

Opis choroby *

Definicja

A rare metabolic disorder for which two forms have been described. Lack of activity of the erythrocyte isoform of adenosine monophosphate (AMP) deaminase has been described in subjects with low plasma uric acid levels without obvious clinical relevance and will not be described further. Myoadenylate deaminase deficiency is an inherited disorder of muscular energy metabolism with a lack of AMP deaminase activity in skeletal muscle. It is characterised by exercise-induced muscle pain, cramps and/or early fatigue.

Dane

Klasyfikacja

Choroba

Synonimy

AMP deaminase deficiency

Niedobór deaminazy AMP deaminase

Niedobór mięśniowej deaminazy monofosforanu

adenozyny

Myoadenylate deaminase deficiency

Kod ORPHA

45

Kod OMIM

615511

Kod ICD10

G71.3

Kod ICD11

5C55.0Y

*Źródło

orphanet