

## 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.

### Other Names for This Gene

- CD312

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

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of ADGRE2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=30817\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=30817[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

### Catalog of Genes and Diseases from OMIM

- ADHESION G PROTEIN-COUPLED RECEPTOR E2; ADGRE2 (<https://omim.org/entry/606100>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/30817>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ADGRE2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ADGRE2[gene]))

## References

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MonkKR, Peeters MC, Piao X, Promel S, Schoneberg T, Schwartz TW, Singer K, Stacey M, Ushkaryov YA, Vallon M, Wolfrum U, Wright MW, Xu L, Langenhan T, Schioth HB. International Union of Basic and Clinical Pharmacology. XCIV. Adhesion Gprotein-coupled receptors. *Pharmacol Rev.* 2015;67(2):338-67. doi:10.1124/pr.114.009647. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25713288>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4394687/>)

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## Genomic Location

The *ADGRE2* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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