

Opis choroby *

Definicja

A rare, genetic, syndromic hair shaft abnormality disorder characterized by short, dry, sulfur-deficient, brittle hair usually associated with highly variable neuroectodermal manifestations, such as ichthyosis, photosensitivity, and intellectual disability.

Dane

Klasyfikacja

Choroba

Kod ORPHA

33364

Kod OMIM

616395

Kod ICD10

L67.8

Kod ICD11

EC21.1

*Źródło

orphanet