

## Opis choroby \*

### Definicja

Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency is a rare, genetic, chronic, primary adrenal insufficiency disorder, due to partial loss-of-function CYP11A1 mutations, characterized by early-onset adrenal insufficiency without associated abnormal external male genitalia. Patients present with signs of adrenal crisis, including electrolyte abnormalities, severe weakness, recurrent vomiting and seizures. Ultrasound reveals absent (or very small) adrenal glands.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

289548

#### Kod OMIM

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#### Kod ICD10

E27.1

#### Kod ICD11

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### \*Źródło

orphanet