Phenotyping and genotyping of retinal dystrophies in the Netherlands.

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

Summary

ID

NL-OMON44665

Source
ToetsingOnline

Brief title
Phenotyping of RD.

Condition

- Eye disorders congenital

Synonym
retinal dystrophy, retinitis pigmentosa

Research involving
Human

Sponsors and support

Primary sponsor: Oogziekenhuis Rotterdam
Source(s) of monetary or material Support: Stichting wetenschappelijk onderzoek Oogziekenhuis (SWOO);Rotterdamse Stichting Blindenbelangen;Landelijke Stichting voor Blinden en Slechtzienden;Stichting Nederlands Oogheelkundig Onderzoek;Stichting Researchfonds Oogheelkunde Nijmegen;Stichting ter Verbetering van het Lot der Blinden

Intervention

Keyword: clinical characteristics, genetics, retinal dystrophy, therapy

Outcome measures

Primary outcome

Extensive clinical characterizations of a large group of retinal dystrophy

patients. Identification of pathologic genetic variants in genes that are known

to, or likely to be involved in retinal dystrophies.

Secondary outcome

n.a.

Study description

Background summary

Retinal dystrophies (RD) represent a group of inherited ophthalmic diseases, which are characterized by dysfunction of, or progressive loss of photoreceptor cells, often accompanied by fundus abnormalities. It represents the most important cause of juvenile blindness in the Western world for which no treatment is currently available. Approximately 115 genes are known to be mutated in these diseases. Together, these mutations cause ~50% of the inherited retinal dystrophies. Knowledge about the genetic causes has improved genetic counselling of the individual patient and enlarged our knowledge about retinal function and dysfunction in retinal dystrophies. It has also led to the first gene therapy trials in patients with RPE65 mutations recently. Now that several types of therapy for retinal dystrophy are being developed, clinical and genetic characterization of retinal dystrophy patients, and discovering new genes associated with these diseases becomes more important.

Study objective

The goal of this study is to collect and clinically characterize a large cohort of patients with hereditary retinal dystrophies in the Netherlands. The study cohort will be used to identify the genetic causes underlying these diseases, and to establish genotype-phenotype correlations. The final goal is to provide a more reliable prognosis to patients, to provide better genetic counselling, to discover new genes associated with retinal dystrophies, and to collect

larger groups of patients carrying a genetic defect in one of the retinal dystrophy genes. The latter will be essential for the implementation of therapeutic strategies in the near future.

Study design

Diagnostic cohort study.

Study burden and risks

It is important that children are entered into the study, since the documentation of the first symptoms and signs of certain retinal disorders will enhance our knowledge on the function and structure of their retina, which will be needed to determine their eligibility for possible therapeutic strategies. Participants do not benefit, risks are considered negligible, procedures are non-invasive and take about 2 hours extra time from patient (and parent). It is anticipated that, in the future, patients with retinal dystrophies will benefit from newly developed therapeutic strategies.

Contacts

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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

One of the following syndromic or non-syndromic retinal dystrophies:
- retinitis pigmentosa,
- Leber congenital amaurosis,
- cone-rod dystrophy,
- cone-dystrophy,
- achromatopsia,
- Stargardt disease,
- choroideremia,
- X-linked juvenile retinoschisis,
- Usher syndrome,
- Bardet Biedl syndrome,
- Best disease,
- retinal dystrophy closely linked to one of those mentioned above.
All modes of inheritance and ages may be considered.

Exclusion criteria

None.

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	18-08-2011
Enrollment:	5100
Type:	Actual

Ethics review

Approved WMO Date:	04-01-2011
Application type:	First submission
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	01-08-2011
Application type:	Amendment
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	15-07-2013
Application type:	Amendment
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	10-10-2013
Application type:	Amendment
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	13-04-2015
Application type:	Amendment
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	28-07-2016

Application type:	Amendment
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	21-07-2017
Application type:	Amendment
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	15-12-2017
Application type:	Amendment
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)

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	NL34152.078.10