6 genes. Ongoing studies should help to elucidate the genetic profile of the disease.

Diagnosis

The development of sexual characteristics is normal, so seeking medical help occurs due to amenorrhea, difficulty or pain during sexual intercourse.

The common age for diagnosis occurs between 13 and 18 years, sometimes anticipated due to accidents, urinary incontinence, or pelvic pain.

The most common exams are:

- Gynecological physical examination, which shows normal external genitalia and altered vaginal canal;
- Imaging exams such as pelvic ultrasound, the main screening test, shows the absence or underdevelopment of the uterus, normal ovaries, and renal characteristics. Magnetic Resonance Imaging can help visualize the internal female reproductive structures in more detail;
- Blood tests demonstrate normal sexual hormonal profile and female karyotype (46,XX).

Treatment options

Medical groups specialized in Rokitansky Syndrome, both Brazilian and worldwide, recommend vaginal dilation as the first treatment option for vaginal construction. Surgical options should be discussed only in cases where dilation is not successful.

It is important that the medical team pays attention and is sensitive to the appropriate time to start dilation. Often, patients prefer to wait a period between diagnosis and treatment.

Dilators

The technique of progressive dilation, known as the Frank method, was described in 1938. The success rate is directly related to the guidance provided by healthcare professionals who educate their patients about this method.

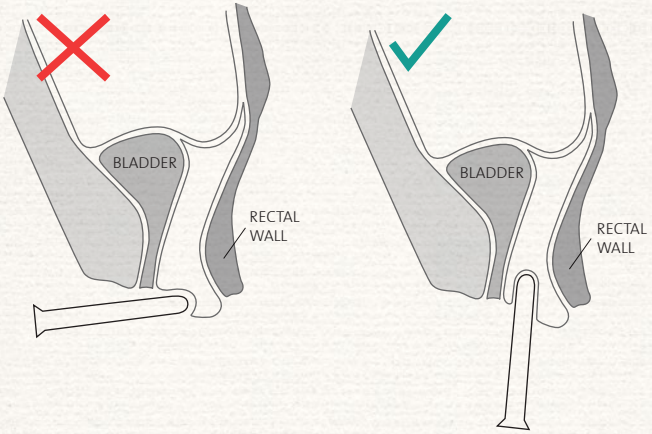
The plastic dilator kit has 5 or 6 sizes, ranging from 6 to 15 cm in length. The first dilator should be used, if possible, daily, with sessions between 20 and 30 minutes. With vaginal extension, the dilator of the immediately larger size is introduced, until treatment is completed with the use of larger dilators, which can reach lengths between 14 and 15 cm.

Treatment varies between 6 and 12 months. There are reports of short duration processes, between 3 and 6 months, when dilation exercises are done 2 to 3 times a day. Reports of longer durations, exceeding 12 months, have been made by patients who frequently interrupt the dilations.

Medical evaluations are recommended on a monthly basis or every two months. It's important to assess if the patient is applying pressure at the correct angle.

The correct use of dilators and their subsequent success depends on good guidance. Physiotherapists specializing in women's health are recommended for this guidance. According to Juliana Schulze, in addition to dilation, physiotherapy can help women better understand their own bodies, with exercises for the pelvic floor and techniques to facilitate dilation and self-awareness, such as electrotherapy, thermotherapy, and manual therapy, for example.

CAREFUL PLACEMENT OF DILATORS



Surgeries

Vaginoplasty or neovaginoplasty is indicated in cases of persistent failure of clinical treatment. At the appropriate time, the medical team should discuss with the patient and their family the surgical possibilities and the team's experience with each technique.

In the 19th century, surgical techniques used parts of the intestine for neovagina formation. Side effects such as mucous secretion in the area, bleeding, and pain led to a decrease in the use of this technique and the development of various others.

DAVYDOV SURGERY

Uses part of the peritoneum for vaginal construction. Cases of discomfort and umbilical pain from traction are reported.

