# Genetics of megacystis-microlonintestinal hypoperistalsis syndrome

Published: 05-10-2011 Last updated: 28-04-2024

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

# 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
Туре:	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** CCMO **ID** NL35920.042.11