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## Netherton syndrome

### What is Netherton syndrome?

Netherton syndrome is a rare inherited disorder that presents with the three following characteristics:

- Ichthyosiform erythroderma – inflamed, red, scaly skin
- Trichorrhexis invaginata (“bamboo hair”) – short, brittle, lustreless hair
- Atopic diathesis – predisposition to allergy problems

Individuals with Netherton syndrome may show some or all of these features with varying degrees of severity of their symptoms.

### What are the signs and symptoms of Netherton syndrome?

Netherton syndrome may be evident at birth or during the first weeks of life. There is widespread reddening ([erythroderma](#)) and the skin is covered in dry fine scales ([ichthyosis](#)). An itchy eczematous rash may be present, especially later in childhood. The skin defect causes the skin to be “leaky” and the newborn infant loses heat, water and proteins, which are all essential for normal growth and development. In addition, babies are more at risk of infection, which can be life-threatening in some cases. Newborns with severe symptoms often have a poor prognosis.

For infants with less severe symptoms, many will suffer from a failure to thrive in the first year of life and by the second year of life although the health of most children will start to improve, most will remain underweight and of short stature. Other signs a patient may develop sometime throughout their lifetime include:

- Ichthyosis linearis circumflexa – development of a distinctive circular scaling on the skin. This condition usually occurs after 2 years of age and tends to go through bouts of flaring followed by months of no skin symptoms. The rash is circular and has a thickened horny margin with a slowly changing pattern.
- “Bamboo hair” – a ball-and-socket-type hair-shaft deformity. Most patients will grow sparse, abnormal hair that is short, spiky, lustreless and brittle. Older patients may lose eyebrows and eyelashes altogether. Some patients may appear to have normal-looking hair but the hair shaft is abnormal under the microscope (subclinical trichorrhexis invaginata).
- Allergic manifestations – many patients are prone to develop food allergy, especially to nuts. Most will have a greater likelihood of developing hay fever, asthma and atopic eczema. Patients have high levels of IgE (allergy antibody) in their blood and may suffer attacks of [angioedema](#) (severe allergic skin reaction) and [urticaria](#).

### What causes Netherton syndrome?

Netherton syndrome is inherited as an autosomal recessive trait. The condition is caused by mutations in the SPINK5 gene that is found on chromosome 5. In some cases there is no family history of the trait and Netherton syndrome is revealed when two unaffected parents who are both carriers of the mutated recessive gene have a child who receives both copies of the recessive gene.

### How is the diagnosis made?

Netherton syndrome should be at the top of the differential diagnosis list in a newborn with erythroderma and abnormal-looking scalp hair, or in an older child with ichthyosis linearis circumflexa and sparse lustreless hair.

Examination of abnormal hair under microscope shows trichorrhexis invaginata (defects of the hair shafts). This does not usually develop until after 2 years of age but may occur earlier.

Skin biopsy and DNA testing may also be performed to confirm diagnosis.

## What is the treatment for Netherton syndrome?

There is no specific treatment for Netherton syndrome. The goals of treatment are to manage the symptoms and prevent skin infections and other complications.

- [Emollients](#) and keratolytics (for example, creams containing [urea](#), lactic acid or [salicylic acid](#)) should be applied to keep the skin moist and hydrated
- [Antibiotics](#) may be used for skin infections
- [Topical steroids](#) may be helpful in older children with eczema but can be absorbed too much in infants with erythroderma causing complications such as pituitary adrenal axis suppression
- Other treatments that have been tried include [photochemotherapy](#) (PUVA) and oral retinoids ([acitretin](#) and [isotretinoin](#)) but these may aggravate the skin problem

Symptoms of Netherton syndrome tend to improve with age. Periods of little or no disease symptoms are interspersed with intermittent exacerbations. Netherton syndrome may increase the risk of skin cancer developing.

### Related information

#### References:

- Book: Textbook of Dermatology. Ed Rook A, Wilkinson DS, Ebling FJB, Champion RH, Burton JL. Fourth edition. Blackwell Scientific Publications.

#### On DermNet NZ:

- [Hair loss](#)
- [Ichthyosis](#)

#### Other websites:

- [Netherton syndrome](#) – emedicine dermatology, the online textbook

#### Books about skin diseases:

See the [DermNet NZ bookstore](#)

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DermNet does not provide an on-line consultation service.

If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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