

Opis choroby *

Definicja

A rare genetic endocrine disease characterized by early onset of severe intractable diarrhea and intestinal malabsorption, followed by obesity and hormonal deficiencies due to insufficient activation of several prohormones, resulting in hypocortisolism, hypothyroidism, diabetes insipidus, hypogonadism, growth deficiency, and diabetes mellitus. Extent and age of onset of hormone deficiencies are variable between patients.

Dane

Klasyfikacja	Synonimy
Podtyp etiologiczny	PCI deficiency Niedobór PCI

Kod ORPHA	Kod OMIM	Kod ICD10
71528	600955	E66.8

Kod ICD11
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*Źródło

orphanet