Ichthyosis Support Group

People who care about ichthyosis

Netherton Syndrome

What is Netherton Syndrome?

Netherton Syndrome (NS) is a rare inherited skin condition, often present at birth or in the first few weeks of life where the skin is very red with fine dry scales. The condition tends to last for a lifetime but the severity can vary from person to person.

Under normal circumstances, the skin acts as a type of barrier, maintaining a constant body temperature, while keeping moisture in and infection out. In NS, however, this function is impaired and the skin becomes "leaky." This leads to a loss of heat, important proteins and moisture and makes the individual prone to infection. It is therefore common for babies with this condition to have to stay in a hospital incubator for a while to prevent dehydration or infection and allow their skin to be kept moist at all times. In very rare cases, new-born's with particularly severe symptoms may not survive. The first year of life is the critical period and some children are in and out of hospital with repeated infections, dehydration and poor weight gain.

What are the signs?

At birth, babies with NS look as if they have been scalded with their skin at its reddest for the first year of life. One of the key features of NS is a failure to thrive, especially in the first two years of life. Growth and weight gain are usually significantly lower than that of children of a similar age and individuals with NS tend to be short in stature. As children get older, their skin tends to improve, although some people stay red all over, while others develop circular patches of red and scaly skin. This may appear to have a "double edge of skin" known as Linearis Circumflexa. In addition to having dry and red skin, individuals with NS usually have other symptoms associated with the condition, including spikey-looking, dull, brittle hair; eye problems, such as excessive watering; and many patients develop allergies, especially to nuts and fish. They may also develop hay fever, asthma and atopic eczema and experience hearing loss due to the build-up of scales inside the ears.

How is the condition diagnosed?

Symptoms vary from person to person but there are a number of common signs that doctors will check to differentiate NS from other skin conditions.

The skin will be red, especially for the first year of life and will usually be rough or "scaly" and quite itchy at times.

Under a microscope, cut hair will show defects in the hair shaft and will look "spiky" or "gelled". This is referred to as "bamboo hair". This method may not always be reliable in early life as the hair changes may not have happened yet.

Sometimes, a small piece of skin may be taken to do a skin biopsy to check for a protein called LEKTI; this will be missing in someone with NS. A skin biopsy is usually done by a dermatologist rather than a GP or paediatrician.

A blood test can also be taken (and sent to a national reference laboratory) to check for a fault in the SPINK-5 gene.

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Why is Netherton Syndrome sometimes misdiagnosed?

Often babies or children are not diagnosed for months or even years as red skin at birth can be a symptom of other conditions, such as other types of ichthyosis, severe eczema or other forms of immune deficiency. It is therefore important to ensure a baby or child has other tests at this stage to rule out conditions other than NS.

Managing Netherton Syndrome

As yet, there is no cure for NS but continual care with moisturisers and anti-infection treatments along with nutritional advice should help keep the skin as healthy as possible.

How successful each treatment is tends to vary from person to person and what works for one person may not work for another. Patients will need to consider different options to find the best treatment for them.

Due to their slowed growth, some babies with NS may need to be fed a special high protein diet to help them gain weight. This is usually given "artificially" via a tube as the baby cannot feed well enough and should be managed by a children's dietician. The feeding tube is either passed through the baby's nose into their stomach (nasogastric feeding) or through their abdominal wall, known as gastrostomy. Some babies with NS may need to be given special feeds into a vein.

Multiple food allergies are common in NS so advice from a dietitian on how to avoid them and what alternate foods to use, is essential to ensure adequate nutrition. In the early years extra dietary supplements are recommended, especially protein. Patients with NS will also be prone to dehydration and should therefore drink plenty of water. To minimise the risk of dehydration, make sure teachers and colleagues are aware of the need for additional hydration throughout the day while at school or in the workplace.

Moisturising creams and skin treatment

It is very important that the skin is kept moisturised at all times and that greasy, moisturising creams and ointments are applied frequently throughout the day.

