Changes in microbiome in patients with Ichthyosis Vulgaris.

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Ethical review	Approved WMO
Status	Recruiting
Health condition type	Cornification and dystrophic skin disorders
Study type	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 keratinisationproces of the skin. Loss-of-function mutations in the gene encoding for filaggrine 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 disease 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 a association between the microbiome and ichthyosis vulgaris. Studies in mice already gave clues for such an association, as changes in microbiome where observed in mices with a matriptase-deficiency (a protease responsible for the conversion of profilaggrin 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-electroforesis, 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 recruted 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 recieve a travelfee 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 throath and sputum.
Biopsies can cause pain and discomfort and can cause scarring. Risk of infection is minimal.

Contacts

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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