

Scottish Differences of Sex Development

SDSD

National Managed Clinical Network

Mayer Rokitansky Küster Hauser (MRKH) Syndrome

Information leaflet

This document has been prepared by Public Services Delivery Scotland (PSD Scotland) on behalf of the Scottish Differences of Sex Development (SDSD) network. Accountable to Scottish Government, PSD Scotland works at the heart of the health service providing national strategic services to the rest of NHS Scotland and other public sector organisations to help them deliver their services more efficiently and effectively. The SDSD network membership is made up of a multidisciplinary group of healthcare professionals who provide care to children and adults born with a difference in sex development or anomaly of the genitalia. The network is supported by a PSD Scotland Programme Team to drive improvement.

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What is MRKH?

MRKH (Mayer Rokitansky Küster Hauser) syndrome is a condition which is present from birth. It affects one in every 4500 – 5000 women. It describes the absence of the uterus (womb), cervix and upper part of the vagina. The ovaries are present and function normally: they produce eggs and the female hormones which are important for a woman's health and for development at puberty.

How do I know if I have MRKH?

When affected women go through puberty, they develop breasts and pubic hair, but they do not start their periods. Since the ovaries are present and producing female hormones, hair and breast growth occurs. Because there is no upper vagina or uterus, a period cannot take place. It is the lack of periods which leads most women to seek advice, usually at around 15-16 years old. Some women seek advice when they have difficulties with sexual intercourse.

Does MRKH affect other areas of the body?

Some women with MRKH have other associated medical issues. In Type 2 MRKH (or "MURCS"), tissues in addition to the reproductive organs may be affected, including the kidneys, the spinal column in the neck, hearing difficulties, and some heart problems.

What investigations will I have?

Your GP will ask you some questions and refer you to a specialist at a hospital, usually a gynaecologist. They will examine you and arrange further tests. These may include:

- A blood test – a chromosome test may be done as part of the investigation into absent periods (see Genetics and MRKH)
- An ultrasound scan or MRI scan of the pelvis to confirm the absence of the upper vagina, uterus and cervix and the presence of ovaries

If there are concerns about other problems which can be associated with MRKH, further investigations may be arranged. For example, an MRI scan of the lower part of the spine and kidneys.

Will I need to have cervical smear test?

No. Since you do not have a uterus or a cervix, it is not necessary to undergo cervical smear tests. In Scotland there is the HPV (human papillomavirus) vaccination programme where girls are vaccinated at around 13 years old. This is before the diagnosis of MRKH in most cases. However, the vaccine is still beneficial to women with MRKH as HPV can live in genital tissue other than the cervix. The vaccine also offers protection against genital warts.

What about sex?

Women with MRKH should be able to enjoy sex. Every woman is different and sex with a partner is an intimate and personal affair. Regardless of sexual orientation penetrative vaginal intercourse can present challenges, and with some help this can be improved.

Almost always, a woman can create an upper vagina by stretching her own tissues. This is usually done by a vaginal dilator – a smooth cylinder-shaped object which has been designed for this purpose. About 95% of women who practice dilator therapy will achieve satisfactory vaginal function. You will always have a nurse or doctor or psychologist to help you and support you with this. More information about dilator therapy is available from the clinical specialist team.

Do I need to use protection when I have sex?

It is always best to practise safe sex. You are still at risk of sexually transmitted diseases and viruses such as HIV or Hepatitis B/C. Use of condoms and dental dams, to practise safe sex, are advised.

Is it normal to feel distressed about MRKH?

Yes. Many women, and their parents, find MRKH very difficult to adjust to initially and can express feelings of shock, anger, depression, isolation, rejection and guilt. Many women find benefit from the support of a Clinical Psychologist and this is an important part of your care. Being in touch with other women who have the same condition can be a support also.

Will I be able to have children?

You will be able to have a biological child as you have functioning ovaries. However, you will not be able to carry a pregnancy as you do not have a uterus. You may need to use a surrogate for this. We will be happy to refer you to assisted conception services for further discussions when you are ready.

Adoption and fostering are other options that may be available and some women and their partners choose positively to accept being childless or child free.

Uterine transplant is in the early stages and is not available as yet. Research is progressing at a very fast pace and may become a reality in a few years.

Genetics and MRKH

How and why does it happen?

MRKH is a result of changes occurring between the 5th and 6th week in a pregnancy, when the areas that are affected in MRKH are developing in the embryo. The exact cause of the variation in development is unknown.

Our chromosomes are the rod-like structures within our cells that are the packages for our genes. It is now known that 20-30% of women with MRKH may have a small chromosome variation, called a microdeletion or microduplication. This may have either been the cause of, or contributed to the variation in the development of the womb.

You may be referred to Clinical Genetics to discuss genetic testing. Even if an initial chromosome test (called a karyotype) has been carried out, a detailed chromosome test (called an array-CGH) is required to detect most small chromosome variations.

In women with MRKH who have a normal detailed chromosome test the cause is usually uncertain. Research is ongoing into potential genes or genetic mechanisms that may be involved in MRKH. It may be that new genetic technology, which allows us to look across all our genes to identify any alteration (genome sequencing), will improve our understanding about MRKH in the future.

Can it be passed onto a child?

If a woman with MRKH is found to have a likely causative chromosome variation, then there is a 1 in 2 (50%) chance that this could be passed on. Often these small chromosome alterations are very variable, so even if this was inherited, we are often not able to predict how any future child may be affected.

If the chromosome test is normal, then it is very difficult to accurately predict the risk of recurrence in a family or passing MRKH onto a child. The current risk is quoted as between 1% and 5%. To date there are no reported cases of any affected offspring from surrogate pregnancies.

Further Information

This leaflet will be available in other languages (for example, Punjabi, Urdu, Polish) or formats (for example, larger print) on request.

Contact: nss.sdsd@nhs.scot

Website: nn.nhs.scot/sdsd

Useful links: nn.nhs.scot/sdsd/patients-and-families/support/

"Vaginal Dilation" Information Leaflet: nn.nhs.scot/sdsd/families/

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