Opis choroby *

Definicja

An extremely rare, autosomal dominant immunological disorder characterized by variable enteropathy, endocrine disorders (e.g. type 1 diabetes mellitus, hypothyroidism), immune dysregulation with pulmonary and blood-borne bacterial infections, and fungal infections (chronic mucocutaneous candidiasis) developing in infancy. Other manifestations include short stature, eczema, hepatosplenomegaly, delayed puberty, and osteoporosis/osteopenia.

Dane

Klasyfikacja
Choroba

Kod ORPHA
391487

Kod OMIM
614162

Kod ICD10
K63.9

Kod ICD11
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*Źródło

orphanet