Research Team Discovers New Gene

Discovery Unravels Mystery of Night Blindness

Today, one in every 10000 Canadians is afflicted with a hereditary disease known as Congenital Stationary night blindness (CNSB). CNSB, a rare X-liked disorder, leaves its host unable to see in the dark, and unable to navigate without the help of an escort. This elusive and debilitating illness has left researchers and ophalthamologists puzzled for many years. That is, until now.

An international team of almost 50 collaborators led by University of Calgary's Dr. Torben Bech-Hansen have isolated the gene for CNSB, and after 13 years of research have discovered a new understanding of night blindness, establishing a new sense of hope for a cure.

Bech-Hansen revealed in a press conference on October 30th the epidemiological study involving the analysis of DNA from 22 families across North America and Europe whose members were afflicted with complete CNSB. After isolating the mutated gene, named NYX, the researchers were able to identify one of its products, a defective protein that they named nyctalopin. "When the nyctolopin protein is mutated, normal neural connection within the retina is not achieved, leading to night blindness" explains Dr. Bech-Hansen.

While a cure may still be many years away, the discovery marks an important step for accurately diagnosing those individuals afflicted with the disease. In addition to a definitive diagnostic test, the research has provided a foundation of understanding about how nerves connect in the retina, and perhaps even more exciting, a first glimpse of how to stimulate retinal regeneration.

Bech-Hansen made reference to the significance of his research collaborators, the benefit of having an interdisciplinary team in achieving this discovery and the importance of funding agencies. Financial support for this latest night blindness discovery was received from the Canadian Foundation Fighting Blindness, The Canadian Institutes of Health Research, the I.D Bebensee Foundation, the Lion's Site center, and the Alberta Children's Hospital Foundation.

The findings of this study are scheduled to be published in the prestigious journal *Nature Genetics* on November 1, 2000. This publication follows a previous article in *Nature Genetics* by Bach-Hansen in 1998 when he identified another gene important for the understanding of impaired night vision.

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