

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Synonimy

Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome
Zespół dysgenezja mózgu-neuropatia-rybia łuska
rogowiec dłoni i stóp

Kod ORPHA

66631

Kod OMIM

609528

Kod ICD10

Q82.8

Kod ICD11

LD27.2

*Źródło

orphanet