

Opis choroby *

Definicja

A rare form of congenital adrenal hyperplasia due to P450 oxidoreductase deficiency and characterized by glucocorticoid deficiency, virilization of external genitalia in females, and undervirilization in males. Findings range from severely affected infants with 46,XX and 46,XY disorders/differences of sex development (DSD) and cortisol deficiency to mildly affected women who appear to have polycystic ovary syndrome, or mildly affected men with gonadal insufficiency.

Dane

Klasyfikacja

Choroba

Synonimy

Congenital adrenal hyperplasia due to cytochrome POR deficiency

Niedobór POR

PORD

Wrodzony przerost nadnerczy z powodu niedoboru cytochromu POR

POR deficiency

PORD

Kod ORPHA

95699

Kod OMIM

613571

Kod ICD10

E25.0

Kod ICD11

5A71.01

[*Źródło](#)

orphanet