

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

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## 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.

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## Literature references

Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics*, 18, 142. [↗](#)

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

Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res*, 46, D649-D655. [↗](#)

Fabregat, A., Korninger, F., Viteri, G., Sidiropoulos, K., Marin-Garcia, P., Ping, P. et al. (2018). Reactome graph database: Efficient access to complex pathway data. *PLoS computational biology*, 14, e1005968. [↗](#)

Reactome database release: 88

This document contains 1 reaction ([see Table of Contents](#))

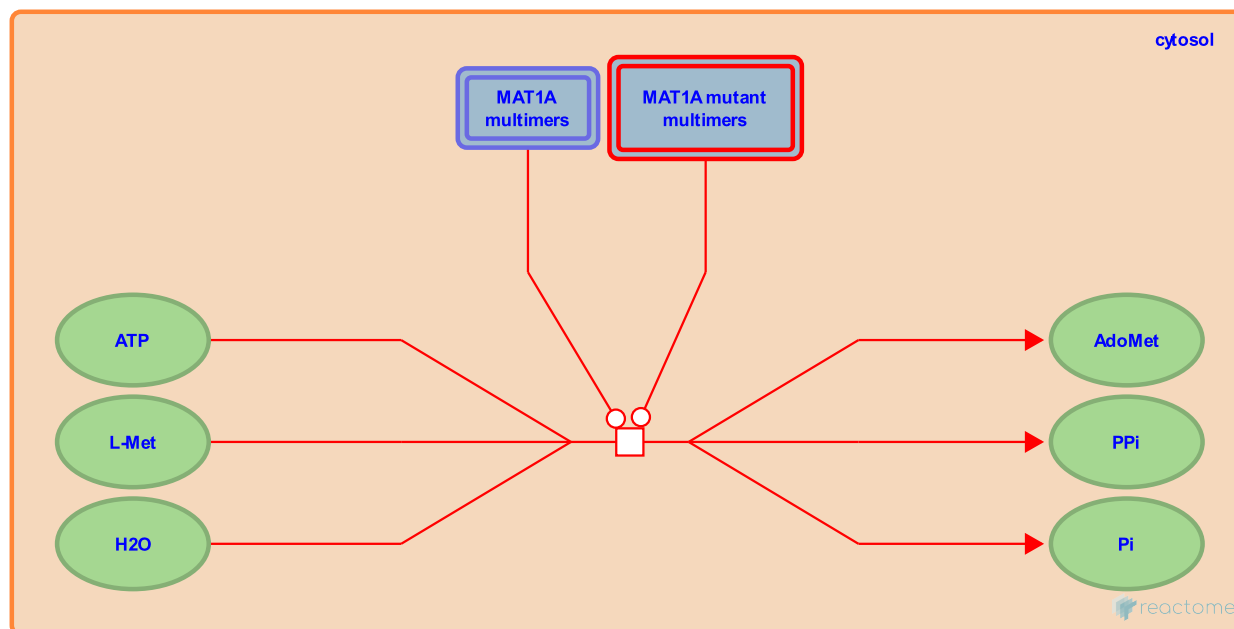
## Defective MAT1A does not transfer Ado from ATP to L-Met ↗

**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. ↗

Guo, J., Mudd, H., Bernardini, I., Hazelwood, S., Tangerman, A., Shotelersuk, V. et al. (1998). Normal brain myelination in a patient homozygous for a mutation that encodes a severely truncated methionine adenosyltransferase I/III. *Am. J. Med. Genet.*, 75, 395-400. ↗

### Editions

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