Long term functional outcome, quality of life, and genetic factors in patients with a cloacal malformation

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The primary objective to our study is assessing functional outcome, quality of life, and genetic mutations in patients with a cloacal malformation. Functional outcome:- Colorectal functiono Objective: assessing colorectal function through the use of...

Ethical review	Approved WMO
Status	Pending
Health condition type	Gastrointestinal tract disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON39349

Source
ToetsingOnline

Brief title
Cloaca: functional outcome, quality of life, and genetic factors

Condition

- Gastrointestinal tract disorders congenital
- Genitourinary tract disorders NEC
- Gastrointestinal therapeutic procedures

Synonym
congenital fusion of rectum, urogenital sinus with a rectovaginal fistula, vagina and urethra

Research involving
Human

Sponsors and support

Primary sponsor: Erasmus MC, Universitair Medisch Centrum Rotterdam

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

Intervention

Keyword: Cloaca, Genetic, Outcome, Quality of life

Outcome measures

Primary outcome

Primary outcome of the study are the scores from the completed questionnaires, the results from the urologic tests, the results from the gynecological consult and the MRI-study, laboratory results from urine and blood samples, and the results from the genetic tests.

Secondary outcome

The results from the questionnaires indicate colorectal function, urological function, gynecological function and sexual functioning. The two days voiding chart diary and the results from the urological tests both indicate bladder function. Laboratory results show the presence of any (chronic) inflammation of the urinary tract. Gynecological consult outcome as well as results from the MRI study will indicate gynecological and sexual function and will give information about fertility status. Laboratory results from the blood sample tests will indicate renal function. Genetic test results will show any genetic mutations that may play a role in the origination as well as the inheritability of cloacal malformations.

Study description

Background summary

A cloacal malformation is a severe congenital abnormality. In this rare condition urethra, vagina and rectum are fused in one common channel, presenting as one single perineal orifice. With a prevalence of one in 50.000 newborns it accounts for approximately 10% of all anorectal malformations. Complex surgical repair is needed in order to construct a close to normal rectourogenital tract. Aim to this surgical approach is achieving appropriate colorectal, urological and gynecological function. Studies on this long term functional outcome of patients born with a cloacal malformation are scarce, but when available, show many cases of impaired colorectal, urological or gynecological function. These studies are all retrospective and with different descriptions and scoring systems for functional outcome. A cross-sectional assessment of functional outcome in patients with a cloacal malformation has never been conducted. Quality of life studies have been done in patients with other anorectal malformations (ARM) in the Netherlands and show lower scores in the ARM group compared to a control group. Quality of life of patients born with a cloacal malformation has never been assessed. Few studies report bladder function that is assessed by urological tests. Assessing long-term functional outcome and the quality of life in patients with a cloacal malformation can show effectiveness of the current postoperative management in these patients. Results might indicate a need for adjusted follow-up of patients with a cloacal malformation in order to achieve higher quality of life.
Further, some studies have been carried out in order to find the genetic origin of anorectal malformations and cloacal malformations in special. Till so far, this has not lead to a specific gene or mutation that leads to this congenital abnormality. Since these patients are born of healthy parents in most cases the onset of the cloacal malformation seems sporadic. Therefore de novo mutations might play an important role in the onset of cloacal malformations. Exome sequencing is a useful technique for identifying de novo mutations, by studying both parents and affected patient. Information about the genetic onset of cloacal malformations will increase knowledge about the development of such a malformation as well as the chances of inheritability of this complex form of ARM.

Study objective

The primary objective to our study is assessing functional outcome, quality of life, and genetic mutations in patients with a cloacal malformation.
Functional outcome:
- Colorectal function
o Objective: assessing colorectal function through the use of a questionnaire
o Question: Do patients with a cloacal malformation have an impaired colorectal function?

- Urological function
o Objective: assessing urological function through the use of a questionnaire, ultrasounds of the urinary tract (pre- and postvoiding), and urine samples.
o Question: Do patients with a cloacal malformation have an impaired urological

function?

- Gynecological and sexual function
o Objective: assessing gynecological and sexual function through the use of a questionnaire.
o Question: Do patients with a cloacal malformation have an impaired gynecological and sexual function? Do patients with a cloacal malformation have a normal fertility?

- Renal function
o Objective: assessing renal function through creatinine clearance rates in blood samples.
o Question: Do patients with a cloacal malformation suffer from renal dysfunction?

Quality of life:
o Objective: assessing quality of life through conducting two questionnaires
o Question: Do patients with a cloacal malformation have a lower quality of life?

Genetic mutations:
o Objective: assessing mutations in both study subjects* and parents of study subjects* DNA by conducting exome sequencing and comparing study subjects* exome with the exome of the parents of the study subject.
o Question: Do patients with a cloacal malformation suffer from genetic mutations that can be related to their congenital abnormality?

Secondary objectives:
o Evaluate postoperative management of patients with a cloacal malformation through test results and questionnaire outcomes.
o Evaluate risk of inheritability and cause of onset of cloacal malformations.

Study design

The study design is one of a cross-sectional study. The study population will consist of patients who were born with a cloacal malformation and were treated in Rotterdam. Participants will spend approximately 4 hours of time to complete the study. Prior to the hospital visit participants will be asked to complete 3 questionnaires, to keep a two days voiding chart diary, and to collect a urine sample. During the hospital visit participants will undergo two urologic test. There will be collected a blood sample from each participants and both her parents. Finally, all patients who reached puberty will undergo a gynecological consult followed by a MRI of the pelvic floor. After this, participants can go home. No further contributions to the study are required.

Study burden and risks

The risks of participating in this study are minimal. All parts of the study are proven to be safe. The collection of a blood sample through a venal punction can be a little annoying. Ultrasound studies as well as a MRI study are without any side-effects. Participants will not benefit directly from participating in the study, because there is no therapeutic aim to this study. Over time participants might benefit because of the re-evaluation of functional outcome that is done in the study. This, together with the assessment of quality of life, might lead to a better postoperative follow-up in these patients. When test results indicate that further investigations are required participants will be referred to their doctor.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

Patiënt is born with a cloacal malformation
Patiënt is born after 1984 and before 2006

Exclusion criteria

Patient is lost in follow-up

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-01-2013
Enrollment:	60
Type:	Anticipated

Ethics review

Approved WMO

Date:	09-09-2013
Application type:	First submission
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)

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	NL38461.078.12