

Opis choroby *

Definicja

Leukotriene C4 synthase deficiency is an extremely rare fatal neurometabolic developmental disorder characterized clinically by muscular hypotonia, psychomotor retardation, failure to thrive, and microcephaly.

Dane

Klasyfikacja

Choroba

Synonimy

LTC4 synthase deficiency

Niedobór syntazy leukotrienu C4

Niedobór syntazy LTC4

Leukotriene C4 synthase deficiency

Kod ORPHA

79507

Kod OMIM

614037

Kod ICD10

E88.8

Kod ICD11

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*Źródło

orphanet