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

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*Źródło

orphanet