

EMG1 of the SSU processome methylates pseudouridine-1248 of 18S rRNA yielding N(1)-methylpseudouridine-1248

May, B., Sharma, S., Vincent, NG.

European Bioinformatics Institute, New York University Langone Medical Center, Ontario Institute for Cancer Research, Oregon Health and Science University.

The contents of this document may be freely copied and distributed in any media, provided the authors, plus the institutions, are credited, as stated under the terms of <u>Creative Commons Attribution 4.0 International (CC BY 4.0)</u> <u>License</u>. For more information see our <u>license</u>.

17/05/2024

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

- 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. 7
- Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics*, 33, 3461-3467. A
- 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. *オ*

This document contains 1 reaction (see Table of Contents)

EMG1 of the SSU processome methylates pseudouridine-1248 of 18S rRNA yielding N(1)-methylpseudouridine-1248 7

Stable identifier: R-HSA-6790906

Type: transition

Compartments: nucleoplasm

Inferred from: EMG1 of the SSU processome methylates pseudouridine-1191 of 18S rRNA yielding N(1)methylpseudouridine-1191 (Saccharomyces cerevisiae)



EMG1 (NEP1) methylates a pseudouridine residue in precursor rRNA (pre-rRNA) to yield N(1)methylpseudouridine (Wurm et al. 2010 and inferred from the yeast homolog) in the nucleolus (Eschrich et al. 2002). Following further modification and nucleolytic processing, the N(1)-methylpseduouridine residue will become N1methyl-N3-(3-amino-3-carboxypropyl) pseudouridine-1248 of the 18S rRNA. A mutation in EMG1 causes Bowen-Conradi Syndrome, which is characterized by growth retardation, microcephaly, severe psychomotor delay, and minor external abnormalities (Armistead et al. 2009). As inferred from the yeast homolog, EMG1 is a component of the small subunit processome (SSU processome) a large complex of proteins that binds the 5' region of pre-rRNA, processes and modifies the 18S rRNA, and assists the assembly of the small ribosomal subunit.

Literature references

- Triggs-Raine, B., Cattini, PA., Wrogemann, K., Meyer, B., Greenberg, CR., Khatkar, S. et al. (2009). Mutation of a gene essential for ribosome biogenesis, EMG1, causes Bowen-Conradi syndrome. *Am. J. Hum. Genet.*, *84*, 728-39.
- Kötter, P., Eschrich, D., Buchhaupt, M., Entian, KD. (2002). Nep1p (Emg1p), a novel protein conserved in eukaryotes and archaea, is involved in ribosome biogenesis. *Curr. Genet., 40*, 326-38. 7
- Kötter, P., Held, M., Wurm, JP., Bahr, U., Wöhnert, J., Engels, JW. et al. (2010). The ribosome assembly factor Nep1 responsible for Bowen-Conradi syndrome is a pseudouridine-N1-specific methyltransferase. *Nucleic Acids Res., 38*, 2387-98. *¬*

Editions

2015-08-15	Authored, Edited	May, B.
2016-01-30	Reviewed	Vincent, NG.
2016-02-12	Reviewed	Sharma, S.