DISEASE:
MEDNIK syndrome

NAME:	MEDNIK syndrome
DESCRIPTION:	A rare disorder of copper metabolism characterized by intellectual deficit, enteropathy, sensorineural hearing loss, peripheral neuropathy, lamellar and erythrodermic ichthyosis, and keratodermia.
ORPHACODE:	171851
SYNONYMS:	Intellectual disability-enteropathy-deafness-peripheral neuropathy-ichthyosis-keratodermia syndrome Intellectual disability-enteropathy-hearing loss-peripheral neuropathy-ichthyosis-keratodermia syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	AP1B1 AP1S1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Ichthyosis (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- adaptor related protein complex 1 subunit beta 1
- adaptor related protein complex 1 subunit sigma 1

Related Gene Panels

- Ichthyosis and erythroderma (98 genes) - KUL