

EMG1 gene

EMG1 N1-specific pseudouridine methyltransferase

Normal Function

The *EMG1* gene provides instructions for making a protein that is involved in the production of cellular structures called ribosomes, which process the cell's genetic instructions to create new proteins. Ribosomes are assembled in a cell compartment called the nucleolus.

The EMG1 protein is involved in the assembly of a part of the ribosome called the small subunit (SSU). In this role, the EMG1 protein functions as part of a protein complex called the SSU processome. In addition to helping to assemble the SSU, the SSU processome is involved in the maturation of a molecule called 18S rRNA, which is a chemical cousin of DNA that is incorporated into the SSU.

Health Conditions Related to Genetic Changes

Bowen-Conradi syndrome

An *EMG1* gene mutation common in the Hutterite population of the United States and Canada causes Bowen-Conradi syndrome, a disorder that affects many parts of the body and is usually fatal in the first few months of life. The mutation that causes Bowen-Conradi syndrome, written as Asp86Gly or D86G, changes the protein building block (amino acid) aspartic acid to the amino acid glycine at position 86 in the EMG1 protein. Studies suggest that the mutation makes the protein unstable, resulting in a decrease in the amount of EMG1 protein that is available in the nucleolus. A shortage of this protein in the nucleolus would impair ribosome production, which may reduce cell growth and division (proliferation); however, it is unknown how *EMG1* gene mutations lead to the particular signs and symptoms of Bowen-Conradi syndrome.

Other Names for This Gene

- 18S rRNA (pseudouridine(1248)-N1)-methyltransferase
- 18S rRNA (pseudouridine-N1-)-methyltransferase NEP1
- 18S rRNA Psi1248 methyltransferase
- C2F
- EMG1 N1-specific pseudouridine methyltransferase

- EMG1 nucleolar protein homolog
- essential for mitotic growth 1
- Grcc2f
- NEP1
- NEP1_HUMAN
- ribosomal RNA small subunit methyltransferase NEP1
- ribosome biogenesis protein NEP1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of EMG1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10436\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10436[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28EMG1%5BTIAB%5D%29+OR+%28NEP1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- EMG1 N1-SPECIFIC PSEUDOURIDINE METHYLTRANSFERASE; EMG1 (<https://omim.org/entry/611531>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10436>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=EMG1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=EMG1[gene]))

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Genomic Location

The *EMG1* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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