Opis choroby *

Definicja

Cataract-deafness-hypogonadism syndrome is an extremely rare multiple congenital abnormality syndrome, described in only three brothers to date, that is characterized by the association of congenital cataract, sensorineural deafness, hypogonadism, mild intellectual deficit, hypertrichosis, and short stature. There have been no further descriptions in the literature since 1995.

Dane

Klasyfikacja Synonimy

Zespół wad wrodzonych Cataract-hearing loss-hypogonadism syndrome

Zespół Schaapa, Taylora i Baraitsera Schaap-Taylor-Baraitser syndrome

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 1383
 Q87.8

Kod ICD11 LD2H.Y

*Źródło

orphanet