Opis choroby *

Definicja

A rare syndrome with 46,XX disorder of sex development characterized by Müllerian duct hypoplasia or agenesis associated with clinical and biological evidence of hyperandrogenism in 46,XX females. Patients present with hypoplastic or absent uterus, variable abnormalities of other reproductive organs, primary amenorrhea, acne, hirsutism, and sometimes renal anomalies. External genitalia and secondary sexual characteristics are normal. Hormonal analysis shows variably elevated serum levels of androstenedione, dehydroepiandrosterone, and/or total and free testosterone.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Müllerian duct failure and hyperandrogenism
Niedobór WNT4
Nieprawidłowości przewodów Mullera i hiperandrogenizm
WNT4 deficiency

Kod ORPHA
247768

Kod OMIM
158330

Kod ICD10
Q51.8

Kod ICD11
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*Źródło

orphanet