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# Erythropoietic protoporphyria life impact and genetic health trajectory (EPP LIGHT): a survey of adults and adolescents in Europe living with EPP

**Submission date**
05/02/2026

**Recruitment status**
No longer recruiting

**Registration date**
17/03/2026

**Overall study status**
Completed

**Last Edited**
17/03/2026

**Condition category**
Nutritional, Metabolic, Endocrine

[_] Prospectively registered
[X] Protocol
[_] Statistical analysis plan
[_] Results
[_] Individual participant data
[X] Record updated in last year

**Plain English summary of protocol**
Background and study aims
Erythropoietic Protoporphyria (EPP) and X-linked Protoporphyria (XLP) are rare, inherited conditions that is a result of an accumulation of protoporphyrin (PPIX). The genes affected in EPP and XLP are different, but these conditions cause similar symptoms. People with EPP/XLP are very sensitive to light. Symptoms after sunlight (indirect and direct) exposure include burning, itching, and pain on exposed areas of the skin, and these areas can become red /inflamed and swollen. Additionally, people with EPP/XLP may develop liver complications.

There is limited information about the impact of EPP in Europe. The aim of the EPP LIGHT is to describe the burden associated with EPP/XLP in terms of health-related quality of life (HRQoL), symptoms and healthcare resource utilization (HCU).

Who can participate?
Those who are 12 or older, have a confirmed diagnosis of EPP or XLP, and live in the UK, France, Germany, Italy, or Spain may participate in the study. They also need to be able to speak, read, and write in English, French, German, Italian, Spanish, or Welsh, and be willing and able to complete a single online questionnaire that takes about an hour.

What does the study involve?
The EPP LIGHT study is a one time online survey for people with EPP or XLP. It uses a mix of validated and study specific questions to understand how light sensitivity affects quality of life, symptoms (including early warning signs and pain), healthcare use (doctor or hospital visits and medicines), and daily activities. Adolescent participants answer slightly different age appropriate questions about wellbeing, friends, and school, and everyone is asked some basic background details.

What are the possible benefits and risks of participating?
There is no benefit in taking part in this study. However, the results may help improve the care of people with EPP or XLP in the future.

Where is the study run from?
The study is conducted by Sciensus, a European life sciences organisation based in the United Kingdom.
The survey is hosted onto the Climedo platform whose servers are located in Germany.

When is the study starting and how long is it expected to run for?
Enrolment onto the study is due to start on 1st August 2025 and to complete on 31st October 2025.

Who is funding the study?
Disc Medicine (USA)

Who is the main contact?
Mathieu Loiseau, Lead Evidence Generation and Patient Support, mathieu.loiseau@sciensus.com

# Contact information

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Scientific

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# Additional identifiers

# Study information

### Scientific Title
EPP LIGHT (Life Impact and Genetic Health Trajectory) study: A cross-sectional online survey of adult and adolescent participants with erythropoietic protoporphyria (EPP) in Europe

### Acronym
EPP LIGHT

### Study objectives
To describe the burden associated with EPP in terms of health-related quality of life (HRQoL), symptoms, healthcare resource utilization (HCU) and preference for treatment.

### Ethics approval required
Ethics approval required

### Ethics approval(s)
Approved 30/05/2025, Wales REC 3 (Castlebridge 4 15-19 Cowbridge Road East, Cardiff, CF11 9AB, United Kingdom; -; Wales.REC3@wales.nhs.uk), ref: 25/WA/0165

### Primary study design
Observational

### Secondary study design
Cross sectional study

### Study type(s)

### Health condition(s) or problem(s) studied
Erythropoietic protoporphyria and X-linked protoporphyria (collectively referred to as EPP)

### Interventions
The research consists of an online survey study for which participants will be required to complete a one-off questionnaire. Participants will be identified with via the relevant Patient Advocacy Groups which will share a study flyer/ advertisement. Potential participants interested in taking part will register via a weblink / QR code and screened for eligibility. Once eligibility has been confirmed, participants will receive a unique weblink for completing the questionnaire.

### Intervention Type
Other

### Primary outcome(s)
1. Health-related quality of life (HRQoL), symptoms, healthcare resource utilization (HCU) and preference for treatment measured using survey at a single timepoint

### Key secondary outcome(s)

**Completion date**
14/11/2025

# Eligibility

**Key inclusion criteria**
Confirmed diagnosis of erythropoietic protoporphyria and X-linked protoporphyria

**Healthy volunteers allowed**
No

**Age group**
Mixed

**Lower age limit**
12 Years

**Upper age limit**
120 Years

**Sex**
All

**Total final enrolment**
101

**Key exclusion criteria**
A cognitive or other (visual, hearing) impairment that would interfere with the ability to participate in the study.

**Date of first enrolment**
01/08/2025

**Date of final enrolment**
31/10/2025

# Locations

**Countries of recruitment**
United Kingdom

England

Northern Ireland

Scotland

Wales

France

Germany

Italy

Spain

**Study participating centre**
**Online study**
N/A
N/A
England
N/A

# Sponsor information

**Organisation**
Disc Medicine

# Funder(s)

**Funder type**

**Funder Name**
Disc Medicine

# Results and Publications

**Individual participant data (IPD) sharing plan**

**IPD sharing plan summary**
Not expected to be made available

**Study outputs**

| Output type | Details | Date created | Date added | Peer reviewed? | Patient-facing? |
|---|---|---|---|---|---|
| Protocol file | | 28/05/2025 | 20/02/2026 | No | No |