

EPCAM gene

epithelial cell adhesion molecule

Normal Function

The *EPCAM* gene provides instructions for making a protein known as epithelial cellular adhesion molecule (EpCAM). This protein is found in epithelial cells, which are the cells that line the surfaces and cavities of the body. The EpCAM protein is found spanning the membrane that surrounds epithelial cells, where it helps cells stick to one another (cell adhesion). In addition, the protein in the cell membrane can be cut at a specific location, releasing a piece called the intracellular domain (EpICD), which helps relay signals from outside the cell to the nucleus of the cell. EpICD travels to the nucleus and joins with other proteins, forming a group (complex) that regulates the activity of several genes that are involved in many cell processes, including growth and division (proliferation), maturation (differentiation), and movement (migration), all of which are important processes for the proper development of cells and tissues.

Health Conditions Related to Genetic Changes

Lynch syndrome

Certain variants (also known as mutations) in the *EPCAM* gene are associated with Lynch syndrome, a condition that increases the risk of developing many types of cancer, particularly cancers of the large intestine (colon) and the rectum (collectively called colorectal cancer). These variants account for up to 3 percent of Lynch syndrome cases.

On chromosome 2, the *EPCAM* gene lies next to another gene called *MSH2*. Each gene provides instructions for making an individual messenger RNA (mRNA), which serves as the genetic blueprint for making the protein. The *EPCAM* gene variants involved in Lynch syndrome remove a region that signals the end of the gene, which leads to formation of a long mRNA that includes both *EPCAM* and *MSH2*.

For unknown reasons, these *EPCAM* gene variants cause the *MSH2* gene to be turned off (inactivated) by a mechanism known as promoter hypermethylation. The promoter is a region of DNA near the beginning of the gene that controls gene activity (expression). Hypermethylation occurs when too many small molecules called methyl groups are attached to the promoter region. The extra methyl groups attached to the *MSH2* promoter reduce the expression of the *MSH2* gene, which means that less protein is produced in epithelial cells.

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Catalog of Genes and Diseases from OMIM

- EPITHELIAL CELLULAR ADHESION MOLECULE; EPCAM (<https://omim.org/entry/185535>)
- DIARRHEA 5, WITH TUFTING ENTEROPATHY, CONGENITAL; DIAR5 (<https://omim.org/entry/613217>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4072>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=EPCAM\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=EPCAM[gene]))

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Genomic Location

The *EPCAM* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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