Opis choroby *

Definicja

A rare, congenital malformation syndrome characterized by the association of anterior ocular chamber cleavage disorder with developmental delay, short stature and congenital hypothyroidism. Additional manifestations include cerebellar hypoplasia, tracheal stenosis, narrow external auditory meatus, and hip dislocation. There have been no further description in the literature since 1995.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2321

Kod OMIM
601427

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet