Defective MAT1A does not transfer Ado from ATP to L-Met

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28/04/2024

Introduction

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

Literature references

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Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics, 33*, 3461-3467. ↗

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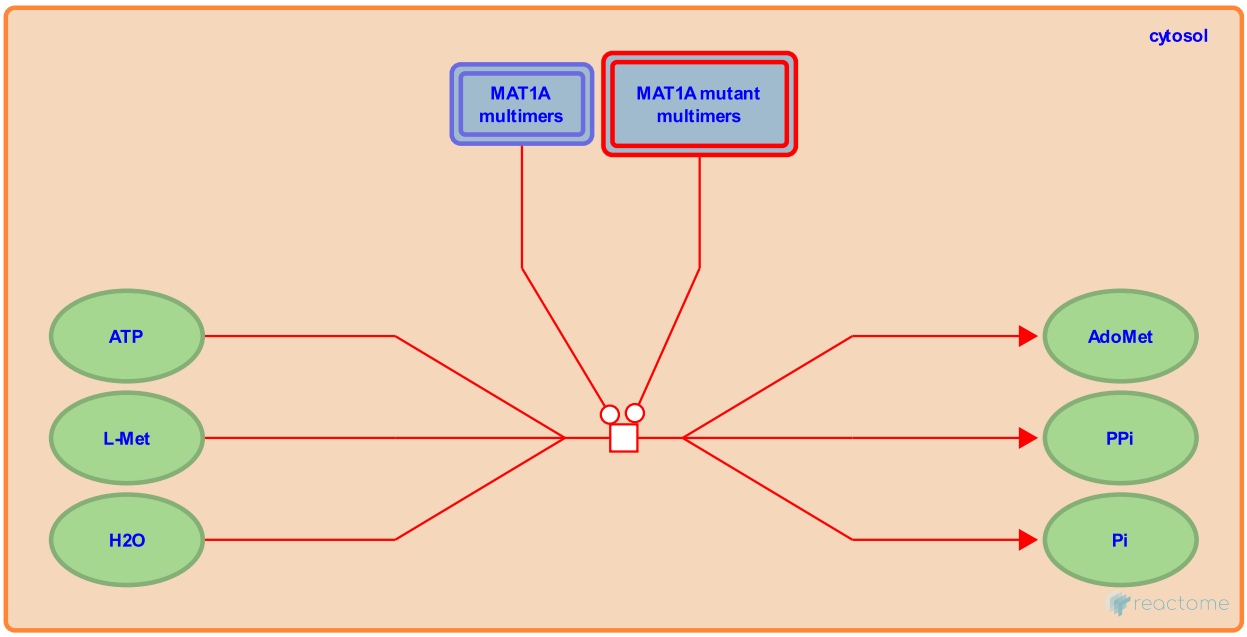
Stable identifier: R-HSA-5603087

Type: transition

Compartments: cytosol

Diseases: hypermethioninemia



S-adenosylmethionine (AdoMet, SAM) is an important methyl donor in most transmethylation reactions. S-adenosylmethionine synthase isoform type-1 (MAT1A) catalyses the formation of AdoMet from methionine and ATP. Defects in MAT1A can cause methionine adenosyltransferase deficiency (MATD; MIM:250850), an inborn error of metabolism resulting in hypermethioninemia. In this condition, methionine accumulates because its conversion to AdoMet is impaired. Frameshift mutations causing complete loss of MAT1A function are K181Vfs*5, H277Afs*75 and V348Gfs*3 (Hazelwood et al. 1998, Chamberlin et al. 1996).

Literature references

Chamberlin, ME., Mudd, SH., Chou, JY., Leonard, JV., Wilson, WG., Ubagai, T. (1996). Demyelination of the brain is associated with methionine adenosyltransferase I/III deficiency. *J. Clin. Invest., 98*, 1021-7.

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Editions

2014-06-27	Authored, Edited	Jassal, B.
2014-11-03	Reviewed	Nakaki, T.