

## 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