

Opis choroby *

Definicja

Intellectual disability-balding-patella luxation-acromicria syndrome is characterised by severe intellectual deficit, patella luxations, acromicria, hypogonadism, facial dysmorphism (including midface hypoplasia and premature frontotemporal balding). It has been described in three unrelated males.

Dane

Klasyfikacja **Synonimy**

Zespół wad wrodzonych Scholte-Begeer-van Essen syndrome
Zespół Scholte, Begeera i van Essena

Kod ORPHA

3041

Kod OMIM

300977

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet