DISEASE:
Erythrokeratodermia-cardiomyopathy syndrome

NAME:	Erythrokeratodermia-cardiomyopathy syndrome
DESCRIPTION:	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.
ORPHACODE:	476096
SYNONYMS:	EKC syndrome
XREF(S):	Orphanet
ANALYTE(S):	DSP
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RELATED CONTENT

Related Genetic Tests

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- Cardiopathies, hereditary (gene panel)

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- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- desmoplakin

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- Cardiopathies, hereditary (102 genes) - KUL