DISEASE:
DK1-CDG

NAME:	DK1-CDG
DESCRIPTION:	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.
ORPHACODE:	91131
SYNONYMS:	CDG syndrome type Im CDG-Im CDG1M Carbohydrate deficient glycoprotein syndrome type Im Congenital disorder of glycosylation type 1m Congenital disorder of glycosylation type Im Dolichol kinase deficiency Hypotonia and ichthyosis due to dolichol phosphate deficiency
XREF(S):	Orphanet ICD-10 OMIM

ANALYTE(S):	DOLK
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Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

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