Opis choroby *

Definicja

A form of classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency characterized by abnormal genital development with variable levels of virilization in females, and normal genitalia in males in association with glucocorticoid insufficiency with absence of salt-wasting, accelerated growth velocity and bone maturation, premature adrenarche and precocious puberty leading to reduced adult height. Females have a normal uterus and various degrees of abnormal vaginal development.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Classic 21-OHD CAH, simple virilizing form
Klasyczny 21-OHD CAH, postać prosta maskulinizująca

Kod ORPHA
315311

Kod OMIM
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Kod ICD10
E25.0

Kod ICD11
5A71.01

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*Źródło

orphanet