Opis choroby *

Definicja

A rare genetic disease characterized by early-onset severe obesity due to mutations in single genes acting on the development and function of the hypothalamus or the leptin-melanocortin pathway, leading to disruption of energy homeostasis and endocrine dysfunction. Patients present with a body mass index over three standard deviations above normal at less than five years of age, accompanied by a variety of signs and symptoms according to the mutated gene, including hyperphagia, insulin resistance, reduced basal metabolic rate, or hypogonadism, among others.

Dane

Klasyfikacja
Choroba

Synonimy
Monogenic obesity due to a leptin-melanocortin pathway anomaly
Otyłość monogenowa z powodu anomalii szlaku leptyna-monokortyna

Kod ORPHA
98267

Kod OMIM
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Kod ICD10
E66.8

Kod ICD11
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*Źródło

orphanet