

# Acyduria fumarowa

Kod Orpha: 24 Kod OMIM: 606812

## Opis choroby \*

### Definicja

Fumaric aciduria (FA), an autosomal recessive metabolic disorder, is most often characterized by early onset but non-specific clinical signs: hypotonia, severe psychomotor impairment, convulsions, respiratory distress, feeding difficulties and frequent cerebral malformations, along with a distinctive facies. Some patients present with only moderate intellectual impairment.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Fumarase deficiency

Niedobór fumarazy

#### Kod ORPHA

24

#### Kod OMIM

606812

#### Kod ICD10

E88.8

#### Kod ICD11

5C53.1

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[\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.