

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 1383	Kod OMIM -	Kod ICD10 Q87.8
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Kod ICD11
LD2H.Y

*Źródło

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