

## Opis choroby \*

### Definicja

A severe form of pseudohypoaldosteronism type 1 characterized by salt wasting in multiple organs including the kidney, colon, and sweat and salivary glands. Presentation is in the first few weeks of life with severe dehydration, vomiting and failure to thrive in association with hyponatremia, hyperkalemia and metabolic acidosis as well as elevated aldosterone and renin levels. No remission is reported and patients suffer from recurrent life-threatening episodes of salt loss.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Autosomal recessive PHA1

Autosomalny recesywny

pseudohipoaldosteronizm typu 1

Autosomal recessive pseudohypoaldosteronism  
type 1

Generalized PHA1

#### Kod ORPHA

171876

#### Kod OMIM

264350

#### Kod ICD10

N25.8

#### Kod ICD11

GB90.41

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#### [\\*Źródło](#)

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