

Opis choroby *

Definicja

Melanocortin 4 receptor (MC4R) deficiency is the commonest form of monogenic obesity identified so far. MC4R deficiency is characterised by severe obesity, an increase in lean body mass and bone mineral density, increased linear growth in early childhood, hyperphagia beginning in the first year of life and severe hyperinsulinaemia, in the presence of preserved reproductive function.

Dane

Klasyfikacja	Synonimy
Podtyp etiologiczny	MC4R deficiency Niedobór MC4R

Kod ORPHA	Kod OMIM	Kod ICD10
71529	618406	E66.8

Kod ICD11
5B81.Y

*Źródło

orphanet