Moisturising creams or emollients used should be perfume-free and without additives to avoid any allergic reactions. Emollients should be used to keep the skin as moist and hydrated as possible.

Soaking in the bath for a period of time is more beneficial than a shower. Bath oils and soap substitutes can also help and these are safe for long-term use.

Steroid creams or retinoid medicines should be avoided as these do not tend to help the skin and can even make the condition worse.

Both children and adults with NS are more susceptible to infection than those without and the use of antibiotics can be important to help protect the skin against infection. Antiseptics and antimicrobials can also provide the skin with additional cover. It is important to seek advice from a pharmacist, dermatology nurse or dermatologist before trying any new cream. It is highly recommended that individuals patch test creams for sensitivity before general use.

Ear treatment

Patients may experience a build up of dead skin in their ears and regular appointments with the Ear, Nose and Throat department at the local hospital for micro suction should help to prevent problems such as potential impaired hearing.

More specific information on caring for the skin, eyes and ears in NS and other forms of ichthyosis can be found on our website.

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Personal care

Apart from keeping the skin hydrated and moist at all times, patients may get tired and overheated quite quickly when doing active sports (e.g running, football etc). They may need to rest more than other individuals and must balance the child's requirements. Good communication between the parent, health and education services is essential.

Red and peeling skin, particularly on the face can be difficult for building self-confidence so individuals with NS may need extra support. As general practitioners may not understand the psychological impact of the condition, other health professionals can provide more help. Some individuals choose to wear wigs due to hair abnormalities.

Support

Some children with NS may require additional teaching support if learning difficulties have been identified. Some children have difficulties with speech due to impaired hearing caused by the build up of dead skin in the ear and a child's progress in school may be affected due to frequent hospital visits or hearing checks during school hours.

Physiotherapy

Patients with NS may have slightly less mobility than other individuals and difficulty extending the arms and legs completely. Physiotherapy may help improve general mobility.

For employed adults with NS, it is important to ensure their place of work knows about the condition and any additional support required. If patients are unable to work because of the condition, they may be entitled to financial support. Visit the DirectGov website (http://www.direct.gov.uk/en/DisabledPeople/FinancialSupport/index.htm) for further information about claiming Disability Living Allowance. Families with babies and children should also be eligible to apply for DLA and Family Fund can provide additional help.

How is the condition inherited?

The condition runs in families as NS is an inherited (i.e genetic) disorder. However, NS is an autosomal recessive form of genetic disorder so the condition usually occurs out of the blue. Each parent provides half their child's genes so if both parents have one copy of the faulty gene they will be carriers of the condition but will not have NS. Their child must have two copies of the faulty gene, one from each parent for NS to occur. Individuals with NS will pass one of faulty gene onto their children so their children will be carriers but are very unlikely to be affected.

Genetic counselling is available for parents with children affected by NS, and for those patients thinking of starting a family. A referral to a clinical geneticist should be requested from your general practitioner or consultant dermatologist.

What genes are affected?

Scientists are not yet sure on the exact cause of NS but patients with NS have been found to have a faulty gene (the SPINK5 gene), which fails to produce a certain protein called LEKTI. Though still in the early stages, researchers are now trying to find ways of replacing this missing protein, offering real hope for the future. There are active medical trials currently in place

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Further help

There are a number of online forums about NS where individuals can share their experiences and detail their own treatment recommendations. Not everything will work for everyone but they can be a good place both to receive and offer support.

Contact the Ichthyosis Support Group for information, advice, details on useful products, and to be connected with other people to share experiences and helpful advice.

To find out more about the ISG or become a member please get in touch in one of the following ways:

By Phone or Fax:

Tel: **0845 602 9202** Fax: **0560 343 8046** (on request)

By Email:

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By Post:

Ichthyosis Support Group

PO Box 1242, Yateley GU47 7FL

Facebook:

facebook.com/ichthyosissupportgroup

Twitter:

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