

Opis choroby *

Definicja

Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome is characterised by ichthyosis, hepatosplenomegaly and late-onset cerebellar ataxia. It has been described in two brothers. Transmission is either autosomal recessive or X-linked.

Dane

Klasyfikacja

Choroba

Synonimy

Dykes-Marks-Harper syndrome

Zespół Dykesa, Markesa i Harpera

Kod ORPHA

2274

Kod OMIM

242520

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet