Genetics of Hirschsprung's disease

Published: 26-05-2011
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Idenification of new Hirschsprung's disease associated genes.

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

Summary

ID

NL-OMON36672

Source
ToetsingOnline

Brief title
Genetics of Hirschsprung's disease

Condition

- Gastrointestinal tract disorders congenital
- Gastrointestinal motility and defaecation conditions

Synonym
Hirschsprung's disease; congenital aganglionosis of the colon

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Groningen
Source(s) of monetary or material Support: Top-subsidie

Intervention

Keyword: exome sequencing, Genetics, Hirschsprung

Outcome measures

Primary outcome

Identification of new Hirschsprung's disease associated genes.

Secondary outcome

Not applicable

Study description

Background summary

Hirschsprung*s disease is a congenital anomaly characterized by the absence of enteric ganglia along a variable length of the bowel. This causes functional obstruction which results in severe constipation and/or megacolon proximal of the aganglionic segment. The current treatment is surgery. Until now around 10 disease associated genes and loci are known which do explain approximately 20/25% of all Hirschsprung cases.

Study objective

Idenification of new Hirschsprung's disease associated genes.

Study design

Genetic analysis of DNA of Hirschsprung patients with 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: having Hirschsprung*s disease.
Family member: not having Hirschsprung*s disease and having a family member (first degree relative) with Hirschsprung*s disease.

Exclusion criteria

Patient: not having Hirschsprung*s disease.
Family member: not having a family member (first degree relative) with Hirschsprung*s disease.

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:	Completed
Start date (anticipated):	01-12-2011
Enrollment:	60
Type:	Actual

Ethics review

Approved WMO Date:	26-05-2011
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	NL35251.042.11