



Netherton's Syndrome

Netherton's syndrome is a rare congenital skin disease (present at birth) of unknown cause which is classified as an ichthyosis. That is because the skin is red and scaly most, if not all the time. There are also hair, infection and allergy problems but because red skin at birth can be due to several diseases, it may not be accurately diagnosed until later in the child's life.

What does it look like?

Most affected infants are "scalded-looking" at birth (congenital erythroderma) and are slow to gain weight, especially in the first year of life. They may, as a result, be kept in hospital until they are growing, perhaps weeks or even months. Their skin is very "leaky" and loses heat, water and protein. These babies may need to be kept in incubators in the special care unit for some time and will need extra fluids and food to compensate. They are more at risk of infection because of their inflamed skin. Skin treatment means regular gentle cleansing, moisturising and careful handling.

What happens next?

There is no cure for the skin condition but in most infants it improves slowly with time. As they get older the skin may remain red and scaly (erythrodermic) all over or just in certain areas, or the child can develop repeated cycles of scaling that occur at intervals of months (ichthyosis linearis circumflexa). The skin may at times be itchy, sensitive or raw and appear thickened at the joints.

What are the other signs?

Patients with Netherton's syndrome have fragile, spiky hair which seems to grow very slowly. This affects the scalp hair, eyebrows, eyelashes and body hair and is a clue to the diagnosis. If the "odd-looking" hair is noticed early on it can be examined under the microscope when it shows



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a characteristic structure due to “bamboo” swellings on the shaft. This sign clinches the diagnosis even in a baby and the swellings are due to areas of weakness and “telescoping” of the hair. This is why the hair breaks so easily. Many children with Netherton’s syndrome become allergic to foods such as fish and nuts and will also notice that they have swellings of the face or lips when in contact with, or are near to the offending items. Hay fever can occur and the children have high levels of the allergy antibody (IgE) in their blood. They are usually shorter and lighter than other children and may not be as active in sports activities as they easily overheat. Their school performance should not be unduly affected but life can be difficult if the skin, especially the face, is constantly red and peeling.

Is there any treatment?

There is no cure as the cause is not known but intensive nursing care in infancy and continued care with moisturisers and anti-infective treatments when necessary will keep the skin as good as possible. Dietary supplements especially protein are often needed in early childhood and extra help with physiotherapy and teaching should be available if needed. Our experience shows that usual treatments for inflamed skin, such as steroid creams or retinoid medicines are ineffective in Netherton’s syndrome and indeed may make things worse.

Is it passed on?

Netherton’s syndrome usually appears out of the blue with no history of such a condition in the family. It is a genetic disease where the parents are each “carriers” of the faulty gene or have a single dose of the gene and unfortunately they both pass it onto the child who has a double dose of the gene. For parents with an affected child there is a one in four chance of any future baby being affected with the same condition. Girls tend to be less severely affected than boys. There is no way of preventing or reliably detecting it before birth. There is research in progress to try and identify the causative gene but as there are millions of genes it is a complicated and slow business.



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