Opis choroby *

Definicja

A rare genetic hypoparathyroidism characterized by severe hypocalcemia, seizures, hyperphosphatemia, and undetectable parathyroid hormone levels, in the absence of parathyroid tissue. Complications include psychomotor and growth delay, delayed dentition, and cataracts.

Dane

Klasyfikacja
Podtyp kliniczny

Kod ORPHA
2239

Kod OMIM
146200

Kod ICD10
E20.8

Kod ICD11
5A50.0Y

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*Źródło

orphanet