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Adenosine monophosphate deaminase deficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Adenosine monophosphate deaminase deficiency. ORPHA:45*

Adenosine monophosphate (AMP) deaminase deficiency is a metabolic disorder for which two forms have been described. Lack of activity of the erythrocyte isoform of 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.