Opis choroby *

Definicja

A rare autosomal recessive microcephalic primordial dwarfism characterized by congenital microcephaly and craniofacial features associated with a spectrum of limb abnormalities ranging from mild to severe. Short stature is frequently observed and often is severe.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
572761

Kod OMIM
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Kod ICD10
Q87.1

Kod ICD11
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*Źródło

orphanet