Identification of White sponge nevus syndrome.

Published: 03-10-2012
Last updated: 26-04-2024

We would like to report a case of an extra-oral manifestation of a White Sponge Nevus (WSN). The purpose of this study was to document a family spanning 6 generations with WSN. It is important to recognize to allow for appropriate genetic counseling...

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON37321

Source
ToetsingOnline

Brief title
White Sponge Naevus syndrome

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Cornification and dystrophic skin disorders

Synonym
Cannon's disease, WSN

Research involving
Human

Sponsors and support

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

Intervention

Keyword: autosomal dominant, Keratin 13, Vulvar leucoplacie, White Sponge Naevus syndrome, WSN

Outcome measures

Primary outcome

The primary study parameter is a mutation in the keratinine 13-gene.

To document phenotypical signs of of soft, white and spongy plaques in the

(oral) mucosa, or extra-oral lesions.

DNA-test can also be offered to subjects without phenotyipical signs of WSN.

Secondary outcome

not applicable.

Study description

Background summary

A search of gynecological and literature revealed very little about white sponge naevus in the genital area. This disease was first reported by Hyde in 1909, but the term *white sponge naevus* was detailed in 1935 by Cannon. White sponge naevus is a rare, benign, autosomal dominant disorder, with no sex predilection. It predominantly affects nonkeratinizing stratified epithelia such as the oral mucosa. However, less frequently it has reported to rarely involve extra-oral mucosal sites including the genital, laryngeal and esophageal mucosal.
Proper evaluation for a correct diagnosis is of utmost importance. Clinical and family history can be helpful in contributing to the correct diagnosis of WSN.

Study objective

We would like to report a case of an extra-oral manifestation of a White Sponge Nevus (WSN). The purpose of this study was to document a family spanning 6 generations with WSN.
It is important to recognize to allow for appropriate genetic counseling of this autosomal dominant disorder and to avoid misdiagnosis leading to ineffective treatment or potentially adverse side effects of unnecessary

medications and patient dissatisfaction over failure to achieve a therapeutic response.

To objectify if these family members with WSN have a DNA-mutation in the keratinine 13-gene (KRT13).

It is uncertain if WSN is premalignant. Is it necessary to perform a yearly routine checkup to detect an early stage of malignancy?

Study design

Family members will be informed about the possibility of predicitve DNA-research through letters.
Information about the opportunity to take part in this study will be given.

To conduct this rsearch:
blood will be aquired for research after written permission. If the subject does not want to know the result, the results will not be announced to them or their other treating physicians. The result of the DNA-test will be send to dr. FJ Hes. There will be no costs for participating in this study. Subjects can leave the study at any time for any reason if they wish to do so without any consequences. The DNA may, if desired, be destroyed.

To collect clinical information:
after written permission of the family members clinical information will be acquired through an informational letter used in the clinical genetic practice.

Mucous membane tissue:
a consultation will take place at the gynaecologist (prof. dr. Peters) if the subject gives permission. A cervix smear can be done if the subject agrees on it. A consultation will tak place at the dermatologist (prof. dr. Vermeer) to inspect the oral cavity on mucous abnormalities.
The subject will be asked for permission to take professional pictures from the mucous abnormalities and to publish the pictures for scientific purpose. The subject will remain anonymous on the picture.
Clinical information and tissue material of previous examinations will be requested after written permission of the subject.

The DNA-test of the KRT-13 will be supervised by prof. dr. Bakker, head of the department of laboratory diagnostic genome analysis. (LDGA). The consultations will be performed onder supervision of dr. Hes.

Collaborator who may be consulted by the subjects and who is not involved in the implementation of the protocol: dr. CJ. van Asperen, department of Clinical Genetics, LUMC.

Study burden and risks

The subjects will receive a questionnaire and an invitation for an informative meeting. If they give permission to participate in this study, they will be asked to withdraw a sample of blood. Inspection of the oral cavity will be performed. Cervix smear usually takes place during screening. A consultation and a pap smear test at the gynaecologist will be offered.
The subject are not at risk.
Given the limited exposure to the subject it seems to us that to conduct in this research is justified.

Contacts

Public
Leids Universitair Medisch Centrum

Albinusdreef 2
2333 ZA Leiden
NL
Scientific
Leids Universitair Medisch Centrum

Albinusdreef 2
2333 ZA Leiden
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

Family members of this family with WSN. Also new subjects with signs of WSN in their family.

Exclusion criteria

Legal incapacity.

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):	10-10-2012
Enrollment:	25
Type:	Actual

Ethics review

Approved WMO Date:	03-10-2012
Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden)
	metc-ldd@lumc.nl

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	NL40694.058.12