Opis choroby *

Definicja

A rare type of familial hypoaldosteronism characterized by early infantile onset of vomiting, diarrhea, severe dehydration, and failure to thrive. Analysis of plasma electrolytes shows hyponatremia, hyperkalemia, and acidosis. Plasma renin activity is elevated, and aldosterone levels are low.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Early-onset familial hyperreninemic hypoaldosteronism
Ciężki niedobór syntazy aldosterony
Severe aldosterone synthase deficiency

Kod ORPHA
556030

Kod OMIM
-

Kod ICD10
E27.4

Kod ICD11
-

---

*Źródło

orphanet