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 Synonimy

Zespół wad wrodzonych Sommer-Rathbun-Battles syndrome

Zespół Sommera, Rathbuna i Battlesa

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 1064
 206750
 Q87.8

Kod ICD11

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*Źródło

orphanet