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

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*Źródło

orphanet