Opis choroby *

Definicja

A rare, genetic, glucose transport disorder characterized by the presence of persistent isolated glucosuria in the absence of both proximal tubular dysfunction and hyperglycemia. The disorder is benign in the majority of cases although it may occasionally manifest with polyuria, enuresis, a mild growth and pubertal maturation delay, hypercalciuria, aminoaciduria and, in severe cases, increased incidence of urinary infections and episodic dehydration and ketosis during pregnancy and starvation.

Dane

Klasyfikacja
Choroba

Synonimy
Familial renal glycosuria
Niedobór SGLT2
SGLT2 deficiency

Kod ORPHA
69076

Kod OMIM
233100

Kod ICD10
E74.8

Kod ICD11
5C61.5

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*Źródło

orphanet