DK1-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. DK1-CDG. ORPHA:91131

DK1-CDG is characterised by muscular hypotonia and ichthyosis. It has been described in four children from two consanguineous families. All the affected children died during early infancy, two from dilated cardiomyopathy. The syndrome is caused by a deficiency in dolichol kinase 1 (DK1), an enzyme involved in the de novo biosynthesis of dolichol phosphate. The mutations identified in the DK1 gene led to a 96 to 98% reduction in DK activity.