Recessive dystrophic epidermolysis bullosa: A clinical observational study of the (inner) eye.

Published: 08-12-2016
Last updated: 16-04-2024

Investigate clinical anomalies in the intraocular tissues, especially the accommodation system, in RDEB patients.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Eye disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON43115

Source
ToetsingOnline

Brief title
Clinical Intraocular RDEB

Condition

- Eye disorders congenital
- Ocular structural change, deposit and degeneration NEC

Synonym
Butterfly disorder, cotton wool skin disorder, crystal skin disorder, severe heriditary blister disease

Research involving
Human

Sponsors and support

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

Intervention

Keyword: Clinical, Dystrophic Epidermolysis Bullosa, Intraocular, Type VII Collagen

Outcome measures

Primary outcome

Outcome of standard ophthalmic investigations, supplemented with fundus imaging, Pentacam and 3D-OCT investigations.

Completion of these clinical investigations, documentation of the outcome in the research file, deriving and storing anonimyzed information out of that file, and interpreting the data. Data used for this study will be encrypted and only involved researchers have access to the data.

Secondary outcome

NA

Study description

Background summary

Collagen type VII (Col VII) is an anchoring fibril forming protein that is primarily known from a dermatological point of view. Col VII anchors tissue layers together, for example the dermal-epidermal layers (thus skin). In fact, it has a lot of resemblance with Velcro. In pathological conditions, this anchoring protein is not fully functional, freely available, or even absent. These conditions are known as dystrophic epidermolysis bullosa. In the recessive variant (RDEB), there is no functional Col VII available. Patients are therefore suffering from extensive blistering of their skin and mucosa, leading to recurrent scar tissue formation and deformations. Many patients die before age 35 because of skin cancer or sepsis. RDEB patients have a low quality of life.
It is known that Col VII is present in the superficial layers of the eye (cornea, conjunctiva). Here, the scar formation also leads to deformations and loss of vision (corneal clouding etc.) Recently, Col VII was discovered in the

inner layers of the eye, at the vitreoretinal junction and in the retina. Pilot studies (our group) have also indicated a possible role in the accommodation system, thus the apparatus that modifies the lens shape in order to focus. We therefore expect abnormalities within the eyes of RDEB patients, or, when this is not the case, possible compensation mechanisms. We want to investigate whether intraocular Col VII has the same anchoring role it has in skin and cornea. Donor eyes are not available. Animal models are not necessarily representative for humans. RDEB patients could be investigated relatively easily and non-invasively in order to compare and comprehend the laboratory data from our earlier pilot studies.

Study objective

Investigate clinical anomalies in the intraocular tissues, especially the accommodation system, in RDEB patients.

Study design

A consecutive clinical case series, in an explorative/descriptive study design (no cause & effect).

Study burden and risks

There are no risks in participating. Countermeasures are taken against damaging participants skin if necessary. Previous experiences however, have shown no such skin reactions. Participants can stop at any time. This would have no consequences for their treatment at ophthalmology or elsewhere in the UMCG. Participant could benefit from early detection of ocular anomalies, or, if no such anomalies are found, are set at ease with the knowledge their eyes are functioning normally.

Contacts

Public
Universitair Medisch Centrum Groningen

Hanzeplein 1
Groningen 9713 GZ
NL
Scientific
Universitair Medisch Centrum Groningen

Hanzeplein 1
Groningen 9713 GZ

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

All RDEB patients that are willing and able (above 8 years of age, both genders, foreign UMCG/blister center visitors).

Exclusion criteria

A clinical condition that would render participation undesirable (very ill patients). Very young children that will not (or cannot) keep still during investigations.

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	31-08-2016
Enrollment:	3
Type:	Actual

Ethics review

Approved WMO

Date:	08-12-2016
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
CCMO	NL57005.042.16