Unravelling the aetiology of pyloric stenosis; a role for the COL3A1 gene?

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Ethical review	Approved WMO
Status	Pending
Health condition type	Gastrointestinal stenosis and obstruction
Study type	Observational invasive

Summary

ID

NL-OMON34013

Source
ToetsingOnline

Brief title
COL3A1 gene and pyloric stenosis

Condition

- Gastrointestinal stenosis and obstruction

Synonym
infantile hypertrophic pyloric stenosis, pyloric stenosis

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Stichting Kindermotiliteit

Intervention

Keyword: aetiology, COL3A1, genetics, pyloric stenosis

Outcome measures

Primary outcome

Primary outcome:

The occurrence of mutations or polymorphisms of the COL3A1 gene in patients

with pyloric stenosis

Secondary outcome

Secundary outcome:

- the results of Collagene IIIa protein analysis in patients with pyloric

stenosis

- the clinical characteristics of patients with pyloric stenosis and their

relatives

Study description

Background summary

Pyloric stenosis is a relatively common gastro-enterologic condition in newborns. A surgical approach, by means of a pylorotomy, is the general treatment of this disorder.
The aetiology of pyloric stenosis is largely unknown.
There are several studies suggesting a genetic basis for pyloric stenosis. Reports of multiple affected individuals in one family have been published. Furthermore, coexistence with congenital malformations and syndromes point to a genetic involvement as well. Up till now, no locus in the human genome has been found to be evidently associated with pyloric stenosis.
It is remarkable that pyloric stenosis is also seen in patients with Ehlers Danlos type IV, an inherited connective tissue disorder, caused by mutations of the COL3A1 gene and that next to this a group of patients with pyloric stenosis is reported with evidently more frequent occurrence of hypermobility of the joints (one of the symptoms of Ehlers Danlos Syndrome) than is seen in the

healthy population.

Study objective

The aim of this study is to improve insight in potentially genetic factors contributing to the development of pyloric stenosis.
We aim to investigate the association of the COL3A1 gene and pyloric stenosis because of concrete signs in the existing literature for an association.

Study design

In a cross-sectional cohort study, we will include consequent newborns diagnosed with pyloric stenosis and therefore undergoing a pylorotomy.
After informed consent is obtained, 0.5 ml of EDTA blood from an existing intravenous canules and a biopsie of connective tissue from the main incision will be taken from the patient.
Parents are asked to fill out a questionnaire concerning the medical history of the child and to answer questions about occurrence of gastrointestinal motility disorders and connective tissue disorders in their families.
Blood and tissue will be analyzed in the DNA and protein laboratory of the VU Medical Centre in Amsterdam. Polymorphisms or mutations will be compared to the normal results of a healthy population, available in the database of the laboratory.

At first, we will include 5 patients and make a subanalysis. Whenever we find mutations or polymorphisms in this group, we will proceed to include more patients, with a maximum of 25 to 30.
Given the explorative character of this study, it is impossible the make an exact power calculation preceding the study.

Study burden and risks

It will take parents about 20 minutes to complete the questionnaire.
Patients will be under complete anesthesia during the pylorotomy. During the procedure, blood from an existing intravenous canule will be taken and a biopsie will be performed.
The extra biopsy taken during the surgical procedure does not involve an increased risk for the patient.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Children (2-11 years)

Inclusion criteria

Infants diagnosed with pyloric stenosis, undergoing a pylorotomy.

Exclusion criteria

Infants diagnosed with pyloric stenosis who already underwent a pylorotomy.
Non-Caucasians

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:	Pending
Start date (anticipated):	01-02-2009
Enrollment:	25
Type:	Anticipated

Ethics review

Approved WMO

Application type:	First submission
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	NL26091.018.08