Non-interventional study of patients with Netherton Syndrome to characterise the natural history of disease (Natural history of Netherton Syndrome)

First published: 27/02/2023

Last updated: 03/11/2026

Study | Discontinued

Administrative details

EU PAS number

EUPAS103733

Study ID

106711

DARWIN EU® study

No

Study countries

- Austria
- China
- France

Germany
Italy
Japan
Netherlands
United Kingdom
United States

Study description

Netherton syndrome (NS) is a rare autosomal recessive disorder that manifests as congenital ichthyosis form erythroderma (CIE) at birth or shortly thereafter.
Epidemiological data on NS is scarce with no reliable data on the incidence.
At present, there is no approved therapy for NS from a regulatory perspective.
Effective management of the symptoms of NS requires a multi-disciplinary approach that targets the specific clinical characteristics of individual patients.
In the context of clinical development of spesolimab for NS, a better understanding of the natural history and real-world management and assessment is key to put results from clinical studies in perspective.
It is also important to understand the burden of NS in terms of the management of its complications and comorbidities.
A multi-country study with medical chart extraction and new data collection is necessary to reliably identify patients diagnosed with NS in real-world clinical practice settings and to collect data to describe patient characteristics, disease course, treatment patterns, healthcare resource utilisation and clinical outcomes.
As well, to better understand the factors that lead to the high burden of NS, primary data on clinician- and patient-reported outcomes should be collected using validated instruments that measure status and progression of skin and hair conditions and patient’s health-related quality of life (HRQoL).

Study status

Discontinued

Research institutions and networks

Institutions

United BioSource Corporation (UBC)

Switzerland

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Institution | **Non-Pharmaceutical company** | **ENCePP partner**

Contact details

Study institution contact

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Study contact

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Primary lead investigator

Paula Chakravarti

Primary lead investigator

Study timelines

Date when funding contract was signed

Planned: 28/02/2023

Actual: 28/02/2023

Study start date
Planned: 28/02/2024

Actual: 09/10/2024

Date of final study report
Planned: 31/12/2026

Actual: 31/03/2025

Sources of funding

- Pharmaceutical company and other private sector

More details on funding

Boehringer Ingelheim

Study protocol

Protocol_and_Protocol_Amendment_06_Feb_2026__Redacted.pdf (1.1 MB)

Regulatory

Was the study required by a regulatory body?

No

Is the study required by a Risk Management Plan (RMP)?

Not applicable

Other study registration identification numbers and links

NCT05902663

Methodological aspects

Study type

Study type list

Study topic:

Disease /health condition

Study type:

Non-interventional study

Scope of the study:

Disease epidemiology

Drug utilisation

Evaluation of patient-reported outcomes

Healthcare resource utilisation

Data collection methods:

Combined primary data collection and secondary use of data

Study design:

Retrospective and prospective cohort

Main study objective:

Primary objective: To assess severity of NS by the Ichthyosis Area Severity Index (IASI). Secondary objective: To assess severity of NS by the Investigator

Global Assessment – Netherton Syndrome (IGA-NS).

Study Design

Non-interventional study design

Cohort

Study drug and medical condition

Medical condition to be studied

Netherton's syndrome

Population studied

Short description of the study population

Patients with Netherton Syndrome

Inclusion criteria [for Part 1 and Part 2]

1. Confirmed diagnosis of NS by at least one of the following:

Genetic testing of mutations in Serine Protease Inhibitor of Kazal Type 5 (SPINK5);

Absence or major deficiency of the protein Lympho-Epithelial Kazal-Type-Related Inhibitor (LEKTI) inskin biopsy;

Clinical assessment (signs and symptoms).

2. Provision of consent or assent (i.e., by parent or legal guardian) as required by local regulations:

[Part 1] to authorise access to existing medical records for study data

collection;
[Part 2] to participate in the longitudinal 52-week evaluation of disease severity and clinical outcome assessments.
[for Part 2 only] 3. Not participating in a clinical trial at the time of study enrolment for Part 2.
Exclusion criteria [for Part 1 and Part 2]
1. Deceased patients and patients whose survival status are not known, who were diagnosed prior to 2002.

Age groups

- Term newborn infants (0 – 27 days)
- Infants and toddlers (28 days – 23 months)
- Children (2 to < 12 years)
- Adolescents (12 to < 18 years)
- **Adult and elderly population (≥18 years)**
  - Adults (18 to < 46 years)
  - Adults (46 to < 65 years)
  - Elderly (≥ 65 years)
    - Adults (65 to < 75 years)
    - Adults (75 to < 85 years)
    - Adults (85 years and over)

Estimated number of subjects

100

Study design details

Setting

This study will be conducted in hospitals/clinical centres where patients with NS are typically
managed in real-world clinical practice. The study will include study sites across several countries
in North America, Europe, and Asia.
For practical reasons, site selection will prioritize centres where a higher number of patients can
be enrolled to achieve the target study sample size. Approximately 22 centres will participate in
this study; more sites may be included to achieve the target study sample size.

Comparators

N/A

Outcomes

IASI and its two subscales (IASI-E erythema and IASI-S scaling), Total score of IGA-NS.

Data analysis plan

There is no pre-specified hypothesis to be tested. Data analysis will be descriptive.
Quantitative variables will be summarised by number of non-missing values (N), mean, standard deviation (SD), standard error of the mean (SE), minimum (min), median, and maximum (max).
Categorical variables will be summarised by counts and percentages for each category.
The primary and secondary outcomes will be analysed descriptively on the overall population as well as for any subgroups deemed appropriate and informative.
Annualised incidence of flares will be calculated overall and for the above

subgroups.

Documents

Study results

1368-0121_Ct.gov Submission.pdf (113.34 KB)

Data management

ENCePP Seal

The use of the ENCePP Seal has been discontinued since February 2025. The ENCePP Seal fields are retained in the display mode for transparency but are no longer maintained.

Data sources

Data sources (types)

Electronic healthcare records (EHR)

Other

Data sources (types), other

Prospective patient-based data collection

Use of a Common Data Model (CDM)

CDM mapping

No

Data quality specifications

Check conformance

Unknown

Check completeness

Unknown

Check stability

Unknown

Check logical consistency

Unknown

Data characterisation

Data characterisation conducted

Unknown