Genetics of megacystis-microlon-intestinal hypoperistalsis syndrome

Published: 05-10-2011
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Identification of the underlying genes of MMIHS

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON39441

Source
ToetsingOnline

Brief title
Genetics of megacystis-microlon-intestinal hypoperistalsis syndrome

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Gastrointestinal motility and defaecation conditions
- Bladder and bladder neck disorders (excl calculi)

Synonym
megacystis-microlon-intestinal hypoperistalsis syndrome

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Groningen
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: congenital, Genetics, megacystis-microlon-intestinal hypoperistalsis syndrome

Outcome measures

Primary outcome

Identification of the underlying genes of MMIHS

Secondary outcome

none

Study description

Background summary

Megacystis-microcolon-intestinal hypoperistalsis syndrome (MMIHS) is a rare congenital disorder. Patients with MMIHS have a distended, hypotonic urinary bladder which is not obstructed (the ureters are sometimes dilated and hydronephrosis can be present), they have hypoperistalsis throughout the entire gastrointestinal tract causing functional small bowel obstruction, they have a micro-colon and some patients have intestinal malrotation. To date, around 100 cases have been reported in the literature. As familial cases of MMIHS have been reported, genetic factors are thought to play a crucial role in the disease development. In some cases the unaffected parents are consanguineous. This makes it is likely that MMIHS has an autosomal recessive pattern of inheritance.

Study objective

Identification of the underlying genes of MMIHS

Study design

Genetic analysis of the DNA of MMIHS patients and family members with homozygosity mapping and exome sequencing

Study burden and risks

not applicable

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

Patient: Diagnosis of megacystis-microlon-intestinal hypoperistalsis syndrome
Family member: Being a family member of a patient with megacystis-microlon-intestinal hypoperistalsis syndrome

Exclusion criteria

Patient: Not a diagnosis of megacystis-microlon-intestinal hypoperistalsis syndrome
Family member: not being a family member of a patient with megacystis-microlon-intestinal

hypoperistalsis syndrome

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)

Primary purpose: Diagnostic

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	01-12-2011
Enrollment:	10
Type:	Actual

Ethics review

Approved WMO Date:	05-10-2011
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Groningen (Groningen)
Approved WMO Date:	13-12-2013
Application type:	Amendment
Review commission:	METC Universitair Medisch Centrum Groningen (Groningen)

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	NL35920.042.11