Opis choroby *

Definicja

A disorder that is characterised by the association of a non-progressive congenital ataxia, severe intellectual deficit, optic atrophy and structural anomalies of the skin vessels. It has been described in five children from a large consanguineous Lebanese family. Short stature and microcephaly were also reported. Transmission is autosomal recessive.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cerebellar ataxia-intellectual disability-optic atrophy-skin abnormalities syndrome
Ataksja móżdżkowa - niepełnosprawność intelektualna - zanik nerwu wzrokowego - nieprawidłowości skóry
SCAR5
SCAR5

Kod ORPHA
83472

Kod OMIM
606937

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet