Study of phenotype in Silver-Russell syndrome patients with known molecular abnormalities

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Inventarisation of the different physical characteristics of SRS at different ages. In this way, differences in phenotype between the 2 diferent causes of SRS will become apparent. Also, with this knowledge, more rpecise and elaborate information...

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

Summary

ID

NL-OMON32891

Source
ToetsingOnline

Brief title
Silver-Russell syndrome

Condition

- Congenital and hereditary disorders NEC

Synonym
Silver-Russell syndrome; growth failure type Silver-Russell

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Child Growth Foundation (United Kingdom)

Intervention

Keyword: genetic, phenotype, Silver-Russell syndrome

Outcome measures

Primary outcome

growth; phsyical characteristics

Secondary outcome

other health problems

Study description

Background summary

Silver-Russell syndroom (SRS) is a rare hereditary condition, resulting in short stature, certain physical characteristics and asymmetries. two improtant genetic causes exist: hypomethylation of th H19 region; and uniparental disomy of chromosme 7.

Study objective

Inventarisation of the different physical characteristics of SRS at different ages. In this way, differences in phenotype between the 2 diferent causes of SRS will become apparent. Also, with this knowledge, more rpecise and elaborate information about SRS can be given to patients and parents of children with the diagnosis SRS.

Study design

children and adults known with a genetic abnormality resulting in SRS will be appraoched by contacting their doctor, who will send them a letter and information with the question, if the patient/parents are willing to cooperate with the study. The stduy itself encompasses, for the adult or child with SRS, answering questions about their own health and a physical (external only) medical examination in the AMC or a hopsital more closeby.

Study burden and risks

burden: low

risk" none

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

Silver-Russell syndrome, proven by DNA analysis (either uniparental disomy 7 or hypomethylation H19)

Exclusion criteria

non-cooperation of patient or parents

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-09-2008
Enrollment:	15
Type:	Anticipated

Ethics review

Approved WMO

Application type:	First submission
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	NL24466.018.08