



## Hidradenitis suppurativa

Hidradenitis suppurativa, also known as acne inversa, is a chronic skin disease characterized by recurrent boil-like lumps (nodules) under the skin. The nodules become inflamed and painful. They tend to break open (rupture), causing abscesses that drain fluid and pus. As the abscesses heal, they produce significant scarring of the skin.

The signs and symptoms of hidradenitis suppurativa appear after puberty, usually in a person's teens or twenties. Nodules are most likely to form in the armpits and groin. They may also develop around the anus, on the buttocks, or under the breasts. In some cases, nodules appear in other areas, such as the nape of the neck, waist, and inner thighs.

The recurrent nodules and abscesses cause chronic pain and can lead to self-consciousness, social isolation, and depression. Rarely, nodules on the buttocks can develop into a type of skin cancer called squamous cell carcinoma.

### Frequency

Hidradenitis suppurativa was once thought to be a rare condition because only the most severe cases were reported. However, recent studies have shown that the condition affects at least 1 in 100 people when milder cases are also considered. For reasons that are unclear, women are about twice as likely as men to develop the condition.

### Causes

In most cases, the cause of hidradenitis suppurativa is unknown. The condition probably results from a combination of genetic and environmental factors. Originally, researchers believed that the disorder was caused by the blockage of specialized sweat glands called apocrine glands. However, recent studies have shown that the condition actually begins with a blockage of hair follicles in areas of the body that also contain a high concentration of apocrine glands (such as the armpits and groin). The blocked hair follicles trap bacteria, leading to inflammation and rupture. It remains unclear what initially causes the follicles to become blocked and why the nodules tend to recur.

Genetic factors clearly play a role in causing hidradenitis suppurativa. Some cases have been found to result from mutations in the *NCSTN*, *PSEN1*, or *PSENEN* gene. The proteins produced from these genes are all components of a complex called gamma- ( $\gamma$ ) secretase. This complex cuts apart (cleaves) many different proteins, which is an important step in several chemical signaling pathways. One of these pathways, known as Notch signaling, is essential for the normal maturation and division of hair follicle cells and other types of skin cells. Notch signaling is also involved in normal immune system function. Studies suggest that mutations in the *NCSTN*,

*PSEN1*, or *PSENEN* gene impair Notch signaling in hair follicles. Although little is known about the mechanism, abnormal Notch signaling appears to promote the development of nodules and lead to inflammation in the skin. Researchers are working to determine whether additional genes, particularly those that provide instructions for making other  $\gamma$ -secretase components, are also associated with hidradenitis suppurativa.

Researchers have studied many other possible risk factors for hidradenitis suppurativa. Obesity and smoking both appear to increase the risk of the disorder, and obesity is also associated with increased severity of signs and symptoms in affected individuals. Studies suggest that neither abnormal immune system function nor hormonal factors play a significant role in causing the disease. Other factors that were mistakenly thought to be associated with this condition include poor hygiene, the use of underarm deodorants and antiperspirants, and shaving or the use of depilatory products to remove hair.

## **Inheritance Pattern**

Hidradenitis suppurativa has been reported to run in families. Studies have found that 30 to 40 percent of affected individuals have at least one family member with the disorder. However, this finding may be an underestimate because affected individuals do not always tell their family members that they have the condition, and hidradenitis suppurativa is sometimes misdiagnosed as other skin disorders.

In some families, including those with an *NCSTN*, *PSEN1*, or *PSENEN* gene mutation, hidradenitis suppurativa appears to have an autosomal dominant pattern of inheritance. Autosomal dominant inheritance means one copy of an altered gene in each cell is sufficient to cause the disorder. In many cases, an affected person inherits the altered gene from a parent who has the condition.

## **Other Names for This Condition**

- acne inversa
- hidradenitides, suppurative
- hidradenitis, suppurative
- suppurative hidradenitides
- suppurative hidradenitis

## **Diagnosis & Management**

### Genetic Testing Information

- What is genetic testing?  
[/primer/testing/geneticTesting](#)
- Genetic Testing Registry: Hidradenitis suppurativa, familial  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1840560/>

### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22hidradenitis+suppurativa%22+OR+%22acne+inversa%22>

### Other Diagnosis and Management Resources

- American Academy of Dermatology: Hidradenitis Suppurativa: Diagnosis, Treatment, and Outcome  
<https://www.aad.org/public/diseases/painful-skin-joints/hidradenitis-suppurativa>

## **Additional Information & Resources**

### Health Information from MedlinePlus

- Encyclopedia: Hair Follicle Anatomy (image)  
<https://medlineplus.gov/ency/imagepages/9703.htm>
- Health Topic: Hidradenitis Suppurativa  
<https://medlineplus.gov/hidradenitissuppurativa.html>

### Genetic and Rare Diseases Information Center

- Hidradenitis suppurativa  
<https://rarediseases.info.nih.gov/diseases/6658/hidradenitis-suppurativa>

### Educational Resources

- British Association of Dermatologists  
<http://www.bad.org.uk/for-the-public/patient-information-leaflets/hidradenitis-suppurativa#.Uzlt8VcjTHk>
- MalaCards: hidradenitis suppurativa  
[https://www.malacards.org/card/hidradenitis\\_suppurativa](https://www.malacards.org/card/hidradenitis_suppurativa)
- Orphanet: NON RARE IN EUROPE: Hidradenitis suppurativa  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=387](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=387)

### Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/hidradenitis-suppurativa/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Hidradenitis+Suppurativa%5BMAJR%5D%29+AND+%28%28hidradenitis+suppurativa%5BTIAB%5D%29+OR+%28acne+inversa%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- ACNE INVERSA, FAMILIAL, 1  
<http://omim.org/entry/142690>
- ACNE INVERSA, FAMILIAL, 2, WITH OR WITHOUT DOWLING-DEGOS DISEASE  
<http://omim.org/entry/613736>
- ACNE INVERSA, FAMILIAL, 3  
<http://omim.org/entry/613737>

### Medical Genetics Database from MedGen

- Hidradenitis suppurativa, familial  
<https://www.ncbi.nlm.nih.gov/medgen/326766>

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