# Sjögren-Larsson Syndrome: clinical and biochemical studies

No registrations found.

| | |
|---|---|
| **Ethical review** | Positive opinion |
| **Status** | Pending |
| **Health condition type** | - |
| **Study type** | Observational non invasive |

## Summary

### ID

NL-OMON22068

**Source**
Nationaal Trial Register

**Brief title**
Sjögren-Larsson Syndrome

**Health condition**

Sjögren-Larsson Syndrome (SLS)

### Sponsors and support

**Primary sponsor:** Radboudumc
**Source(s) of monetary or material Support:** N/A

### Intervention

### Outcome measures

**Primary outcome**

- To find out more about biochemical abnormalities in SLS patients using targeted and untargeted metabolomics

- To study neuro-retinal changes over time to learn more about involved neurons and pathomechanism of ophthalmologic abnormalities in SLS patients

### Secondary outcome

N/A

## Study description

### Background summary

Rationale: Sjögren-Larsson Syndrome (SLS) is an autosomal recessive inherited disorder, with a clinical triad of intellectual disability, spastic di- or tetraplegia and ichthyosis. This syndrome is caused by a deficient microsomal fatty aldehyde dehydrogenase (FALDH). FALDH is part of the fatty alchohol nicotinamide adenine dinucleotide (NAD) oxidoreductase complex (FAO) and catalyzes oxidation of many different medium- and long-chain fatty aldehydes into fatty acids.

Deficiency results in the accumulation of fatty aldehydes and fatty alcohols in body fluids and tissues, which is considered the principal causative mechanism leading to the overall clinical phenotype of SLS. The FALDH gene, named ALDH3A2, is located on gene 17p11.2, and mutations in this gene have been identified in SLS patients. Our research group already did several studies in this patient group. With new techniques we would like to find out more about the biochemical abnormalities. Further elucidation of the underlying (biochemical) mechanisms of disease, especially the identification of affected pathways and involved lipid species, potentially leads to the development of novel therapeutic strategies. With new ophthalmologic diagnostic techniques, we aim to get a completer image of ophthalmologic abnormalities in this group.

Objectives:

1. To further unravel biochemical abnormalities in SLS patients

2. To study neuro-retinal changes over time to learn more about involved neurons and pathomechanism of ophthalmologic abnormalities in SLS patients

Study design: Monocenter, interdisciplinary, prospectivecross-sectional, observational cohort study

Study population: Patients with genetically proven Sjögren-Larsson Syndrome in the Netherlands

Main study parameters/endpoints:

1. With targeted and untargeted metabolomics, we aim to determine which lipids accumulate in
SLS patients.

2. With application of novel non-invasive imaging techniques, combined with normal ophthalmologic outpatient visits, we aim to learn more about the structure of the retina and the
abnormalities in the retina.

**Study design**

One-off measuring moment where patients come to the ophtalmology policlinic in the Radboud UMC for eye examination. On the same day blood and urine will be collected for metabolic research.

**Intervention**

Observational cohort study, ophthalmologic and metabolic studies

# Contacts

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# Eligibility criteria

### Inclusion criteria

genetically confirmed SLS patient

### Exclusion criteria

not genetically confirmed SLS patient

## Study design

### Design

| | |
|---|---|
| Study type: | Observational non invasive |
| Intervention model: | Other |
| Allocation: | Non controlled trial |
| Masking: | Open (masking not used) |
| Control: | N/A , unknown |

### Recruitment

NL

| | |
|---|---|
| Recruitment status: | Pending |
| Start date (anticipated): | 15-02-2017 |
| Enrollment: | 25 |
| Type: | Anticipated |

## Ethics review

Positive opinion

| | |
|---|---|
| Date: | 08-02-2017 |
| Application type: | First submission |

## Study registrations

## Followed up by the following (possibly more current) registration

ID: 45720
Bron: ToetsingOnline
Titel:

## Other (possibly less up-to-date) registrations in this register

No registrations found.

## In other registers

| Register | ID |
|---|---|
| NTR-new | NL6138 |
| NTR-old | NTR6277 |
| CCMO | NL58544.091.16 |
| OMON | NL-OMON45720 |

# Study results