Opis choroby *

Definicja

An extremely rare genetic endocrine disease characterized by primary adrenal insufficiency, dystrophic myopathy, hepatic steatosis, severe psychomotor delay, megalocornea, failure to thrive, chronic constipation, and terminal bladder ectasia which can lead to death. There have been no further descriptions in the literature since 1982.

Dane

Klasyfikacja
Choroba

Kod ORPHA
977

Kod OMIM
300270

Kod ICD10
E27.4

Kod ICD11
5A74.Y

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*Źródło

orphanet