Opis choroby *

Definicja

A rare, genetic, endocrine disease characterized by defect in conversion of cortisone to active cortisol, resulting in ACTH-mediated excessive androgen release from adrenal glands. Premature adrenarche is typical with precocious pseudopuberty, proportionate tall stature and accelerated bone maturation in males, and hirsutism, oligoamenorrhea, central obesity and infertility in females. Imaging studies may indicate adrenal hyperplasia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
11-beta-hydroxysteroid dehydrogenase deficiency type 1
Niedobór dehydrogenazy 11-beta-hydroksysteroidowej typu 1

Kod ORPHA
168588

Kod OMIM
614662

Kod ICD10
E25.8

Kod ICD11
5A71.Y

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*Źródło

orphanet