

## What Is Netherton Syndrome?

Netherton syndrome is a less common form of ichthyosis. It generally affects the skin, hair and immune system.

Infants with Netherton syndrome are usually born with red, scaly skin that may leak fluid. In older children and adults, the scaling of the skin may have a circular pattern. Bamboo hair is common; hair that has ridges or knots similar to bamboo stalks, and is fragile and breaks easily. People with Netherton symptoms are also more prone to allergies, asthma and eczema.

## What causes it?

Netherton syndrome is not contagious. It is caused by a change or variant in the SPINK5 gene so it does work as it should. As a result, there are not enough outer layers of skin (stratum corneum). The skin is too thin and breaks down easily; it can also lead to abnormal hair growth and immune system changes that can make allergies, asthma and eczema more likely. People with Netherton syndrome inherit two copies of the altered SPINK5 gene – one from each parent.

## How common is it?

Netherton syndrome is very rare. According to the National Organization of Rare Disorder, there are about 150 reported cases, with the total population of people living with Netherton estimated to be fewer than 5,000.

Other names you might hear it called:

Bamboo hair syndrome

Comel-Netherton syndrome

Ichthyosis erythroderma. With hypotrichosis and hyper-IgE

Ichthyosis linearis circumflexa

## Signs & Symptoms

Infants with Netherton syndrome may be born prematurely. They may:

- Have scaly or reddened skin (erythroderma)
- Be born with a thick shell-like covering of the skin (collodion membrane) that usually sheds within the first weeks of life
- Develop recurrent infections of the skin or blood (sepsis)
- Have very little hair
- Be at risk of dehydration or have higher than normal sodium levels in the blood due to excessive loss of water and fluid from the skin's surface
- Have difficulty maintaining a normal body temperature

Older children and adults with Netherton syndrome tend to have red, scaly skin throughout their lives. The scaling may have a circular look to it and be itchy. People with Netherton syndrome may also have:

- Thin or fragile hair that breaks easily called Bamboo hair
- Frequent bacterial skin infections, as well as viral infections including herpes and the human papilloma virus due, in part, to how thin and broken the skin is
- Infections of the bloodstream (sepsis)

- Skin buildup in the ears that can affect their hearing
- Difficulty moving hands, arms, feet and legs because of skin tightness
- Immune system-related problems, including food allergies, asthma and eczema are common
- Inability to sweat

It's not uncommon for babies or children with Netherton syndrome to be misdiagnosed with atopic dermatitis (red, itchy patches of skin), cradle cap, psoriasis, or ARCI-CIE at first.

## What to Expect

- Infants and children affected by Netherton syndrome may not grow or gain weight as would usually be expected.
- Developing a daily routine to support the viability of the skin is important, but may be a bit of trial and error at first. You may need to try a variety of skin care products and approaches before finding the one that gives you/your child the best results.
- Every person's experience with Netherton syndrome is unique. In general, the skin is red, dry and itchy and can peel. How severe the condition will be varies by person. While you may hear about treatments that were a sure win for some people, everyone is different. For a complete product listing, send an email to [info@firstskinfoundation.org](mailto:info@firstskinfoundation.org).
- It's important to find a dermatologist who knows how to treat Netherton syndrome, and will coordinate with other specialists, including asthma/allergy, ear, nose and throat, and primary care to optimally manage your care.
- While you can live a fulfilling life with Netherton syndrome, you may need to be flexible and make adjustments.

## Treatments and Self-Care

Treatment for Netherton syndrome and related health issues can be challenging.

### Skin care

- Bathing frequently using a mild, soap-free cleanser to soften the skin is helpful for many people
- Applying moisturizers to the skin after bathing and throughout the day can reduce skin dryness and help the skin be more flexible and comfortable. In particular, regular use of emollients and moisturizing creams containing petrolatum or lanolin and/or skin barrier repair therapies with ceramides or cholesterol is often recommended.
- Other topical creams and treatments, for example topical calcineurin inhibitors (pimecrolimus and tacrolimus) may be helpful, but need to be used with caution and sparingly as some of the medications can be absorbed into the bloodstream.
- Topical steroids are sometimes recommended, but use should be limited and not continuous. For example, itch and discomfort might be treated with a few days of steroids. Steroid substitutes called calcineurin inhibitors may also be considered, but they, too can be absorbed and should not be used over large body surface areas or chronically.
- Because the skin is so thin, treatments to exfoliate or remove scale, such as the alpha-hydroxy acids (lactic acid, glycolic acid), salicylic acid and oral retinoids are not helpful in the management of this disorder, and may actually aggravate the symptoms.

### Other measures to manage Netherton syndrome may include:

- Watching out for possible infections. Ask your doctor's office if you have concerns.
- Good nutrition is important, as is a higher calorie diet for young children who burn more calories than their same-sized peers because of the loss of water through the skin.
- Oral antihistamines to help with itchy eczema.
- Topical or oral antibiotics to treat infection(s).

- Medications for allergies or asthma; for people with food allergies or sensitivities, avoiding certain foods seems to help the skin.

## Things to Keep in Mind

- Netherton syndrome is a chronic condition that needs lifelong, daily care. Researchers are trying to better understand Netherton syndrome and find better treatments.
- It is not contagious. No one can catch it by touching or being close to you or to your child.
- People (some well meaning, others less so) may stare at you, or at your child with HI, and ask questions like “What’s wrong with your skin/your child?” or “Does it hurt?”.
  - Other children may tease, bully or exclude your child because of their appearance.
  - Explaining to people that you, or your child, were born with Netherton syndrome may help them to understand that you’re not sick, it’s not something they can catch, and that you live with the condition every day.
  - Although your skin may look different from other people’s, you can still do most things that other people do.
  - You are not responsible for other people’s reactions to your or your child’s appearance, nor should you feel obligated to explain yourself.
- Parents of a child with Netherton syndrome may feel a need to be very protective of them.
  - Do your best to treat your child in the same way as unaffected children in the family, while allowing for the fact that he or she needs a special skin care regimen and must take certain precautions. Try to hold them to the same expectations and encourage them to follow their dreams and ambitions just as you do any other children.
  - Reinforce in your child with Netherton syndrome that their skin condition does not define who they are or what they can achieve in their lives.
  - Talking with other parents and families can be helpful.

## Questions to Ask

- What makes Netherton syndrome different from other types of ichthyosis?
- Why do babies and kids with Netherton syndrome gain weight so slowly?
- What are the best treatments for Netherton syndrome?
- What’s the link with allergies, asthma and eczma?
- What tends to make Netherton syndrome worse? Are there triggers?
- Are there activities I need to be more careful about, especially given how thin the skin is?
- How can I best protect myself from the sun?
- Can children with Netherton syndrome swim in pool, lake or ocean water?
- Is the sun more harmful?
- Where can I find support or other parents to talk to?



Foundation for  
Ichthyosis & Related  
Skin Types

PO Box 1067, Lansdale, PA 19446-0687

Phone: 800-545-3286

Website: [www.firstskinfoundation.org](http://www.firstskinfoundation.org)

Email: [info@firstskinfoundation.org](mailto:info@firstskinfoundation.org)

***Improve lives and seek cures for those affected by ichthyosis and related skin types.***

*January 2023*