Identifying the gene causing Congenital Short Bowel Syndrome

Published: 27-05-2010
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Identification of the causative gene for CSBS.

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

Summary

ID

NL-OMON35033

Source
ToetsingOnline

Brief title
Congenital Short Bowel Syndrome

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Gastrointestinal motility and defaecation conditions

Synonym
Congenital Short Bowel 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: CSBS, Genetics

Outcome measures

Primary outcome

Identification of the causative gene for CSBS

Secondary outcome

Geen

Study description

Background summary

Congenital Short Bowel Syndrome (CSBS) is a rare gastrointestinal disorder. Children are born with a shortened small intestine what leads to severe malabsorption problems. The only described symptomatic treatment is parenteral nutrition. Because this treatment often gives complications, i.e. infections and liver failure, many children die.
In literature around 40 patients have been published. Often siblings are affected and often the parents are consanguineus, this makes a autosomal recessive pattern of inheritence very likely.

Study objective

Identification of the causative gene for CSBS.

Study design

Genetic analysis of the DNA of the families with homozygosity mapping and sequence analysis

Study burden and risks

nvt

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 Congenital Short Bowel Syndrome
Family member: Being a family member of a patient with Congenital Short Bowel Syndrome

Exclusion criteria

Patient: Not a diagnosis of Congenital Short Bowel Syndrome
Family member: not being a family member of a patient with Congenital Short Bowel Syndrome

Study design

Design

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

Primary purpose: Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	14-06-2010
Enrollment:	3
Type:	Actual

Ethics review

Approved WMO

Date:	27-05-2010
Application type:	First submission
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	NL31708.042.10