Opis choroby *

Definicja

A rare genetic skin disease characterized by excessive salt wasting in sweat, leading to hyponatremic dehydration, hyperkalemia, and poor feeding and slow weight gain in infancy. Laboratory examination shows hyponatremia, hyperkalemia, increased aldosterone, and increased sweat chloride concentrations.

Dane

Klasyfikacja
Choroba

Synonimy
Carbonic anhydrase XII deficiency
Carbonic anhydrase XII deficiency

Kod ORPHA
542657

Kod OMIM
143860

Kod ICD10
L98.8

Kod ICD11
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*Źródło

orphanet