Genetics of hidradenitis suppurativa

Published: 30-08-2013
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To determine which gen defects are associated with hidradenitis suppurativa.

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

Summary

ID

NL-OMON47104

Source
ToetsingOnline

Brief title
Genetics of hidradenitis suppurativa

Condition

- Skin appendage conditions

Synonym
acne inversa, hidradenitis suppurativa

Research involving
Human

Sponsors and support

Primary sponsor: Erasmus MC, Universitair Medisch Centrum Rotterdam
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: ectopic acne, genetics, hidradenitis suppurativa, mutations

Outcome measures

Primary outcome

The identification of pathogenic variants will be carried out at the Erasmus MC Genomics Core Facility of the Department of Internal Medicine, where the data analysis will also be carried out.

The skin collected from surgery will be examined on protein level to see if possible genetic defects are expressed there. This is to verify the relevance of the found genetic abnormalities.

Secondary outcome

N.A.

Study description

Background summary

Hidradenitis suppurativa (HS) is a chronic recurrent inflammatory skin disease affecting apocrine gland-bearing body areas such as axillae, groin and perianal regions Characteristic for this disease are painful, inflammatory nodules and sterile abscesses, followed by tissue fibrosis and sinus tract formation. HS usually develops after puberty and the overall prevalence is estimated around 1% (1). Women are more frequently affected than men (female: male ratio, 3:1). About 85-95% of the patients are active smokers (2). The exact pathogenesis of HS is still unknown. The primary event is thought to be occlusion of the terminal hair follicle caused by infundibular hyperkeratosis and hyperplasia of the follicular epithelium. (3) Bacterial cultures from HS lesions are often sterile or only grow commensal skin flora. (4) The diagnosis is made based on the clinical presentation. Available treatments are limited, antibiotics, retinoids, corticosteroids and cyclosporine can give temporarily relieve. Radical excision with secondary wound healing is the therapy of first choice. (5)

In approximately 40% of the patients, HS occurs in one or more family members. (4) It is hypothesized that in some cases there is an autosomal dominant inheritance with a variable penetrance. (6) To date, several genetic loci have been identified which are associated with HS, but so far no causative genes

have been found. Mutations in the gamma-secretase gens (PSENEN, PSEN1 and NCSTN) were identified in families with multiple family members suffering from a special form of HS. However, how these mutations cause HS is still unknown. (7)

1. Revuz JE, Canoui-Poitrine F, Wolkenstein P, Viallette C, Gabison G, Pouget F, et al. Prevalence and factors associated with hidradenitis suppurativa: results from two case-control studies. Journal of the American Academy of Dermatology. Elsevier; 2008;59(4):596-601.
2. König A, Lehmann C, Rompel R, Happle R. Cigarette smoking as a triggering factor of hidradenitis suppurativa. Dermatology. Karger Publishers; 1999;198(3):261-4.
3. Kurzen H, Kurokawa I, Jemec GBE, Emtestam L, Sellheyer K, Giamarellos-Bourboulis EJ, et al. What causes hidradenitis suppurativa? Experimental dermatology. Copenhagen: Munksgaard, c1992-; 2008;17(5):455-6.
4. Jemec G, Revuz J, Leyden J. Hidradenitis Suppurativa. 1st ed. Heidelberg, Germany: Springer; 2006.
5. Ritz JP, Runkel N, Haier J, Buhr HJ. Extent of surgery and recurrence rate of hidradenitis suppurativa. International journal of colorectal disease. Springer; 1998;13(4):164-8.
6. Von der Werth JM, Williams HC, Raeburn JA. The clinical genetics of hidradenitis suppurativa revisited. British Journal of Dermatology. Wiley Online Library; 2000;142(5):947-53.
7. Wang B, Yang W, Wen W, Sun J, Su B, Liu B, et al. γ-secretase gene mutations in familial acne inversa. Science. American Association for the Advancement of Science; 2010;330(6007):1065.

Study objective

To determine which gen defects are associated with hidradenitis suppurativa.

Study design

The collected blood and saliva are processed at the laboratory of Internal Medicine of Erasmus MC. DNA will be disposed of after sequencing.
The skin collected from surgery will be examined on protein level to see if possible genetic defects are expressed there. This is to verify the relevance of the found genetic abnormalities.

Study burden and risks

From patients visiting the department of dermatology venous blood will be drawn or saliva will be collected. Skin will also be collected from surgery.
The burden and risks for participation are minimal for the patients.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

doctor confirmed diagnosis of hidradenitis suppurativa

Exclusion criteria

none

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):	01-09-2013
Enrollment:	1000
Type:	Anticipated

Ethics review

Approved WMO Date:	30-08-2013
Application type:	First submission
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	10-12-2018
Application type:	Amendment
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	NL45264.078.13