

Examining what causes spontaneous eye movements in albinism

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Infographic: Cause of spontaneous eye movements in albinism. Credit: Netherlands Institute for Neuroscience - Eline Feenstra

People with albinism often have poor vision. A new study from the Netherlands Institute for Neuroscience reveals the underlying cause.



In Europe, albinism occurs in 1 in 20,000 individuals. However, in some populations, it is much more common, affecting 1 in 1,000 people. People with albinism lack pigment and frequently have poor eyesight. For example, they may see at a distance of 20 meters what someone with pigment sees at 80 meters. But what exactly causes this?

One of the causes is the spontaneous back-and-forth movement of the eye, called 'pendular nystagmus.' This phenomenon not only makes seeing difficult but also hinders social eye contact. Treatment sometimes involves medications or surgery on the eye muscles, but these methods have unpleasant side effects and are not fully effective. Understanding the underlying mechanisms behind this condition is essential for developing alternative treatment strategies.

Moving train

Pendular nystagmus resembles the eye movements people make when looking outside while riding a moving train: the eyes automatically move along with the moving landscape and then spring back to the resting position. Nerve cells in a small <u>brain</u> area (known as the nucleus of the optic tract) selectively respond to this movement and become active. In healthy individuals, this activity leads to the tightening of the eye muscles to stabilize the image. However, in <u>albinism</u>, this process works a bit differently.

Jorrit Montijn, Valentina Rugiccini and their colleagues, under the supervision of Alexander Heimel, have now demonstrated in albino mice that the cells in this brain area are no longer selective for the direction of image <u>movement</u>. As a result, the image cannot stabilize, leading to the condition of pendular nystagmus. Currently, surgery of this brain area is not possible, but the research provides hope that in the future, pendular nystagmus can be reduced through manipulation of activity in this brain region.



Ruling out cortex

Jorrit Montijn: "We show that the nucleus of the optic tract might be the source of the problem. Previous research already suggested that this area is involved in eye movements, but it could not be ruled out that (also) other areas, such as the cortex, cause pendular nystagmus. By simultaneously measuring both the cortex and the nucleus of the optic tract in the same mice, we were able to eliminate this question."

"We now know that there is something wrong with this area, but we still don't know what can be done about it. The next steps would be to translate this into practice. One possible option could be Deep Brain Stimulation of the area, but this still needs to be tested, and it is not known if it has an effect. Another option is perhaps surgery or even gene therapy in the future. It is now up to more clinically oriented scientists to investigate this."

The paper is published in the journal *Investigative Opthalmology* & *Visual Science*.

More information: Jorrit S. Montijn et al, Impaired Direction Selectivity in the Nucleus of the Optic Tract of Albino Mice, *Investigative Opthalmology & Visual Science* (2023). DOI: 10.1167/iovs.64.11.9

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