Foundation for Ichthyosis & Related Skin Types, Inc.®

Erythrokeratodermia Variabilis

What is Erythrokeratodermia Variabilis?

Erythrokeratodermia variabilis (EKV) is a very rare inherited skin disorder of cornification associated with noninflammatory erythema. It is present at birth or becomes apparent in infancy.

What are the Signs & Symptoms?

Although its signs and symptoms vary, the condition is characterized by two major features. The hallmark of erythrokeratodermia variabilis (EKV) is the seemingly independent occurrence of transient, figurate erythema (reddening of the skin) and hyperkeratosis (abnormal thickening of the outer layer of skin). Frequently, one of these features predominates; occasionally, one may be absent. Skin lesions in EKV may constantly change their appearance and vary among patients.

How is it Diagnosed?

Doctors frequently use genetic testing to help define which ichthyosis a person actually has. This may help them to treat and manage the patient. Another reason to have a genetic test is if you or a family member wants to have children. Genetic testing, which would ideally be performed first on the person with ichthyosis, is often helpful in determining a person's, and their relative's, chances to have a baby with ichthyosis. Genetic testing may be recommended if the inheritance pattern is unclear or if you or a family member is interested in reproductive options such as genetic diagnosis before implantation or prenatal diagnosis.

Results of genetic tests, even when they identify a specific mutation, can rarely tell how mild or how severe a condition will be in any particular individual. There may be a general presentation in a family or consistent findings for a particular diagnosis, but it's important to know that every individual is different. The result of a genetic test may be "negative," meaning no mutation was identified. This may help the doctor exclude certain diagnoses, although sometimes it can be unsatisfying to the patient. "Inconclusive" results occur occasionally, and this reflects the limitation in our knowledge and techniques for doing the test. But we can be optimistic about understanding more in the future, as science moves quickly and new discoveries are being made all the time. You can receive genetic testing through the Yale University's Disorders of Keratinization study with Dr. Keith Choate or for more information about genetic tests performed you can visit GeneDx, www.genedx.com.

What is the Treatment?

Treatment may include the use of moisturizers 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. Please consult your doctor.

References.
https://ghr.nlm.nih.gov/condition/erythrokeratodermia-variabilis-et-progressiva
http://www.ichthyosis.org.uk/ekv-erythrokeratoderma-variabilis/

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

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