

Opis choroby *

Definicja

A rare, primary form of mineralocorticoid resistance characterized by mild to profound salt wasting either restricted to the kidney (renal pseudohypoaldosteronism type 1), or generalized affecting many organs (generalized pseudohypoaldosteronism type 1). Clinical presentation is in the neonatal period with failure to thrive, vomiting and dehydration with biochemical findings of hyperkalaemia, metabolic acidosis and, elevated plasma aldosterone and renin concentration.

Dane

Klasyfikacja

Choroba

Synonimy

PHA type 1

PHA type 1

PHA1

Kod ORPHA

756

Kod OMIM

264350

Kod ICD10

N25.8

Kod ICD11

GB90.41

*Źródło

orphanet