

MRKH syndrome caused by Intrauterine Placental Transfusion between sex-discordant twins?

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Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Reproductive tract and breast disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON43018

Source

ToetsingOnline

Brief title

MIPT study

Condition

- Reproductive tract and breast disorders congenital
- Congenital reproductive tract and breast disorders

Synonym

Mayer - Rokitansky - Kuster - Hauser (MRKH) syndrome

Research involving

Human

Sponsors and support

Primary sponsor: Vrije Universiteit Medisch Centrum

Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: chimerism, MRKH syndrome, twins

Outcome measures

Primary outcome

The presence of chimerism in MRKH patients, determined by Y-chromosome-specific real-time quantitative polymerase chain reaction.

Secondary outcome

In addition we want to test if the chimerism is only confined to blood, by testing (micro)chimerism in non-hematopoietic tissue (buccal smear) in MRKH patients. If the chimerism is present in the blood, but not in this non-hematopoietic tissue, this supports the idea that the chimerism is a result of blood sharing via intrauterine placental connections.

Study description

Background summary

Mayer Rokitansky Küster Hauser (MRKH) syndrome is a congenital disorder, characterized by aplasia of the uterus and the upper two thirds of the vagina. The aetiology of this disease is unknown. The freemartin phenomenon represents a similar phenotype in cattle, in which female calves are lacking the Müllerian duct derivatives. This phenomenon occurs in sex-discordant calf-twins, in which a shared placenta allows blood exchange. It is speculated that placental transfusion of antimüllerian hormone (AMH) from male to female calf-fetus is a possible cause. This placental transfusion also results in blood chimerism; the existence of two blood cell lines in one organism derived from two genetically distinct zygotes.

We hypothesize that in humans too, transfusion of AMH intra-uterine from a male co-twin to a female co-twin is responsible for the development of MRKH in the female co-twin. The Müllerian duct in human develops in the sixth week of pregnancy. This means that possible twin-to-twin-transfusion of AMH - resulting in regression of the duct - takes place in an early stage in fetal development. Therefore it can also occur in the case of a spontaneous reduction of one fetus

(vanishing twin). A vanishing twin can leave its traces by microchimerism: in which a second cell line is present with a low concentration in the surviving fetus.

Study objective

The aim of this study is to determine whether male microchimerism is present in patients with MRKH syndrome. This would be a sign of intrauterine cell trafficking - and possible AMH transfer- from male to female co-twin. This placental blood transfusion could be the cause of the origin of the MRKH syndrome.

Study design

Observational case control study.

Study burden and risks

This is a non-therapeutic study. In the course of this study we ask the subjects for one visit to the outpatient clinic or a home visit for blood sampling and a short questionnaire.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age

Adults (18-64 years)

Elderly (65 years and older)

Inclusion criteria

Patients:

- Diagnosed with MRKH syndrome
- 18 - 50 years;Control group: (already sampled as control patients in a previous study)
- 18 - 50 jaar

Exclusion criteria

- not willing or able to sign the informed consent
- reported pregnancy (in the control group)

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

Recruitment

NL

Recruitment status: Recruitment stopped

Start date (anticipated):	02-03-2017
Enrollment:	96
Type:	Actual

Ethics review

Approved WMO	
Date:	28-12-2016
Application type:	First submission
Review commission:	METC Amsterdam UMC
Approved WMO	
Date:	19-10-2018
Application type:	Amendment
Review commission:	METC Amsterdam UMC

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL57503.029.16
Other	TC = 5961