

Opis choroby *

Definicja

Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome is characterised by metaphyseal dysplasia, short-limb dwarfism, mild intellectual deficit and conductive hearing loss, associated with repeated episodes of otitis media in childhood. It has been described in three brothers born to consanguineous Sicilian parents. Variable manifestations included hyperopia and strabismus. The mode of inheritance is autosomal recessive.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Metaphyseal dysostosis-intellectual disability-conductive hearing loss syndrome

Kod ORPHA

2502

Kod OMIM

250420

Kod ICD10

Q78.5

Kod ICD11

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*Źródło

orphanet