

## 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