MUC1 gene
mucin 1, cell surface associated

Normal Function

The *MUC1* gene provides instructions for making a protein called mucin 1. This protein is one of several mucin proteins that make up mucus, a slippery substance that lubricates and protects the lining of the airways, digestive system, reproductive system, and other organs and tissues. In addition to its role in mucus, mucin 1 is involved in cell signaling and kidney development.

Although most mucin proteins are released from the cell, mucin 1 spans the cell membrane. It is found in epithelial cells, which are the cells that line the surfaces and cavities of the body. In particular, mucin 1 is found in the respiratory tract, female reproductive organs, and gastrointestinal tract. Like other mucins, mucin 1 has a region called the mucin domain that contains repeated stretches of protein building blocks (amino acids); the number of repeats can vary from 20 to 100. This protein is modified by the addition of numerous chains of sugar molecules, which are attached to certain amino acids in the mucin domain. The sugars spread out from the protein like branches on a tree and prevent access to the cell surface below, protecting the body from foreign invaders. The sugars also attract water molecules, helping lubricate and hydrate the tissues.

The portion of mucin 1 that reaches inside the cell, called the cytoplasmic tail (or MUC1-CT), relays signals from outside the cell to the cell's nucleus; these signals instruct the cell to undergo certain changes. Through this process, mucin 1 is thought to be involved in cell growth and division (proliferation), helping cells stick to one another (cell adhesion), cell movement (motility), and cell survival. The cytoplasmic tail can also detach from the cell membrane and move to the nucleus, although the mechanism is unclear. Some researchers suggest that, in the nucleus, MUC1-CT helps control the activity of other genes. In addition, mucin 1 is present in cells that form the kidneys and is thought to play a role in development of these organs.

Health Conditions Related to Genetic Changes

**Medullary cystic kidney disease type 1**

Mutations in the *MUC1* gene cause medullary cystic kidney disease type 1 (MCKD1). This condition is characterized by impairment of kidney function that usually begins in adulthood and progressively worsens. Some affected individuals develop fluid-filled pockets in the kidneys called medullary cysts.

This condition occurs when a single DNA building block (nucleotide) called cytosine is inserted into the *MUC1* gene. These insertions occur in one particular region of
the gene, the part that provides instructions for the repeating mucin domain. These mutations lead to the production of an altered mucin 1 protein, although it is unclear how this change causes kidney disease. Why the effects of MUC1 gene mutations are limited to the kidneys is also unknown.

**Chromosomal Location**

Cytogenetic Location: 1q22, which is the long (q) arm of chromosome 1 at position 22

Molecular Location: base pairs 155,185,824 to 155,192,915 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- ADMCKD
- ADMCKD1
- breast carcinoma-associated antigen DF3
- CA 15-3
- cancer antigen 15-3
- carcinoma-associated mucin
- CD227
- DF3 antigen
- EMA
- episialin
- H23 antigen
- H23AG
- KL-6
- krebs von den Lungen-6
- MAM6
- MCD
• MCKD
• MUC-1
• MUC-1/SEC
• MUC-1/X
• MUC1/ZD
• MUC1_HUMAN
• mucin-1
• mucin 1, transmembrane
• peanut-reactive urinary mucin
• PEM
• PEMT
• polymorphic epithelial mucin
• PUM
• tumor-associated epithelial membrane antigen
• tumor associated epithelial mucin
• tumor-associated mucin

Additional Information & Resources

Educational Resources
• Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): The Epithelial Surfaces of the Body Are the First Defenses Against Infection
  https://www.ncbi.nlm.nih.gov/books/NBK27105/#A154

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28MUC1%5BTI%5D%29+AND+%28Genes%5BMH%5D+OR+Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22+AND+5Bdp%5D

Catalog of Genes and Diseases from OMIM
• MUCIN 1, TRANSMEMBRANE
  http://omim.org/entry/158340
Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MUC1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MUC1%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4582
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P15941

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21966898
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23361236
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3567759/
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17850209
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11439371
- OMIM: MUCIN 1, TRANSMEMBRANE
  http://omim.org/entry/158340
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16904320

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/MUC1