

Name:

Date:

Class:

Information Gathering Worksheet **Answer Key Leber Congenital Amaurosis**

Which genetic disorder are you researching? Leber Congenital Amaurosis

Be sure to cite your sources and use reliable sources throughout.

What are the symptoms of this disorder?

Leber congenital amaurosis, also known as LCA, is an eye disorder that is present from birth (congenital). People with this disorder typically have severe visual impairment beginning at birth or shortly afterward. The visual impairment tends to be severe and may worsen over time.

Leber congenital amaurosis is also associated with other vision problems, including an increased sensitivity to light (photophobia), involuntary movements of the eyes (nystagmus), and extreme farsightedness (hyperopia). The pupils, which usually expand and contract in response to the amount of light entering the eye, do not react normally to light. Instead, they expand and contract more slowly than normal, or they may not respond to light at all.

Source: <https://medlineplus.gov/genetics/condition/leber-congenital-amaurosis>

How common is this disorder?

Leber congenital amaurosis occurs in 2 to 3 per 100,000 newborns. It is one of the most common causes of blindness in children.

Source: <https://medlineplus.gov/genetics/condition/leber-congenital-amaurosis>

Which specific cells, tissues, and organs are affected by this disorder?

This condition primarily affects the retina, which is the specialized tissue at the back of the eye that detects light and color. Leber congenital amaurosis can result from variants (also known as mutations) in at least 20 genes, all of which are necessary for function of the retina and normal vision. These genes play a variety of roles in the development and function of the retina. For example, some of the genes associated with this disorder are necessary for the normal development of light-detecting cells called photoreceptors. Other genes are involved in phototransduction, the process by which light entering the eye is converted into electrical signals that are transmitted to the brain. Still other genes play a role in the function of cilia, which are microscopic finger-like projections that stick out from the surface of many types of cells. Cilia are found in the retina's photoreceptors and are necessary for vision.

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If applicable, what are the current treatments? How successful are they?

Currently, there is no way to prevent or stop vision loss in LCA in most patients. However, there are new treatments being tested in clinical trials, including gene therapy, oral medications, and eye injections, that show great promise.

Source: <https://rarediseases.org/rare-diseases/leber-congenital-amaurosis>

What specific gene is mutated in people with this disorder?

Variants in any of the genes associated with Leber congenital amaurosis disrupt the development and function of the retina, resulting in early vision loss. Variants in the CEP290, CRB1, GUCY2D, and RPE65 genes are the most common causes of Leber congenital amaurosis, while variants in the other genes generally account for a smaller percentage of cases. In about 30 percent of all people with Leber congenital amaurosis, the cause of the disorder is unknown, though research is ongoing.

Source: <https://medlineplus.gov/genetics/condition/leber-congenital-amaurosis>

Is this mutation dominant or recessive?

Leber congenital amaurosis usually has an autosomal recessive pattern of inheritance.

Source: <https://medlineplus.gov/genetics/condition/leber-congenital-amaurosis>

What specific mutation causes this disorder?
List the healthy and mutated DNA sequence below.
Include at least 15 nucleotides before and after the mutation.

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, which reduces the production of functional protein to low levels in cells.

Healthy sequence:

5' GACTGCCAATAGGGATAGGTATGAGATATTCACAATTACAACCTGGCCAGG 3'

Mutated sequence:

5' GACTGCCAATAGGGATAGGTATGAGATACTCACAATTACAACCTGGCCAGG 3'

Source: <https://medlineplus.gov/genetics/gene/cep290/#conditions>
<https://www.ncbi.nlm.nih.gov/snp/rs281865192>