Pseudoxanthoma elasticum: a cohort study and design of a registry.

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To collect data in order to create a registry that can be used for research in PXE, partly with an international questionnaire, and to measure PPi, ENPP1 activity, anti-retinal antibodies and cytokines, and retinal calcification in all patients...

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Congenital and hereditary disorders NEC
Study type	Observational invasive

Summary

ID

NL-OMON52835

Source
ToetsingOnline

Brief title
Pseudoxanthoma elasticum: the registry

Condition

- Congenital and hereditary disorders NEC
- Retina, choroid and vitreous haemorrhages and vascular disorders
- Arteriosclerosis, stenosis, vascular insufficiency and necrosis

Synonym
Pseudoxanthoma elasticum, PXE

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Utrecht
Source(s) of monetary or material Support: Vrienden van het UMC

Intervention

Keyword: Pseudoxanthoma elesticum, PXE, Pyrophosphate, Registry

Outcome measures

Primary outcome

Data will be collected for future research for which the purpose is not known

at this time.

Secondary outcome

Plasma PPi, ENPP1 activity, anti-retinal antibodies and cytokines will be

measured in patients included in the PXE registry, progression of retinal

calcification will be measured using late phase ICG angiogram.

Study description

Background summary

Pseudoxanthoma Elasticum (PXE) is a rare, but severe genetic disease affecting elastic fibres in the skin, vasculature and eyes. Patients suffer from vascular morbidity and visual impairment at a relatively young age. Up to now, no preventative treatment exists and little is known on the prognosis of an individual patient. New insights into this disease, e.g. the course of disease, associations between different clinical parameters and possible future treatments, are needed. The aim of this study is to make a registry, where clinical data is collected for research. To improve international collaboration for this rare disease we want add an international questionnaire to this registration.

Recently, low levels of the calcification inhibitor inorganic pyrophosphate (PPi) were shown to be present in PXE patients and PPi and ENPP1 acitvity (the enzym that forms PPi) levels might therefore be predictive for disease onset and progression and level may explain the large variation in severity of the disease. To get more insights into the pathophysiology of PXE a second aim of this study is to measure PPi and ENPP1 activity in patients included in the PXE registry and link this to clinical parameters. Furthermore, there is a need to quantify ophthalmological disease in an earlier stage than the final visual loss, which is a subjective measure. Up to now, it is not possible to measure

the severity of retinal calcification disease. We have made progress in quantifying progression of retinal disease using indocyanine green angiograms (ICGA) and hypothesize that these can be used to monitor ocular disease. Thus, a third aim of this study is to perform ICGA in patients with PXE and to measure progression of retinal calcification.
Lastly, in some patients there is acute vision loss, one hypothesis is that this is cause by an auto-immune phenomenon. Therefore we want to analyse the blood of PXE patients for anti-retinal antibodies and cytokines.

Study objective

To collect data in order to create a registry that can be used for research in PXE, partly with an international questionnaire, and to measure PPi, ENPP1 activity, anti-retinal antibodies and cytokines, and retinal calcification in all patients included in the PXE national registry and link this to other clinical and ophthalmological parameters.

Study design

Longitudinal observational cohort study

Study burden and risks

The burden for patients to participate in this study is minimal. A total of 27 ml extra blood will be collected as much as possible along with blood collection for routine medical care. Furthermore, patients will receive one injection each visit every two years with ICG which is also used in routine ophthalmological care for decades. This will take minimal extra time and has minimal risks. Participation or refusal to participate in the study will neither have consequences for their treatment.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)

Inclusion criteria

- Diagnosed with PXE
- Given informed consent
- Age: 18 years or older

Exclusion criteria

Age under 18

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):	12-03-2019
Enrollment:	500
Type:	Actual

Ethics review

Approved WMO Date:	04-01-2019
Application type:	First submission
Review commission:	METC NedMec
Approved WMO Date:	18-04-2019
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	14-07-2021
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	08-09-2022
Application type:	Amendment
Review commission:	METC NedMec

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	NL67568.041.18