

# Changes in microbiome in patients with Ichthyosis Vulgaris.

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<b>Ethical review</b>	Approved WMO
<b>Status</b>	Recruiting
<b>Health condition type</b>	Cornification and dystrophic skin disorders
<b>Study type</b>	Observational invasive

## Summary

### ID

NL-OMON36870

### Source

ToetsingOnline

### Brief title

Changes in microbiome in Ichthyosis Vulgaris.

### Condition

- Cornification and dystrophic skin disorders

### Synonym

Fish scale disease, Ichthyosis Vulgaris

### Research involving

Human

### Sponsors and support

**Primary sponsor:** Universitair Medisch Centrum Sint Radboud

**Source(s) of monetary or material Support:** ZonMw

## Intervention

**Keyword:** Changes, Epidermis, Ichthyosis Vulgaris, Microbiome

## Outcome measures

### Primary outcome

Systematic analysis of the microbiome of patients with ichthyosis vulgaris and comparison with controls and first degree relatives.

### Secondary outcome

Creating a cell bank of ichthyosis vulgaris patients for in vitro studies.

## Study description

### Background summary

Ichthyosis vulgaris is the most common genetic cause of a disturbed keratinisation process of the skin. Loss-of-function mutations in the gene encoding for filaggrin causes ichthyosis vulgaris. The exact mechanism in which these mutations lead to development of ichthyosis vulgaris is not entirely clear.

60-70% of patients with ichthyosis vulgaris also suffer from atopic dermatitis. Recent research has shown that filaggrin mutations are responsible for 10-15% of cases of atopic dermatitis. Both diseases have in common that they are associated with a change in barrier function of the skin.

It is known that microbes have the potential to make changes in the barrier function of the skin. Recent developments in molecular biology made it possible to analyse the microbiome on human skin. Several skin diseases are associated with changes in the microbiome. Analysis of the microbiome of patients with atopic dermatitis and psoriasis have already been done. The relation between atopic dermatitis and filaggrin mutations has not yet been investigated. Possibly, there exists an association between the microbiome and ichthyosis vulgaris. Studies in mice already gave clues for such an association, as changes in microbiome were observed in mice with a matriptase-deficiency (a protease responsible for the conversion of pro-filaggrin to filaggrin.) In this research we want to collect material to investigate this hypothesis.

### Study objective

The objective of this study is to analyse the microbiome of the skin of patients with ichthyosis vulgaris (FLG -/-). This microbiome will be compared to the microbiome of healthy volunteers (FLG +/+) and first grade family members of patients with ichthyosis vulgaris (FLG +/-).

We hope to get clues of the role of micro-organisms in this disease. Insights in the variations in microbiome in patients with ichthyosis vulgaris and atopic dermatitis can contribute to the development of more specific targeted treatments.

Also, we want to collect keratinocytes of patients with ichthyosis vulgaris. These collections can contribute to further research in the skin barrier function.

## **Study design**

This is a small scale study with a pilot character. This study can lead to a bigger cohort study, for example in patients with atopic dermatitis. We want to approach 50 patients with ichthyosis vulgaris to collect material for genetic and celbiologic research. This material will consist of collection of sputum, a swab of the throat, skinswabs and 5 biopsies. This material will be analysed by means of histologic investigation, DNA isolation, genotyping with PCR and gel-electrophoresis, immunohistochemic research and microbiome analysis.

The study will consist of the following groups:

- 1: Healthy volunteers (n=10)
- 2: Patients with ichthyosis vulgaris (n=50 approached, n=10 for final analysis)
- 3: First grade familiemembers of patients with ichthyosis vulgaris (n=10)

Analysis:

1. Genotyping of FLG status (Sputum DNA)
2. Histologic confirmation of diagnosis (Biopsies of Ichthyosis vulgaris patients)
3. Microbiome analysis of skin and throat (Patients with Ichthyosis vulgaris and their first grade family members)
4. Epidermal keratinocytes will be cultured for in vitro 3D skin cultures.

Group 1

Cells and DNA of this group is already in our database.

Group 2 and 3

Volunteers are recruited from our list and the list of patients and the list of the Canisius Wilhelmina Hospital . Patients will receive an information letter at home along with a declaration paper. A week later we will call the patient and ask if he/she is willing to participate. If the patient is willing to participate, he/she is invited in the UMC. St. Radboud. In the hospital an anamnesis will be taken. Material will be collected in form of collection of sputum, skin swabs, throat swabs and five 3 mm skin biopsies. DNA-analysis will

be performed according to standard methods.

Patients will receive a travel fee based on use of public transportation and a fee for collection of materials.

### **Study burden and risks**

Patients will be asked to attend at the UMC St. Radboud. Here, the examinations will take about 1-2 hours. Materials that will be collected consist of: 5 skin biopsies, skin swab, a swab of the throat and sputum.

Biopsies can cause pain and discomfort and can cause scarring. Risk of infection is minimal.

## **Contacts**

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### **Scientific**

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## **Trial sites**

### **Listed location countries**

Netherlands

## **Eligibility criteria**

### **Age**

Adults (18-64 years)

Elderly (65 years and older)

## Inclusion criteria

Patients with Ichthyosis vulgaris, 18-65 years of age. First grade family members of patients with Ichthyosis Vulgaris, 18-65 years of age

## Exclusion criteria

<18 or >65 years of age. Exclusioncriteria will be conform the exclusioncriteria of the human microbiome project.

## Study design

### Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

### Recruitment

NL	
Recruitment status:	Recruiting
Start date (anticipated):	08-01-2013
Enrollment:	40
Type:	Actual

## Ethics review

Approved WMO	
Date:	30-11-2012
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

## 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	NL41569.091.12