

## 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