Exome Sequencing and Molecular Profiling in Patients with Hidradenitis Suppurativa (HS)

Published: 19-09-2024
Last updated: 18-01-2025

To Identify genetic variants and clinical phenotypes associated with risk of HS and to characterize the key cellular and molecular mechanisms and pathways involved in the pathogenesis of moderate-to-severe HS.

Ethical review	Approved WMO
Status	Pending
Health condition type	Skin appendage conditions
Study type	Observational invasive

Summary

ID

NL-OMON57185

Source
ToetsingOnline

Brief title
HSProGen

Condition

- Skin appendage conditions

Synonym
acne ectopica, Hidradenitis Suppurativa

Research involving
Human

Sponsors and support

Primary sponsor: The European Hidradenitis Supurativa Foundation (EHSF) e.V.
Source(s) of monetary or material Support: Regeneron,The European Hidradenitis Supurativa Foundation (EHSF) e.V.

Intervention

Keyword: exome sequencing, molecular profiling

Outcome measures

Primary outcome

Whole-exome sequencing and/or array genotyping to identify single variants and/or gene-based burden tests significantly associated with HS status and severity.

Secondary outcome

Transcriptional profiling through bulk RNA sequencing.

Study description

Background summary

Despite some progress in understanding and treating Hidradenitis suppurativa (HS), significant improvements in therapeutic options will require a better understanding of HS pathophysiology, particularly the underlying genetic variants that may serve as important drivers of the disease.

Study objective

To Identify genetic variants and clinical phenotypes associated with risk of HS and to characterize the key cellular and molecular mechanisms and pathways involved in the pathogenesis of moderate-to-severe HS.

Study design

An exploratory observational study as part of a Europe wide consortium.

Study burden and risks

All research data will be handled in accordance with the Dutch Data Protection Act and privacy regulations of Erasmus MC. Extra time during the visit is required from participants as they need to fill out a questionnaire and more information is needed from the physician. Furthermore, a swap and (optional)

blood sample is collected. Risks are minimal. A buccal swap is a non-invasive method. Venepuncture has a small risk for pain, minor bleeding, and/or hematoma. The patient will not directly benefit from this research, but participation contributes to increased knowledge about HS and subsequently improving treatment and care.

Contacts

Public
The European Hidradenitis Supurativa Foundation (EHSF) e.V.

Auenweg 38
Dessau 06847
DE
Scientific
The European Hidradenitis Supurativa Foundation (EHSF) e.V.

Auenweg 38
Dessau 06847
DE

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

• Male or female, age 18 years or older at the screening visit
• Patients must have a diagnosis of HS (in compliance with Dessau criteria as modified in San Francisco) for at least one year (365 days) prior the screening visit
• HS lesions must be present in at least two distinct classical anatomic areas

(e.g., left and right axilla; or left axilla and left inguino-crural fold)
• Willing and able to comply with clinic visits and study-related procedures
• Provide informed consent signed by study patient or legally acceptable representative
• Able to understand and complete study-related material

Exclusion criteria

- Presence of skin comorbidities (e.g. inverse psoriasis, epidermal inclusion cyst) that may interfere with study assessments for HS
- Severe concomitant illness(es)
- Pregnant or breastfeeding women

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	15-10-2024
Enrollment:	400
Type:	Anticipated

Ethics review

Approved WMO

Date:	19-09-2024
Application type:	First submission
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)

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	NL84959.078.23