

Opis choroby *

Definicja

Pili torti-developmental delay-neurological abnormalities syndrome is characterized by growth and developmental delay, mild to moderate neurologic abnormalities, and pili torti. It has been described in a brother and his sister born to consanguineous Puerto Rican parents.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2891

Kod OMIM

261990

Kod ICD10

Q84.1

Kod ICD11

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*Źródło

orphanet