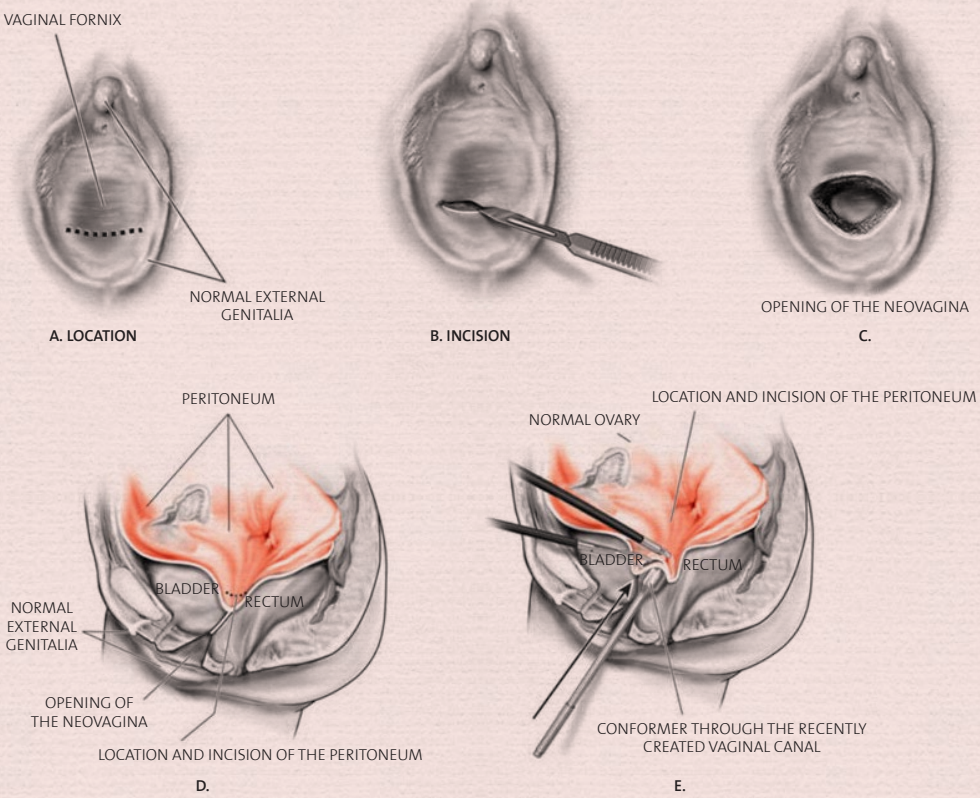


IMAGE: WWW.MIKLOSANDMOORE.COM

MCINDOE SURGERY

It used skin grafts on a rubber mold. Today called McIndoe-Banister, the technique has evolved in relation to molds and grafts. In the postoperative period, daily use of dilators is necessary for approximately 3 months.

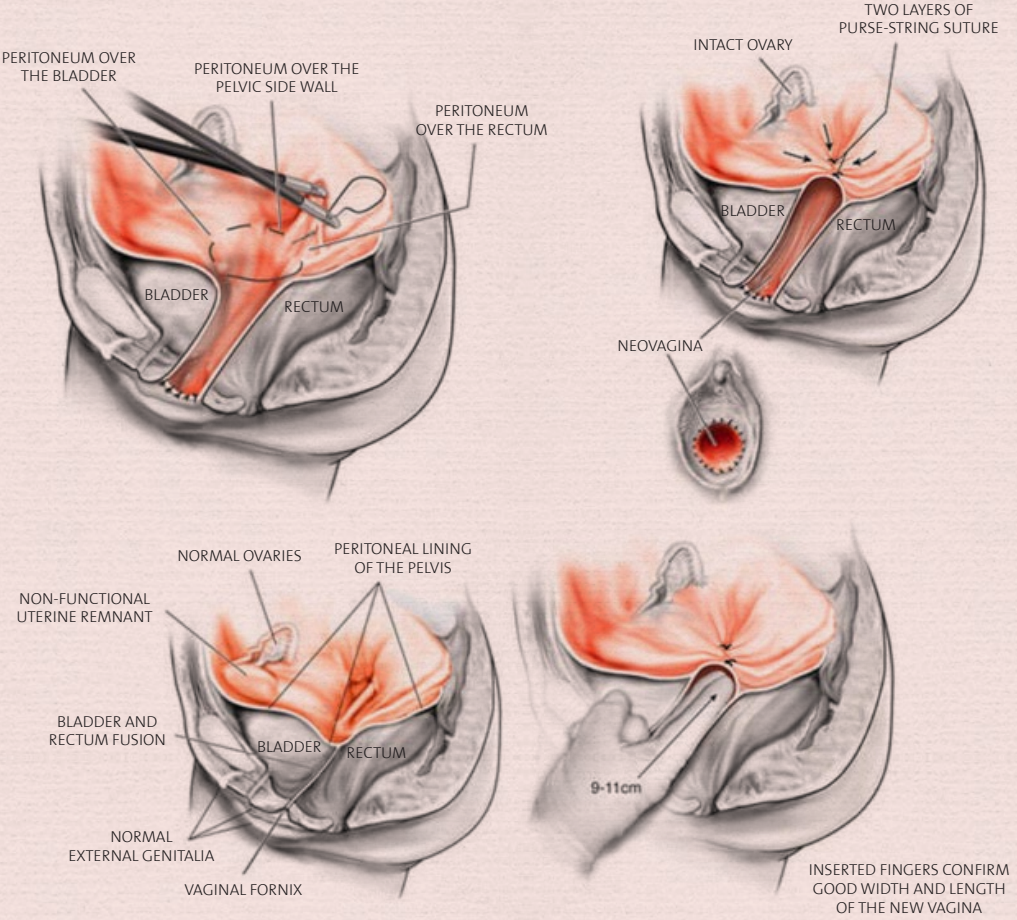


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IMAGE: WWW.MIKLOSANDMOORE.COM

WILLIAMS' SURGERY

Uses the skin of the labia majora to form a "vaginal pouch." Women who undergo this technique mention discomfort during sexual activity due to the axis of the new vagina, as well as the occurrence of hair growth.

VECCHIETTI SURGERY

Uses laparoscopy to fix threads that are externally attached to a traction system in the abdomen. The main complaints from patients are time, costs, care with the equipment, and a second surgery to remove the device.

Important guidelines

Your patient will likely have a normal sex life after treatment and will have options for motherhood. Sexuality and motherhood are important issues and should be approached with sensitivity and attention by healthcare professionals.

Surrogacy

Your patient can use her own eggs, her partner's sperm, or a donor's, and will be the biological mother of the child.

Uterine transplant

The first step is the in vitro fertilization procedure, in which eggs and sperm are collected and embryos are created and frozen for later use (up to this point, the same process as surrogacy). Next, it is necessary to find a compatible uterine donor and perform the transplant surgery. The months-long interval, or even up to a year, during which immunosuppressants are used, is necessary until the embryo transfer stage. The immunosuppressant is continued throughout the pregnancy and is only stopped after the cesarean section and removal of the transplanted uterus. Complications in all stages are reported. There are already some babies born from transplanted uteruses in the world. We remember that the first uterine transplant from a deceased donor occurred in Brazil in 2016. Currently, the procedure is performed on an experimental basis and is only indicated for women who have not undergone previous surgical neovaginoplasty.

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If you are assisting a patient with Rokitansky Syndrome,
please contact the **Roki Institute**.
Our commitment is to help you!

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