Opis choroby *

Definicja

A rare form of congenital adrenal hyperplasia (CAH) due to 3-beta-hydroxysteroid dehydrogenase (HSD3B2) deficiency and characterized by salt-wasting and non-salt wasting CAH with a wide variety of symptoms, including glucocorticoid and mineralocorticoid deficiencies in both sexes. Salt wasting can lead to dehydration and hypotension in the first few weeks of life. Affected males had undervirilization manifesting as a micropenis to severe perineoscrotal hypospadias. Females show normal or mildly virilized external genitalia (mild clitoromegaly, labial fusion) due to dehydroepiandrosterone (DHEA) accumulation and conversion to androgens by the normal HSD3B1.

Dane

Klasyfikacja
Choroba

Synonimy
CAH due to 3-beta-hydroxysteroid dehydrogenase deficiency
CAH z powodu niedoboru dehydrogenazy 3-beta-hydroksysteroidowej

Kod ORPHA
90791

Kod OMIM
201810

Kod ICD10
E25.0

Kod ICD11
5A71.01

---

*Źródło

orphanet