

Opis choroby *

Definicja

Dilated cardiomyopathy with ataxia (DCMA) is characterized by severe early onset (before the age of three years) dilated cardiomyopathy (DCM) with conduction defects (long QT syndrome), non-progressive cerebellar ataxia, testicular dysgenesis, and 3-methylglutaconic aciduria.

Dane

Klasyfikacja

Choroba

Synonimy

3-methylglutaconic aciduria type 5
Acyduria 3-metyloglutakonowa typu 5
MGA5
Zespół DCMA
DCMA syndrome
MGA5

Kod ORPHA

66634

Kod OMIM

610198

Kod ICD10

E71.1

Kod ICD11

5C50.E0

*Źródło

orphanet