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Lay Protocol Synopsis

Full study title:	**Natural History and Disease Burden of X-linked Hypohidrotic Ectodermal Dysplasia (XLHED): An Observational, Multicentre, International Study**
Brief title	**EdeReaLife**
Lay language title:	**XLHED Natural History Study: Impact on Patients and Families**
EU CT Number:	**Not applicable**

What is the purpose of the study? (Rationale)

X-Linked Hypohidrotic Ectodermal Dysplasia (XLHED) is a rare genetic condition that affects more severely males, mainly causing issues with sweat glands, teeth, and hair. This condition can significantly impact the daily lives of patients and their families.

The aim of this study is to understand how XLHED affects the lives of young male patients and their families over time. By studying the natural course of the disease and its impact, the study could improve the understanding of the challenges faced by these patients and their families.

In this non-interventional study, no treatment is tested. Instead, the researchers observe and collect information about the health and daily experiences of young male patients with XLHED. They do this by gathering information from medical records and asking parents to fill out questionnaires at different points in time.

What are the objectives of the study and how are they evaluated?

Objectives (Goals)	Endpoints (Measurements to answer researchers'questions)
To describe the natural history and the daily life impact of XLHED on patients and their family.	Researchers: • Describe the demographic characteristics and clinical profile of young male patients with XLHED. • Describe how these patients access healthcare access and their care paths. • Measure how XLHED affects the health-related quality of life (HRQL) of young male patients. • Assess how parents report the impact on the family. • Assess the costs families bear related to managing XLHED and its complications.

How is the study conducted? (Study Design)

This study is an observational study, designed to gather information without changing the usual care of the participants. The study is conducted in two European countries: France and Germany, each with one expert site. Young male patients with XLHED, from birth to just before their 11th birthday, can be included. These patients should not be participating in any other treatment study for XLHED.

The study collects data over a 2-year follow-up period, with assessments at three time points: at the start of the study, 1 year after enrollment, and 2 years after enrollment.

Investigators collect information through electronic forms (eCRFs) completed by healthcare professionals and questionnaires completed by the parents or legal guardians of the patients. The data come from medical records and observations made during routine clinical examinations. No extra tests or procedures are done for this study.

The study aims to describe the natural history and the burden of XLHED on the daily lives of patients and their families. Safety and ethical considerations are strictly followed, and the study is approved by appropriate review boards before any patients are enrolled.

Who can take part in the study? (Study Population)

The study started in July 2023.

Young male patients with X-Linked Hypohidrotic Ectodermal Dysplasia (XLHED) can take part in this study. To be eligible, patients must:

- Be males from birth to just before their 11th birthday.
- Have a confirmed diagnosis of XLHED, either through genetic testing or by showing the typical symptoms (such as issues with sweat glands, teeth, and hair) and having a mother with a confirmed genetic diagnosis of XLHED.
- Have not received any treatment with a specific medicine called ER004 or participated in any studies related to ER004.
- Have their parent(s) or legal guardian(s) agree to the study and provide consent according to the country's regulations.

Patients are not eligible if they:

- Have received any treatment with ER004 or are currently participating in any studies related to ER004.
- Have a negative genetic test for XLHED.
- Have any other medical condition that could interfere with the evaluation of the impact of XLHED.

The study takes place at one expert site in each of two European countries: France and Germany. Parents or legal guardians of patients who attend these expert centers are informed about the study and can decide if they want their child to participate.

What are the study treatments and how are they administered? (Interventions)

This study does not involve any specific treatments or procedures. It is an observational study, designed to gather information about the natural history of XLHED without giving any new treatments. Participants continue to receive their usual care as recommended by their healthcare providers. No new procedures are tested or recommended as part of this study.

Ethical considerations

This study is conducted following the ethical principles of the Declaration of Helsinki and the General Data Protection Regulation (GDPR), as well as other local regulations. This means that the study is done in a way that respects the rights and well-being of the participants.

The study does not involve any changes to the usual care that patients receive, so it does not affect their physical or mental health. Each patient and their parent(s) or legal guardian(s) are fully informed about the study before they decide to participate. Their written consent or verbal agreement is obtained, according to the local regulations of each country.

An independent data monitoring committee or similar review group is not set up, as this is a non-interventional study without any investigational treatments.

What are the possible benefits and risks in taking part in the study? (Benefit /risk)

Possible Benefits:

By participating in this study, the participants will contribute to a better understanding of how XLHED affects the daily lives of young male patients and their families. This information can help improve future care and support for individuals with XLHED.

There are no direct individual benefits expected from participating in this study, as no new treatments or interventions are being tested. However, the study aims to provide group benefits by gaining insights that could help the medical community better understand and manage XLHED in the future.

Possible Risks and Burdens:

The risks and burdens associated with this study are considered minimal. Since this is an observational study, participants do not undergo any additional diagnostic, therapeutic, or monitoring procedures beyond their usual care.

Participants and their caregivers may experience some inconvenience due to the need to complete questionnaires and attend follow-up visits at the expert reference centers. These visits occur at three time points: at the start of the study, 1 year after enrollment, and 2 years after enrollment.