Opis choroby *

Definicja

An extremely rare syndrome reported in two siblings of non consanguineous parents that is characterized by the association of ocular abnormalities (partial aniridia, congenital glaucoma, telecanthus) with frontal bossing, hypertelorism, unilateral renal agenesis and mild psychomotor delay. There have been no further descriptions in the literature since 1974.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Sommer-Rathbun-Battles syndrome
Zespół Sommera, Rathbuna i Battlesa

Kod ORPHA
1064

Kod OMIM
206750

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet