

Opis choroby *

Definicja

A group of inborn errors of biotin metabolism characterized by reduced activities of biotin-dependent enzymes resulting in a wide spectrum of symptoms, including feeding difficulty, breathing difficulties, lethargy, seizures, skin rash, alopecia, and developmental delay. This group includes biotinidase deficiency and biotin holocarboxylase synthetase deficiency.

Dane

Klasyfikacja	Synonimy
Grupa fenomenów	MCD MCD

Kod ORPHA	Kod OMIM	Kod ICD10
148	-	E53.8

Kod ICD11
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*Źródło

orphanet