

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

Published: 04-01-2019

Last updated: 12-04-2024

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...

<b>Ethical review</b>	Approved WMO
<b>Status</b>	Recruiting
<b>Health condition type</b>	Congenital and hereditary disorders NEC
<b>Study type</b>	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 elasticum, 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 activity (the enzyme 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 caused 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**

### **Public**

Universitair Medisch Centrum Utrecht

Heidelberglaan 100

Utrecht 3584 CX

NL

### **Scientific**

Universitair Medisch Centrum Utrecht

Heidelberglaan 100

Utrecht 3584 CX

## 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

CCMO

### ID

NL67568.041.18