EGFR Gene

Subjects: Genetics
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Definition

Epidermal growth factor receptor

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1. Normal Function

The *EGFR* gene provides instructions for making a receptor protein called the epidermal growth factor receptor, which spans the cell membrane so that one end of the protein remains inside the cell and the other end projects from the outer surface of the cell. This positioning allows the receptor to attach (bind) to other proteins, called ligands, outside the cell and to receive signals that help the cell respond to its environment. Ligands and receptors fit together like keys into locks. Epidermal growth factor receptor binds to at least seven different ligands. The binding of a ligand to epidermal growth factor receptor allows the receptor to attach to another nearby epidermal growth factor receptor protein (dimerize), turning on (activating) the receptor complex. As a result, signaling pathways within the cell are triggered that promote cell growth and division (proliferation) and cell survival.

2. Health Conditions Related to Genetic Changes

2.1. Cholangiocarcinoma

2.2. Lung cancer

At least eight mutations in the *EGFR* gene have been associated with lung cancer. Lung cancer is a disease in which certain cells in the lungs become abnormal and multiply uncontrollably to form a tumor. Lung cancer may not cause signs or symptoms in its early stages. Nearly all these *EGFR* gene mutations occur during a person's lifetime (somatic) and are present only in cancer cells.

Somatic mutations in the *EGFR* gene most often occur in a type of lung cancer called non-small cell lung cancer, specifically a form called adenocarcinoma. These mutations are most common in people with the disease who have never smoked. Somatic *EGFR* gene mutations occur more frequently in Asian populations than in white populations, occurring in 30 to 40 percent of affected Asians compared to 10 to 15 percent of whites with lung cancer.

Most of the somatic *EGFR* gene mutations that are associated with lung cancer delete genetic material in a part of the gene known as exon 19 or change DNA building blocks (nucleotides) in another region called exon 21. These gene changes result in a receptor protein that is constantly turned on (constitutively activated), even when it is not bound to a ligand. As a result, cells constantly receive signals to proliferate and survive, leading to tumor formation. When these genetic changes occur in cells in the lungs, lung cancer can develop. Additional genetic, environmental, and lifestyle factors contribute to a person's cancer risk.

Lung cancers with *EGFR* gene mutations tend to respond to treatments that specifically target the overactive epidermal growth factor receptor protein that allows cancer cells to constantly grow and divide.

3. Other Names for This Gene

- cell growth inhibiting protein 40
- cell proliferation-inducing protein 61
- erb-b2 receptor tyrosine kinase 1

- ERBB
- ERBB1
- HER1
- mENA
- NISBD2
- PIG61
- proto-oncogene c-ErbB-1
- receptor tyrosine-protein kinase erbB-1

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Keywords

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