HPSE2 gene

heparanase 2 (inactive)

Normal Function

The *HPSE2* gene provides instructions for making a protein called heparanase 2. Little is known about this protein, but its structure is similar to that of another protein called heparanase 1. Heparanase 1 is an enzyme that splits (cleaves) molecules called heparan sulfate proteoglycans (HSPGs) by removing the heparan sulfate portion (the side chain).

HSPGs are important parts of the lattice of proteins and other molecules outside the cell (extracellular matrix) and of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues. Cleavage of HSPGs by heparanase 1 may lead to changes in the basement membrane or extracellular matrix that allow cell movement or release of substances from the cell. The specific function of the heparanase 2 enzyme is not well understood, but studies suggest that it may block the action of heparanase 1.

Health Conditions Related to Genetic Changes

Migraine

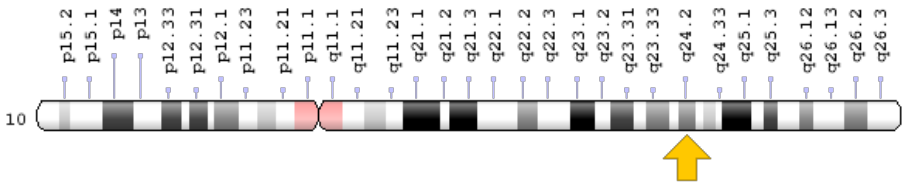
Ochoa syndrome

At least nine *HPSE2* gene mutations have been identified in people with Ochoa syndrome (also called urofacial syndrome), a disorder that causes urinary problems and unusual facial expressions. These mutations result in changes in the heparanase 2 protein that likely prevent it from functioning. The connection between *HPSE2* gene mutations and the features of Ochoa syndrome are unclear. Because the areas of the brain that control facial expression and urination are in close proximity, some researchers have suggested that the genetic changes may lead to an abnormality in this brain region that may account for the symptoms of Ochoa syndrome. Other researchers believe that a defective heparanase 2 protein may lead to problems with the development of the urinary tract or with muscle function in the face and bladder.

Chromosomal Location

Cytogenetic Location: 10q24.2, which is the long (q) arm of chromosome 10 at position 24.2

Molecular Location: base pairs 98,457,077 to 99,235,875 on chromosome 10 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- heparanase 2
- heparanase-2
- HPA2
- HPR2
- HPSE2_HUMAN

Additional Information & Resources

Clinical Information from GeneReviews

- Urofacial Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK154138

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HPSE2%5BTIAB%5D%29+OR+%28heparanase+2%5BTIAB%5D%29%29+OR+%28%28heparanase-2%5BTIAB%5D%29+OR+%28HPR2%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

- HEPARANASE 2
  http://omim.org/entry/613469

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_HPSE2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HPSE2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:18374
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:60495
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/60495
- UniProt
  https://www.uniprot.org/uniprot/Q8WWQ2

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https://ghr.nlm.nih.gov/gene/HPSE2

Reviewed: March 2012
Published: September 10, 2019

Lister Hill National Center for Biomedical Communications
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