Erythrokeratodermia-cardiomyopathy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Erythrokeratodermia-cardiomyopathy syndrome. ORPHA:476096

Erythrokeratodermia-cardiomyopathy syndrome is a rare, genetic erythrokeratoderma disorder characterized by generalized cutaneous erythema with fine white scales and pruritus refractory to treatment, progressive dilated cardiomyopathy, palmoplantar keratoderma, sparse or absent eyebrows and eyelashes, sparse scalp hair, nail dystrophy, and dental enamel anomalies. Variable features include failure to thrive, developmental delay, and development of corneal opacities. Histology shows psoriasiform acanthosis, hypogranulosis, and compact orthohyperkeratosis.