

Opis choroby *

Definicja

Lipodystrophy-intellectual disability-deafness syndrome is an extremely rare form of genetic lipodystrophy (see this term), reported in 3 patients from one family to date, characterized by generalized congenital lipodystrophy, low birth weight, progressive sensorineural deafness occurring in childhood, intellectual deficit, progressive osteopenia, delayed skeletal maturation, skeletal abnormalities described as slender, undermineralized tubular bones, and dense metaphyseal striations in the distal femur, ulna and radius of older patients. Autosomal recessive inheritance has been suggested.

Dane

Klasyfikacja

Choroba

Synonimy

Lipodystrophy-intellectual disability-hearing loss syndrome
Zespół Rajaba i Sprangera
Rajab-Spranger syndrome

Kod ORPHA

50811

Kod OMIM

608154

Kod ICD10

Q78.8

Kod ICD11

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*Źródło

orphanet