

Opis choroby *

Definicja

Chédiak-Higashi syndrome (CHS) is a rare severe genetic disorder generally characterized by partial oculocutaneous albinism (OCA, see this term), severe immunodeficiency, mild bleeding, neurological dysfunction and lymphoproliferative disorder. A classic, early-onset form and an attenuated, later-onset form (Atypical CHS; see this term) have been described.

Dane

Klasyfikacja

Choroba

Synonimy

Chédiak-Higashi disease
Choroba Chédiaka i Higashiego
Zespół Chédiaka, Higashiego i Steinbrinka
Chédiak-Higashi-Steinbrink syndrome

Kod ORPHA

167

Kod OMIM

214500

Kod ICD10

E70.3

Kod ICD11

EC23.20

*Źródło

orphanet