CEP290 gene

centrosomal protein 290

Normal Function

The *CEP290* gene provides instructions for making a protein that is present in many types of cells, including in the eye's light receptor cells (photoreceptors). 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). The CEP290 protein is likely necessary for vision by playing a role in transporting proteins within photoreceptors.

Health Conditions Related to Genetic Changes

Leber congenital amaurosis

Many variants (also known as mutations) in the *CEP290* gene have been found to cause Leber congenital amaurosis. Leber congenital amaurosis is an eye disorder that primarily affects the retina, which is the specialized tissue at the back of the eye that detects light and color. People with this disorder typically have severe visual impairment beginning near birth or shortly afterward. Variants in the *CEP290* gene account for 15 to 22 percent of all cases of Leber congenital amaurosis.

A particular genetic change, written as 2991+1655A>G, is the most common *CEP290* gene variant associated with Leber congenital amaurosis. This variant creates a premature stop signal in the instructions for making the CEP290 protein, whichreduces the production of functional protein to low levels in cells. Other CEP290 gene changes responsible for this disorder result in the production of abnormally short, completely nonfunctional versions of the CEP290 protein.

It is unclear how variants 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. 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.

Bardet-Biedl syndrome

MedlinePlus Genetics provides information about Bardet-Biedl syndrome

Joubert syndrome

MedlinePlus Genetics provides information about Joubert syndrome

Meckel syndrome

MedlinePlus Genetics provides information about Meckel syndrome

Senior-Løken syndrome

MedlinePlus Genetics provides information about Senior-Løken syndrome

Other disorders

Several dozen variants in the *CEP290* gene have also been identified in other syndromes associated with abnormal cilia. These conditions, which are known as ciliopathies, affect many body systems and include Joubert syndrome, Meckel syndrome, Senior-Løken syndrome, and Bardet-Biedl syndrome (mentioned above). 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 variants 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 variants 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.

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CEP290 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80184[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CEP290%5BTIAB%5D%29+OR+%28NPHP6%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CENTROSOMAL PROTEIN, 290-KD; CEP290 (https://omim.org/entry/610142)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/80184)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CEP290[gene])

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Genomic Location

The *CEP290* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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