Mayer-Rokitansky-Küster-Hauser Syndrome

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Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a disorder that occurs in females and mainly affects the reproductive system.

genetic conditions

1. Introduction

This condition causes the vagina and uterus to be underdeveloped or absent, although external genitalia are normal. Affected women usually do not have menstrual periods due to the absent uterus. Often, the first noticeable sign of MRKH syndrome is that menstruation does not begin by age 16 (primary amenorrhea). Women with MRKH syndrome have a female chromosome pattern (46,XX) and normally functioning ovaries. They also have normal breast and pubic hair development. Although women with this condition are usually unable to carry a pregnancy, they may be able to have children through assisted reproduction.

When only reproductive organs are affected, the condition is classified as MRKH syndrome type 1. Some women with MRKH syndrome also have abnormalities in other parts of the body; in these cases, the condition is classified as MRKH syndrome type 2. In this form of the condition, the kidneys may be abnormally formed or positioned, or one kidney may fail to develop (unilateral renal agenesis). Affected individuals commonly develop skeletal abnormalities, particularly of the spinal bones (vertebrae). Females with MRKH syndrome type 2 may also have hearing loss or heart defects.

2. Frequency

MRKH syndrome affects approximately 1 in 4,500 newborn girls.

3. Causes

The cause of MRKH syndrome is unknown. Changes in several genes that are involved in development before birth have been identified in females with MRKH syndrome. However, each has been found in only a few affected individuals, and it is unclear whether these changes cause MRKH syndrome. Researchers are working to determine how genetic changes might lead to problems with reproductive system development in females.

The reproductive abnormalities of MRKH syndrome are due to incomplete development of the Müllerian duct. This structure in the embryo develops into the uterus, fallopian tubes, cervix, and the upper part of the vagina. The cause of the abnormal development of the Müllerian duct in affected individuals is unknown. Originally, researchers suspected that MRKH syndrome was caused by environmental factors during pregnancy, such as medication or maternal illness. However, subsequent studies have not identified an association with any specific maternal drug use, illness, or other factor. Researchers now suggest that in combination, genetic and environmental factors contribute to the development of MRKH syndrome, although the specific factors are often unknown.

It is also unclear why some affected individuals have abnormalities in parts of the body other than the reproductive system. Certain tissues and organs, such as the kidneys, develop from the same embryonic tissue as the Müllerian duct, and researchers suspect that problems during development could affect these organs as well.

3.1. The Genes Associated with Mayer-Rokitansky-Küster-Hauser Syndrome

- LHX1
- SHOX

3.1.1. Additional Information from NCBI Gene:

- TBX6

4. Inheritance

Most cases of MRKH syndrome occur in females with no history of the disorder in their family.

Less often, MRKH syndrome is passed through generations in families. Its inheritance pattern is usually unclear because the signs and symptoms of the condition frequently vary among affected individuals in the same family. However, in some families, the condition appears to have an autosomal dominant pattern of inheritance. Autosomal dominant inheritance means that one copy of the altered gene in each cell is typically sufficient to cause the disorder, although the gene involved is usually unknown.

5. Other Names for This Condition

- congenital absence of the uterus and vagina (CAUV)
- genital renal ear syndrome (GRES)
- MRKH syndrome
- Mullerian agenesis
- Mullerian aplasia
- Mullerian dysgenesis
- Rokitansky Kuster Hauser syndrome

- Rokitansky syndrome

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