Molecular basis of Pierpont syndrome

Published: 19-07-2013
Last updated: 22-04-2024

1,detection of the gene causing Pierpont syndrome2. study of the molecular and cellular mechanisms leading to the various manifestations of Pierpont syndrome3. better understanding of the regulation of subcutaneous fat depositions

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

Summary

ID

NL-OMON38587

Source
ToetsingOnline

Brief title
Molecular basis of Pierpont syndrome

Condition

- Congenital and hereditary disorders NEC

Synonym
Pierpont syndrome; lipomatosis and mental retardation

Research involving
Human

Sponsors and support

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

Intervention

Keyword: etiology, functional analysis, molecular research, Pierpont syndrome

Outcome measures

Primary outcome

Detection of the gene causing Pierpont syndrome.

Secondary outcome

- Understanding the molecular and cellular mechanisms leading to the various

manifestations of Pierpont syndrome.

- better understanding of the regulation of subcutaneous fatdepositions

Study description

Background summary

Pierpont syndrome is an entity characterized by marked intellectual disability, unusual face, and abnormal fat depositions. Affected individuals develop epilepsy and show a progressively difficult behaviour. A single boy had a brain tumour as a young child. There is at present no therapy to influence the natural history of the entity. The cause is unknown and we assume it is an autosomal dominant de novo mutation.

Study objective

1,detection of the gene causing Pierpont syndrome
2. study of the molecular and cellular mechanisms leading to the various manifestations of Pierpont syndrome
3. better understanding of the regulation of subcutaneous fat depositions

Study design

Whole exome sequencing of 3 persons with the clinical diagnosis Pierpont syndrome, and of one of them also the parents for trio analysis (vcomparison of variants in results of child with those in parents); if results provide insufficient information whole exome sequencing will also be performed in the parents of the two other patients.

Study burden and risks

The risk of blood sampling is limited. There is no benefit of participating in

this study to the participants themselves but there is a group benefit as the study should provide essential information needed for adequate genetic counselling and future interventions with respect to epilepsy and behaviour.

Contacts

Public
Academisch Medisch Centrum

Meibergdreef 9
Amsterdam 1105AZ
NL
Scientific
Academisch Medisch Centrum

Meibergdreef 9
Amsterdam 1105AZ
NL

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

Patients: clinical diagnosis Pierpont syndrome
Parents: having a child with clinically diagnosed Pierpont syndrome; able to read and understand the written information

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):	03-09-2013
Enrollment:	11
Type:	Actual

Ethics review

Approved WMO

Date:	19-07-2013
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	NL45117.018.13