

Erythrokeratoderma Variabilis

Erythrokeratoderma variabilis (EKV) is a very rare, inherited skin disease. There are approximately 50 affected families in the UK. The name comes from the clinical findings of redness (erythema) and thickened skin (hyperkeratosis).

When does it begin?

90% of affected individuals develop skin findings in the first year of life.

What are the signs?

Affected individuals have 2 major skin findings - (1) transient red patches that move around and (2) thickening of skin to form yellow-brown areas on the arms, legs and sometimes the trunk. Some patients notice the red patches are affected by changes in temperature e.g. weather or room temperature. These changes can cause discomfort to the patient. About 50% of patients have thickening or peeling of the skin on the palms and soles. Hair, nails and teeth are not involved.

What is the cause?

EKV usually runs in families but sporadic cases with no family history can occur. It is caused by mutations (small mistakes or faults) in genes encoding connexins (proteins that are involved in communication between cells in the upper layer of the skin). The two connexins that are mutated in about 60% of cases are connexin 30.3 and connexin 31. In about 40% of cases, no mutation is found in connexins. It is not known what gene causes EKV in these families.

How is it passed on?

Most cases of EKV are autosomal dominant. This means that if a parent is affected, there is a 50% chance in each pregnancy that the new-born baby will inherit the mutation from the affected parent and will develop EKV.

How is EKV diagnosed?

The diagnosis is usually made from the appearance of the skin lesions by a dermatologist with experience of paediatric dermatology/genetic skin disease.

What is the treatment?

Treatment may include the use of moisturisers and creams containing keratolytics (that remove the thickened skin) including urea, salicylic acid and propylene glycol. Patients with EKV usually respond very well to oral retinoids. Retinoids are vitamin A-derived drugs that are used to used for many forms of ichthyosis and EKV. Retinoids are teratogenic (harmful to the growing baby in the womb) and are not prescribed and must not be taken for women of child-bearing age who are planning a pregnancy.

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 agents should be perfume-free and without additives to avoid any allergic reactions. Bath oils are important so that bathing does not cause drying or irritation of the skin. Bathing is considered to be more beneficial than showering.

/cont..>



People who care about ichthyosis

Emollients and keratolytic (anti-scaling) creams which contain urea, and/or lactic acid and propylene glycol (see product's ingredients label for these) can be used to keep the skin as moist and hydrated as possible, however keratolytics can cause irritation if applied to inflamed and/or broken skin.

Steroid creams should be avoided as these do not help the skin.

Antibiotics are also important to treat secondary skin infections and gentle antiseptics can provide the skin with additional protection. Washing with an emollient containing an antimicrobial agent may help minimise the risk of skin infection.

It is important to check with your doctor or pharmacist before trying any new treatment. It is advisable to patch test any new cream for sensitivity before general use, and even "herbal" and "natural" creams may contain unsuitable ingredients.

Oral treatment (tablets) may be needed in severe ichthyosis. A group of drugs known as retinoids (synthetic vitamin A derivatives) are sometimes used (e.g. acitretin). They can reduce the thickness of the scale and help improve the appearance by reducing the overactive growth of the outer skin layer. Although often effective, they have a number of side-effects which should be fully discussed and considered before starting the tablet. Monitoring blood tests are needed while taking retinoid tablets, and pregnancy must be avoided as the drug can severely damage any unborn baby (teratogenic).

Personal care

Apart from keeping the skin hydrated and moisturised at all times, patients may get tired and overheated quickly when doing active sports (e.g. running, football etc). They may need to rest more than other individuals and teachers should be aware of this so they do not push children to continue participating. Individuals should be given the flexibility to access fluids throughout the day and not just when participating in sporting activities.

Peeling skin, particularly on the face can be difficult for building self-confidence so individuals may need extra support. General practitioners need to understand the psychological impact of the condition and provide adequate support to the family, which may require the involvement of other health professionals.

Individuals may experience scaling on the scalp, and patients should manage their scalp to lessen the scaling where possible. Please see our leaflet for advice about how to look after your scalp.

More specific information on caring for the skin, eyes and ears can be found on the Ichthyosis Support Group website at www.ichthyosis.org.uk

/cont...>



People who care about ichthyosis

Further help

There are a number of online forums 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:

isg@ichthyosis.org.uk

By Post:

Ichthyosis Support Group

PO Box 1242, Yateley GU47 7FL

Facebook:

facebook.com/ichthyosissupportgroup

Twitter:

twitter.com/ISG_Charity