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Lipodystrophy-intellectual disability-deafness syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Lipodystrophy-intellectual disability-deafness syndrome. ORPHA:50811

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.