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