Opis choroby *

Definicja

A rare disorder characterised by growth retardation with prenatal onset, cataracts, microcephaly, intellectual deficit, immune deficiency, delayed ossification and enamel hypoplasia. It has been described in two siblings. Transmission is autosomal recessive.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2643

Kod OMIM
251190

Kod ICD10
Q87.1

Kod ICD11
LD24.D

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*Źródło

orphanet