

## 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

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#### \*Źródło

orphanet