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
Grupa fenomenów

Synonimy
MCD
MCD

Kod ORPHA
148

Kod OMIM
-

Kod ICD10
E53.8

Kod ICD11
-

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*Źródło

orphanet