

## Opis choroby \*

### Definicja

A rare genetic disease characterized by onset of neurological deterioration in the first two years of life, progressing to severe intellectual disability, profound ataxia, mild dyskinesia, axial hypotonia, camptocormia, and oculomotor apraxia. Some patients also develop nephropathy with features of tubulointerstitial nephritis, hypertension, and a tendency for hyperkalemia.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Cerebrorenal syndrome, Perez type  
Zespół mózgowo-nerkowy, typ Pereza

#### Kod ORPHA

505242

#### Kod OMIM

617595

#### Kod ICD10

E83.2

#### Kod ICD11

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#### \*Źródło

orphanet