Characteristics of Patients with Dystrophic Epidermolysis Bullosa

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UMC Groningen part of research: to recruit patients with Dystrophic Epidermolysis Bullosa, in order to harvest keratinocytes for defining several characteristics of the EB population: antibodies to type VII collagen, presence of type VII NC1 domain...

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

Summary

ID

NL-OMON34434

Source
ToetsingOnline

Brief title
Characteristics of DEB

Condition

- Skin and subcutaneous tissue disorders congenital
- Epidermal and dermal conditions

Synonym
Epidermolysis Bullosa; Butterfly children

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Groningen
Source(s) of monetary or material Support: Afdeling Dermatologie betaalt het onderzoek.

Intervention

Keyword: characteristics, COLVII, Dystrophic Epidermolysis Bullosa, reprogramming cell lines

Outcome measures

Primary outcome

This study is to determine characteristics of a specific population and to

obtain tissue for research purposes. Subject*s participation will end once all

study-related procedures are completed.

See Certification of Human Subjects Approvals, to Alfred T. Lane, MD,

Dermatology Stanford University California USA, approved 11/10/2009 and CIRM

Disease Team Award Research Proposal, Application number DR1-01454

Secondary outcome

Not applicable.

Study description

Background summary

Rationale:
Epidermolysis Bullosa (EB) is a complex family of inherited blistering skin disorders that share abnormalities of the cutaneous basement membrane zone. Children with one subtype lacking normal collagen VII (COL7A1) develop a severe, scarring phenotype called recessive dystrophic epidermolysis bullosa (RDEB). More commonly, patients with the dominant-negative form of collagen VII deficiency, called Dominant Dystrophic Epidermolysis Bullosa (DDEB), develop debilitating blisters and ulcers. Untill now, current therapy for DDEB and RDEB is limited to palliative woundcare.
Goal is to screen subjects with DEB and evaluate the characteristics of the subjects and their cells in order to develop new strategies of therapy. The final goal is to develop a curative treatment for patients with DDEB and RDEB.

Study objective

UMC Groningen part of research: to recruit patients with Dystrophic Epidermolysis Bullosa, in order to harvest keratinocytes for defining several characteristics of the EB population: antibodies to type VII collagen, presence of type VII NC1 domain, incidence of comorbidity, routine laboratory values of this population, accuracy of earlier clinical or biopsy based diagnosis and specific mutation identification from the genetic testing results.
Aim is to use skin cells from the skinbiopsies for use in reprogramming the somatic cells into genetically-corrected iPS cells. This research may be helpful in elucidationg a future treatment for EB.

Study design

Pre-clinical study, exploring the skin cells of a group of 20 adult Dutch DEB - patients, one visit research.

Study burden and risks

Burden and risks:
Burden is minimal and risks are low.
Burden:
- one visit EB-consultation UMC Groningen
- 2 skinbiopsies 6 mm; taking skinbiopsies (local anesthesia) may give some pain;
- photographs of skin
- physical skin examination
Risks:
- after biopsies, the skin may show skin infection (<1%); this is easy to treat with antibiotic ointments and wound dressings the patients are used to.

This trial will characterize a subset of subjects with DEB. This will not provide a direct benefit to these patients, but may prove beneficial to the patient him/herself and to the research community: the results of this research gives knowledge about a possible therapy, that is: corrected autologous epidermal sheets. Because current therapy for DEB is limited to palliative wound care, the benefit generated by this therapy would be immense.

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

1: clinical diagnosis DEB (via IF, EM and DNA)
2: age: 18-85 years
3: except from DEB: good health
4: possibility of adequate communication
5: living in the Netherlands or Belgium (concerning travelling time to UMC Groningen)

Exclusion criteria

1: minors < 18 y
2: not able to communicate
3: medical instability or limiting ability to travel to UMC Groningen
4: DEB affected patients with co morbidity
5: incapacitated adults

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Treatment

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	01-05-2011
Enrollment:	20
Type:	Actual

Ethics review

Approved WMO

Date:	25-08-2010
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Groningen (Groningen)

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
ClinicalTrials.gov	NCTnumber:01019148

Register	ID
CCMO	NL32561.042.10