

Opis choroby *

Definicja

A rare disorder characterised by the association of aplasia cutis congenita with high myopia, congenital nystagmus and cone-rod dysfunction. It has been described in two siblings (brother and sister). Transmission is autosomal dominant.

Dane

Klasyfikacja

Choroba

Synonimy

Gershoni-Baruch-Leibo syndrome

Zespół Gershoni, Baruchi i Leibo

Kod ORPHA

1117

Kod OMIM

601075

Kod ICD10

Q84.8

Kod ICD11

LD27.Y

*Źródło

orphanet