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

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*Źródło

orphanet