

Opis choroby *

Definicja

Gaucher disease (GD) is a lysosomal storage disorder encompassing three main forms (types 1, 2 and 3), a fetal form and a variant with cardiac involvement (Gaucher disease - ophthalmoplegia - cardiovascular calcification or Gaucher-like disease).

Dane

Klasyfikacja

Choroba

Synonimy

Acid beta-glucosidase deficiency

Niedobór glukocerebrozydazy

Niedobór kwaśnej beta-glukozydazy

Glucocerebrosidase deficiency

Kod ORPHA

355

Kod OMIM

610539

Kod ICD10

E75.2

Kod ICD11

5C56.0Y

*Źródło

orphanet