

Molecular pathology of skin disease

Published: 23-10-2006

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To elucidate the molecular basis of skin disease.

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Skin and subcutaneous tissue disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON30415

Source

ToetsingOnline

Brief title

Molecular pathology

Condition

- Skin and subcutaneous tissue disorders congenital
- Skin neoplasms malignant and unspecified
- Skin and subcutaneous tissue disorders NEC

Synonym

genodermatoses, hereditary skin disease

Research involving

Human

Sponsors and support

Primary sponsor: Academisch Ziekenhuis Maastricht

Source(s) of monetary or material Support: Ministerie van OC&W,ZonMW,Berliner Stiftung fur Dermatologie;La Roche Research Foundation

Intervention

Keyword: Experimental, Gene, Molecular, Skin

Outcome measures

Primary outcome

Not applicable. This is not a clinical trial.

Secondary outcome

Not applicable

Study description

Background summary

The department of dermatology of the university hospital Maastricht specializes in the diagnosis and treatment of inherited malignant and non-malignant skin disorders, including complex malformation syndromes. We have a state of the art molecular biology laboratory that has facilities for sequencing and imaging.

There are three lines of research that are guided by our clinical work:

1. Functional characterisation of mutations in gap junction proteins;
2. Functional analysis of mutations in lamins A/C and their associated proteins in lipodystrophies and progeroid syndromes
3. Molecular genetic diagnosis and functional analysis of inherited and congenital syndromic and non-syndromic skin disorders

Study objective

To elucidate the molecular basis of skin disease.

Study design

Not applicable, this is not a clinical trial

Study burden and risks

Patient are not subjected to any other procedures outside of the normal diagnostic ones. As such there is no added burden or risk for them.

Contacts

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Scientific

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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

Inherited and/or congenital skin disorder

Exclusion criteria

Refusal to participate in the project
Refusal of parents/legal guardian to grant permission

Study design

Design

Study type: Observational invasive

Masking: Open (masking not used)

Control: Uncontrolled

Primary purpose: Basic science

Recruitment

NL

Recruitment status: Recruiting

Start date (anticipated): 01-01-2007

Enrollment: 100

Type: Actual

Ethics review

Approved WMO

Date: 23-10-2006

Application type: First submission

Review commission: METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)

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

CCMO

ID

NL11251.068.06