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

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*Źródło

orphanet