

Opis choroby *

Definicja

A rare chromosomal anomaly syndrome characterized by complete or partial loss of an X chromosome in phenotypic females, clinically manifesting with short stature, primary ovarian insufficiency as well as cardiovascular, renal, liver, autoimmune diseases, hearing loss and neurocognitive abnormalities.

Dane

Klasyfikacja

Zespół wad wrodzonych 45,X syndrome

Synonimy

Zespół 45,X

Zespół 45,X/46,XX

45,X/46,XX syndrome

Kod ORPHA

881

Kod OMIM

-

Kod ICD10

Q96.4

Kod ICD11

LD50.0

*Źródło

orphanet