Opis choroby *

Definicja

A form of pseudohypoaldosteronism type 1 characterized by mild mineralocorticoid resistance that is restricted to the kidneys and that usually improves in early childhood. Typical presentation is in the neonatal period with weight loss, failure to thrive, vomiting and dehydration in association with hyponatremia, hyperkalemia and metabolic acidosis as well as elevated aldosterone and renin levels.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Autosomal dominant PHA1
Autosomalny dominujący pseudohipoaldosteronizm typu 1
Autosomal dominant pseudohypoaldosteronism type 1
Renal PHA1

Kod ORPHA
171871

Kod OMIM
177735

Kod ICD10
N25.8

Kod ICD11
GB90.41

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*Źródło

orphanet