

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	Synonimy
Choroba	Gershoni-Baruch-Leibo syndrome Zespół Gershoni, Baruchi i Leibo

Kod ORPHA	Kod OMIM	Kod ICD10
1117	601075	Q84.8

Kod ICD11
LD27.Y

*Źródło

orphanet