

## 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