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

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*Źródło

orphanet