

ZMPSTE24 gene

zinc metallopeptidase STE24

Normal Function

The *ZMPSTE24* gene provides instructions for making a protein that acts as a protease, which is an enzyme that cuts (cleaves) other proteins. The ZMPSTE24 protein cuts an immature version of the lamin A protein (prolamin A) at a particular location; this cleavage is an essential step in the maturation of lamin A.

Mature lamin A is a component of the nuclear envelope, which is the membrane that surrounds the nucleus in cells. The nuclear envelope regulates the movement of molecules into and out of the nucleus, and researchers believe it may play a role in regulating the activity of certain genes.

Health Conditions Related to Genetic Changes

Mandibuloacral dysplasia

At least four mutations in the *ZMPSTE24* gene cause a form of mandibuloacral dysplasia called mandibuloacral dysplasia with type B lipodystrophy (MADB). This condition is characterized by a variety of signs and symptoms, which can include bone abnormalities, mottled or patchy skin coloring, and loss of fatty tissue under the skin affecting all parts of the body (type B lipodystrophy). *ZMPSTE24* gene mutations that cause MADB lead to a reduction of functional ZMPSTE24 protein. As a result, prolamin A is not processed efficiently, and it builds up in cells. In addition, there is a shortage of mature lamin A. Some researchers speculate that these changes damage the nucleus, making cells more fragile. It is not known how the effects of *ZMPSTE24* gene mutations relate to the specific signs and symptoms of MADB.

Other disorders

Mutations in the *ZMPSTE24* gene that completely eliminate the function of the ZMPSTE24 protein have been identified in newborns with a disorder called lethal restrictive dermopathy. Infants with this disorder have tight, rigid skin; underdeveloped lungs; and other abnormalities. They do not usually survive past the first week of life. Without any functional ZMPSTE24 protein, prolamin A accumulates and mature lamin A is absent; however, it is unclear how these changes lead to the severe signs and symptoms of lethal restrictive dermopathy.

Other Names for This Gene

- CAAX prenyl protease 1 homolog
- FACE-1
- FACE1
- FACE1_HUMAN
- farnesylated proteins-converting enzyme 1
- HGPS
- prenyl protein-specific endoprotease 1
- PRO1
- STE24
- Ste24p
- zinc metalloproteinase Ste24 homolog

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ZMPSTE24 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10269\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10269[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ZMPSTE24%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+1800+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- RESTRICTIVE DERMOPATHY 1; RSDM1 (<https://omim.org/entry/275210>)
- ZINC METALLOPROTEINASE STE24; ZMPSTE24 (<https://omim.org/entry/606480>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10269>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ZMPSTE24\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ZMPSTE24[gene]))

References

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Genomic Location

The *ZMPSTE24* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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