

Wrodzona biegunka spowodowana niedoborem komórek enteroendokrynowych

Kod Orpha: 83620 Kod OMIM: 610370

Opis choroby *

Definicja

A very rare genetic gastroenterological disease characterized by severe malabsorptive diarrhea (requiring parenteral nutrition and disappearing at fasting) due to a lack of intestinal enteroendocrine cells. It is associated with early-onset (within the first weeks of life) dehydration, metabolic acidosis and diabetes mellitus (that can develop until late childhood). Patient may display various degrees of pancreatic insufficiency that does not explain diarrhea, as it is not reduced with pancreatic enzyme supplementation. Central hypogonadism (developing in the second decade), as well as an association with celiac disease have been reported.

Dane

Klasyfikacja

Choroba

Synonimy

Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells
Anedokrynoza jelitowa

Kod ORPHA

83620

Kod OMIM

610370

Kod ICD10

P78.3

Kod ICD11

DA90.Y

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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