

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

*Źródło

orphanet