Molecular Basis of Robin Sequence (RS)

Published: 31-07-2013
Last updated: 24-04-2024

1. Identification of gene(s) causing RS. 2. Understanding of the molecular and cellular mechanisms leading to the various manifestations of RS.3. Enabling molecular diagnostics for RS.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Congenital and hereditary disorders NEC
Study type	Observational invasive

Summary

ID

NL-OMON40532

Source
ToetsingOnline

Brief title
Molecular Basis of Robin Sequence (RS)

Condition

- Congenital and hereditary disorders NEC
- Head and neck therapeutic procedures

Synonym
Robin Sequence

Research involving
Human

Sponsors and support

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

Intervention

Keyword: Genetics, Molecular, Robin, Sequence

Outcome measures

Primary outcome

The genetic cause(s) for isolated and syndromic RS.

Secondary outcome

Understanding the pathogenesis of RS.

Study description

Background summary

Robin Sequence (RS) is a rare condition characterized by micro- or retrognathia, cleft palate and respiratory insufficiency. RS is clinically characterized by varying degrees of airway obstruction, causing respiratory insufficiency and feeding difficulties. No consensus has been reached in finding the best management plan for these children. Often conservative treatment consisting of prone positioning, continuous positive airway pressure or placement of a nasopharyngeal airway is sufficient, but sometimes a surgical intervention like a tongue-lip adhesion/glossopexia, mandibular distraction or a tracheostomy is needed.
Different hypothesis have been made to explain the nature of RS such as a mechanic restraint in utero preventing the mandible to grow, intrinsic mandibular malformation, or a neurological immaturity causing involuntary control of the tongue. None of these however are confirmed by evidence based research. Different genes have been suggested to be involved in RS, but none was shown to cause the complete RS phenotype.

Study objective

1. Identification of gene(s) causing RS.
2. Understanding of the molecular and cellular mechanisms leading to the various manifestations of RS.
3. Enabling molecular diagnostics for RS.

Study design

This is a non-therapeutic observational study conducted in the Academic Medical Centre outpatient clinic performed during 1year.

Study burden and risks

The risk of blood sampling is limited. There may be a benefit for patients who will be diagnosed with RS in the future as the diagnosis may be confirmed molecularly more easily but there is no benefit for the present participants. Knowing the cause may lead to adequate genetic counselling and in the future to more effective treatment. The study cannot be performed only in adults as growth of the mandible cannot be studied in adults; and the number of adults with RS is very small. Therefore we find it acceptable to perform the study in children.

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

Group I: all patients (Age 0-17 years) with RS known in the AMC, Amsterdam
Group II: the parents of participants in group I.
*Patients and their parents able to read and understand the written information
*Informed consent signed

Exclusion criteria

None

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	07-01-2014
Enrollment:	180
Type:	Actual

Ethics review

Approved WMO Date:	31-07-2013
Application type:	First submission
Review commission:	METC Amsterdam UMC
Approved WMO Date:	20-06-2014

Application type:	Amendment
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	NL44491.018.13