



Erythrokeratoderma Variabilis

EKV (erythrokeratoderma variabilis) 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 does it look like?

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 noticed that the red patches are affected by changes in temperature eg room temperature or warm weather. 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 in 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 for many forms of ichthyosis and EKV. Retinoids are teratogenic (harmful to the growing baby in the womb) and are not prescribed for women of child-bearing age who are planning a pregnancy.

What is the prognosis? In some patients, the skin findings improve with age. EKV patients have a normal life span.

