

CEP290 Gene

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centrosomal protein 290

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1. Normal Function

The *CEP290* gene provides instructions for making a protein that is present in many types of cells. Although this protein's function is not well understood, studies suggest that it plays an important role in cell structures called centrosomes and cilia. Centrosomes are involved in cell division and the assembly of microtubules, which are proteins that transport materials in cells and help the cell maintain its shape. Cilia are microscopic, finger-like projections that stick out from the surface of cells. Cilia are involved in cell movement and many different chemical signaling pathways. They are also necessary for the perception of sensory input (such as vision, hearing, and smell).

2. Health Conditions Related to Genetic Changes

2.1. Leber congenital amaurosis

At least 35 mutations in the *CEP290* gene have been found to cause Leber congenital amaurosis. Mutations in this gene account for 15 to 22 percent of all cases of this condition.

A particular genetic change, written as 2991+1655A>G, is the most common *CEP290* gene mutation associated with Leber congenital amaurosis. This mutation reduces the production of functional *CEP290* protein to very low levels in cells. Other genetic changes responsible for this disorder result in the production of abnormally short, nonfunctional versions of the *CEP290* protein.

It is unclear how mutations in the *CEP290* gene cause the characteristic features of Leber congenital amaurosis. A shortage of the *CEP290* protein clearly affects the development of the retina, which is the specialized tissue at the back of the eye that detects light and color. Light-sensing cells (photoreceptors) in the retina contain cilia, which are essential for normal vision. Abnormalities involving these cilia may lead to the severe, early visual impairment characteristic of Leber congenital amaurosis.

2.2. Bardet-Biedl Syndrome

Bardet-Biedl syndrome

2.3. Joubert Syndrome

Joubert syndrome

2.4. Meckel Syndrome

Meckel syndrome

2.5. Senior-Løken Syndrome

Senior-Løken syndrome

2.6. Other Disorders

Several dozen mutations in the *CEP290* gene have been identified in other syndromes associated with abnormal cilia. These conditions, which are known as ciliopathies, include Joubert syndrome, Meckel syndrome, Senior-Løken syndrome, and Bardet-Biedl syndrome. The features of these disorders overlap significantly. They each affect multiple organ systems, most commonly the brain and spinal cord (central nervous system), retina, and kidneys. Meckel syndrome is typically the most severe of the *CEP290*-associated ciliopathies; affected individuals usually die before or shortly after birth.

The *CEP290* gene mutations responsible for these disorders lead to the production of an abnormally short version of the CEP290 protein. The abnormal protein likely disrupts cilia function in many different parts of the body. However, it is unclear how mutations in this single gene can cause multiple disorders. Researchers speculate that changes in other genes, particularly genes involved in cilia function, may contribute to the varied signs and symptoms of these conditions.

3. Other Names for This Gene

- 3H11Ag
- BBS14
- cancer/testis antigen 87
- CE290_HUMAN
- centrosomal protein 290kDa
- centrosomal protein of 290 kDa
- CT87
- CTCL tumor antigen se2-2
- FLJ13615
- FLJ21979
- JBTS5
- JBTS6
- KIAA0373
- LCA10
- MKS4
- monoclonal antibody 3H11 antigen
- nephrocytsin-6
- NPHP6
- POC3
- POC3 centriolar protein homolog
- prostate cancer antigen T21
- rd16
- SLSN6
- tumor antigen se2-2

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