

PRESS RELEASE

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WHAT CAUSES INHERITED DEGENERATION OF THE RETINA?

A team of scientists from the Institute of Molecular Genetics of the Czech Academy of Sciences (IMG) has described in their recent research how mutations causing degeneration of the human retina affect neurons in the mouse cerebellum. This is another step towards understanding the development of an inherited degenerative disorder called retinitis pigmentosa. Their results were published in the Life Science Alliance journal a few days ago.

The human DNA contains approximately 20,000 genes, which are like "pages" in our "genetic book". Each page contains instructions for the production of a specific protein. For example, light-sensitive cells in the eye synthesize proteins important for detecting light based on this "genetic book". Before the protein is made, the information from DNA is transcribed into a molecule called pre-mRNA, which is an exact copy of the DNA page. Only a small part (about one-tenth) contains the information for the production of the protein, while the rest is removed in a process called RNA splicing. RNA splicing is carried out by huge molecular scissors called the "spliceosome". Mutations in some of the proteins that make up the spliceosome cause inherited degeneration of the retina - retinitis pigmentosa. This disease cannot be treated currently, and we do not even know why the disease affects the retina specifically.

To better study this disease, David Staněk's team from the IMG inserted mutations causing retinitis pigmentosa into laboratory mice. It turned out that mutations that kill neurons in the human retina affect neurons in the mouse cerebellum. "For the first time, we saw the death of neurons almost in direct transmission, which helped us uncover what is happening in these cells and why they die," explains David Staněk. This unique model will be used in the future to test new procedures for the treatment of retinitis pigmentosa.

More information:

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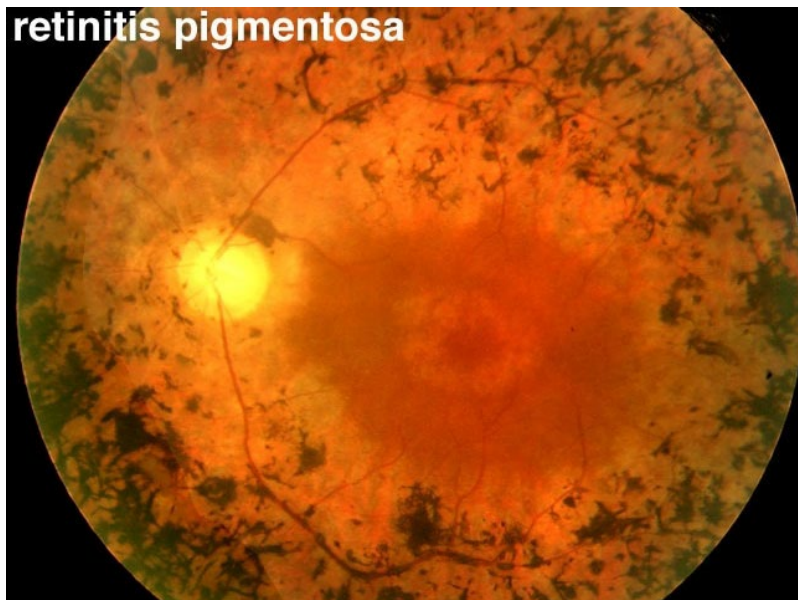
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Retina of an eye affected by degeneration (retinitis pigmentosa). Source: Webvision: The Organization of the Retina and Visual System



Cerebellum affected by degeneration. Source: Krausová et al. (2023), Life Science Alliance