

Opis choroby *

Definicja

Perrault syndrome (PS) is characterized by the association of ovarian dysgenesis in females with sensorineural hearing impairment. In more recent PS reports, some authors have described neurologic abnormalities, notably progressive cerebellar ataxia and intellectual deficit.

Dane

Klasyfikacja

Choroba

Synonimy

XX gonadal dysgenesis-deafness syndrome

Dysgenezja gonad XX - głuchota

XX gonadal dysgenesis-hearing loss syndrome

Kod ORPHA

2855

Kod OMIM

617565

Kod ICD10

Q87.8

Kod ICD11

LD2H.Y

*Źródło

orphanet