

DISEASE:
CEDNIK syndrome

NAME:	CEDNIK syndrome
DESCRIPTION:	A rare, genetic, neurocutaneous disease characterized by severe developmental abnormalities of the nervous system and aberrant differentiation of the epidermis. Patients present with a unique constellation of clinical signs described with the acronym CEDNIK: CErebral Dysgenesis, Neuropathy, Ichthyosis, and palmoplantar Keratoderma.
ORPHACODE:	66631
SYNONYMS:	Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>SNAP29</u>
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