Phenotyping X-linked adrenoleukodystrophy (X-ALD) in women: a cohort study

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1. What proportion of X-ALD carriers have symptoms en what are those symptoms (as evaluated by history, neurologic examination and electrophysiology)?2. At what age did symptoms fist manifest themselves and how is the progression of those symptoms?3...

Ethical review Approved WMO

Status Pending

Health condition type Metabolic and nutritional disorders congenital

Study type Observational invasive

Summary

ID

NL-OMON31235

Source

ToetsingOnline

Brief title

Phenotyping X-linked adrenoleukodystrophy (X-ALD) in women

Condition

- Metabolic and nutritional disorders congenital
- Spinal cord and nerve root disorders

Synonym

Schilder's disease, X-linked adrenoleukodystrophy

Research involving

Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum

Source(s) of monetary or material Support: Ministerie van OC&W

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Intervention

Keyword: adrenoleukodystrophy, carrier, heterozygote, X-ALD

Outcome measures

Primary outcome

- 1. Symptoms found by (neurologic) history and physical examination
- 2. Score on SF-36 and ALDS
- 3. Electrophysiological studies (EMG, SSEP, BAEP)

Secondary outcome

- 1. Biochemical parameters (plasma C26:0, ALDP expression in fibroblasts)
- 2. X-inactivation

Study description

Background summary

X-linked adrenoleukodystrophy is a metabolic disorder characterised by impaired peroxisomal beta-oxidation of very long chain fatty acids (>C22:0). These fatty acids accumulate in plasma and tissues of patients. Clinical presentation and progression of the disease has been well decribed for male patients over the last decades. For a long time it was assumed that carriers would develop no or only mild symptoms, as is the case in many X-linked diseases. However, there are carriers with severe and disabling symptoms. How many carriers develop symptoms and how severe these symptoms are has not yet been systematically studied.

Study objective

- 1. What proportion of X-ALD carriers have symptoms en what are those symptoms (as evaluated by history, neurologic examination and electrophysiology)?
- 2. At what age did symptoms fist manifest themselves and how is the progression of those symtpoms?
- 3. Are there differences in biochemical (C26:0 in plasma, ALDP expression) or genetic (X-inactivation) between symptomatic and asymptomatic X-ALD carriers?

Study design

Observational cohort study

Study burden and risks

Participation entails one visit to the hospital for part of a day (approximately 4 hours) for history/physical examination, blood sample/skin biopsy and electrophysiological studies. Questionaires must be completed prior to this visit (SF-36, ALDS), which will usually take less than 30 minutes.

Contacts

Public

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age

Adults (18-64 years) Elderly (65 years and older)

Inclusion criteria

- Age > 18 years
- Carrier for X-ALD (either proven by ABCD1 gene mutation analysis or obligate carrier on basis of family history)

Exclusion criteria

- Age < 18 years
- Unable to visit hospital or to perform necessary tasks (questionaires, etc)

Study design

Design

Study type: Observational invasive

Masking: Open (masking not used)

Control: Uncontrolled

Primary purpose: Basic science

Recruitment

NL

Recruitment status: Pending

Start date (anticipated): 01-05-2007

Enrollment: 40

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 NL17119.018.07