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

orphanet