



## ADGRE2 gene

adhesion G protein-coupled receptor E2

### Normal Function

The *ADGRE2* gene provides instructions for making a protein found in several types of immune system cells, including mast cells. Mast cells, which are found in many body tissues including the skin, are important for the normal protective functions of the immune system. They also play a role in allergic reactions, which occur when the immune system overreacts to stimuli that are not harmful. The specific role of the ADGRE2 protein in mast cells is not well understood.

The ADGRE2 protein consists of two parts (subunits) that interact with each other: an alpha subunit that lies on the outside surface of the cell and a beta subunit that crosses the cell membrane and extends into the cell.

### Health Conditions Related to Genetic Changes

#### Vibratory urticaria

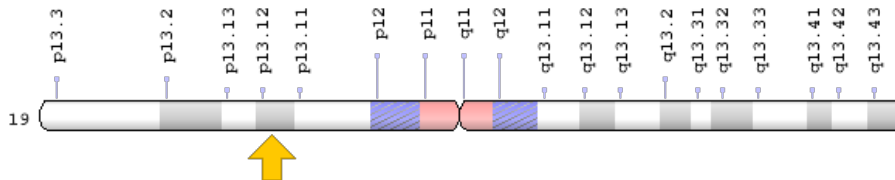
At least one mutation in the *ADGRE2* gene has been identified in people with vibratory urticaria, a condition in which vibration, repetitive stretching, or friction on the skin results in allergy symptoms such as hives (urticaria), swelling (angioedema), redness (erythema), and itching (pruritus) in the affected area. The reaction can be brought on by towel drying, hand clapping, running, a bumpy ride in a vehicle, or other repetitive stimulation.

The *ADGRE2* gene mutation that causes vibratory urticaria, written as Cys492Tyr or C492Y, replaces the protein building block (amino acid) cysteine with the amino acid tyrosine at position 492 of the protein sequence. The substitution alters the protein structure and leads to a less stable interaction between the two subunits. This fragile connection can be more easily broken; vibration, friction, or stretching of the skin can disrupt the association between subunits in mast cells. Researchers suggest that once the subunits are disconnected, the beta subunit signals the mast cells to react and produce the allergy symptoms in the skin that occur in vibratory urticaria.

## Chromosomal Location

Cytogenetic Location: 19p13.12, which is the short (p) arm of chromosome 19 at position 13.12

Molecular Location: base pairs 14,732,392 to 14,778,560 on chromosome 19 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CD312
- EGF-like module-containing mucin-like hormone receptor-like 2
- EMR2

## Additional Information & Resources

### Educational Resources

- Immunology (fifth edition, 2001): Mast Cells Reside in Tissues and Orchestrate Allergic Reactions  
<https://www.ncbi.nlm.nih.gov/books/NBK27112/#A1737>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ADGRE2%5BTIAB%5D%29+OR+%28adhesion+G+protein-coupled+receptor+E2%5BTIAB%5D%29%29+OR+%28%28CD312%5BTIAB%5D%29+OR+%28EGF-like+module-containing+mucin-like+hormone+receptor-like+2%5BTIAB%5D%29+OR+%28EMR2%5BTIAB%5D%29+OR+%28VBU%5BTIAB%5D%29+OR+%28adhesion+G+protein-coupled+receptor+E2+isoform+a+precursor%5BTIAB%5D%29+OR+%28adhesion+G+protein-coupled+receptor+E2+isoform+h+precursor%5BTIAB%5D%29%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>

## Catalog of Genes and Diseases from OMIM

- ADHESION G PROTEIN-COUPLED RECEPTOR E2  
<http://omim.org/entry/606100>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ADGRE2.html](http://atlasgeneticsoncology.org/Genes/GC_ADGRE2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ADGRE2%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:3337](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3337)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:30817>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/30817>
- UniProt  
<https://www.uniprot.org/uniprot/Q9UHX3>

## **Sources for This Summary**

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<http://omim.org/entry/606100>
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<https://ghr.nlm.nih.gov/gene/ADGRE2>

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