

Otyłość - zapalenie jelit - niedoczynność tarczycy - przerost serca - opóźnienie rozwoju

Kod Orpha: 88643 Kod OMIM:

Opis choroby *

Definicja

Obesity-colitis-hypothyroidism-cardiac hypertrophy-developmental delay syndrome is characterised by precocious obesity, congenital hypothyroidism, neonatal colitis, cardiac hypertrophy, craniosynostosis and developmental delay. It has been described in two brothers, one of whom died within the first month of life. The parents of the two children were nonconsanguineous and in good health, however, the pregnancies were complicated by a maternal HELLP syndrome (Haemolysis, Elevated Liver enzymes and Low Platelets). The mode of inheritance has not yet been clearly established.

Dane

Klasifikacja

Choroba

Kod ORPHA
88643

Kod OMIM
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Kod ICD10
E03.8

Kod ICD11
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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