

Opis choroby *

Definicja

Hereditary hyperferritinemia with congenital cataracts is characterized by the association of early onset (although generally absent at birth) cataract with persistently raised plasma ferritin concentrations in the absence of iron overload.

Dane

Klasyfikacja

Choroba

Synonimy

Bonneau-Beaumont syndrome

HHCS

Zespół hiperferrytemia dziedziczna-zaćma

Zespół Bonneau i Beaumont

HHCS

Hereditary hyperferritinemia with congenital cataracts

Kod ORPHA

163

Kod OMIM

600886

Kod ICD10

H26.0

Kod ICD11

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*Źródło

orphanet