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

Synonimy
45,X syndrome
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