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
Podtyp etiologiczny

Synonimy
PCI deficiency
Niedobór PCI

Kod ORPHA
71528

Kod OMIM
600955

Kod ICD10
E66.8

Kod ICD11
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*Źródło

orphanet