Opis choroby *

Definicja

A rare generalized, genetic disorder of proximal tubular transport characterized by excessive urine output with loss of low molecular weight solutes (amino acids, glucose, low-molecular weight proteins, organic acids, carnitine, calcium, phosphate, potassium, bicarbonate) and water, and which can be life threatening.

Dane

Klasyfikacja
Choroba

Synonimy
DeToni-Debré-Fanconi syndrome
Pierwotny zespół nerkowo-kanalikowy Fanconiego
Primary Fanconi renal syndrome

Kod ORPHA
3337

Kod OMIM
618913

Kod ICD10
E72.0

Kod ICD11
GB90.42

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*Źródło

orphanet