NYX Gene

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nyctalopin

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1. Introduction

The *NYX* gene provides instructions for making a protein called nyctalopin, which plays an important role in a specialized tissue at the back of the eye called the retina. Within the retina, nyctalopin is located on the surface of light-detecting cells called photoreceptors. The retina contains two types of photoreceptors: rods and cones. Rods are responsible for vision in low light. Cones provide vision in bright light, including color vision.

Nyctalopin appears to play a critical role in normal vision. Studies suggest the protein helps relay visual signals from rods and cones to other retinal cells called bipolar cells. This signaling is an essential step in the transmission of visual information from the eyes to the brain.

2. Health Conditions Related to Genetic Changes

2.1. X-linked congenital stationary night blindness

More than 50 mutations in the *NYX* gene have been identified in people with X-linked congenital stationary night blindness. Mutations in this gene are responsible for the complete form of the disorder, which is characterized by difficulty seeing in low light (night blindness), loss of sharpness (reduced acuity), severe nearsightedness (high myopia), involuntary movements of the eyes (nystagmus), and eyes that do not look in the same direction (strabismus).

Many *NYX* mutations change single protein building blocks (amino acids) in nyctalopin. *NYX* mutations can change the size or shape of the protein or prevent it from attaching to the surface of photoreceptor cells. A loss of functional nyctalopin disrupts the ability of photoreceptor cells to transmit visual signals, which impairs vision. The function of rods is severely disrupted, while the function of cones is only mildly affected.

2.2. Other disorders

At least two mutations in the *NYX* gene have been found to cause high myopia without the other vision problems characteristic of X-linked congenital stationary night blindness. The mutations responsible for high myopia each change a single amino acid in nyctalopin, which is predicted to result in an unstable protein. Researchers are uncertain why these mutations cause high myopia without any other vision abnormalities.

3. Other Names for This Gene

- CLRP
- CSNB1
- CSNB1A
- CSNB4
- leucine-rich repeat protein
- MGC138447
- NYX_HUMAN

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