

Wrodzony przerost nadnerczy spowodowany niedoborem 17-alfa-hydroksylazy

Kod Orpha: 90793 Kod OMIM: 202110

Opis choroby *

Definicja

A rare form of congenital adrenal hyperplasia due to 17-alpha-hydroxylase (CYP17A1) deficiency and characterized by glucocorticoid deficiency, mineralocorticoid excess leading to hypokalemic hypertension and sex steroid deficiency (hypergonadotrophic hypogonadism). Undervirilization and even female phenotype in 46,XY males, primary amenorrhea in females and lack of pubertal development in both sexes is common. Residual CYP17A1 activity is associated with the severity of this condition with a large spectrum of variability, from presenting in early infancy, to unusually mild courses with isolated sex steroid deficiency but normal ACTH-stimulated cortisol in adult patients.

Dane

Klasyfikacja

Choroba

Synonimy

CAH due to 17-alpha-hydroxylase deficiency

CAH spowodowany niedoborem 17-alfa-hydroksylazy

Złożony Niedobór 17-hydroksylazy/17,20-liazy

Combined 17-hydroxylase/17,20-lyase deficiency

Kod ORPHA

90793

Kod OMIM

202110

Kod ICD10

E25.0

Kod ICD11

5A71.01

[*Źródło](#)

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Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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