Study of rare genetic disorder that effects the eyes

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Nagano prefecture is home to a group of people affected with a rare genetic neurodegenerative disorder called familial amyloid polyneuropathies (FAP). This disease impacts the gene encoding protein transthyretin (TTR) which is produced in the liver and also eyes. Liver transplants are often a treatment for this disease, but severe eyesight problems such as cloudiness and glaucoma remains, despite such procedures. This research is a retrospective observational study of what ophthalmologists have experienced in their practice over the years.

A group of doctors at Shinshu University Hospital decided to put together this study with the hope that sharing information with other researchers and doctors around the world might help further and bolster understanding and treatment for this eye disease, TTR-related FAP, which is referred to as hereditary ATTR amyloidosis, an autosomal dominant disorder.

The team led by Shinji Kakihara strived to articulate how doctors have cared for patients who have this disease and share issues that need addressing as well as positive outcomes from their care, on account of the fact that many ophthalmologists remain unaware of TTR-related FAP.

Patients are usually diagnosed with hereditary transthyretin amyloidosis by age 50 and have an overall survival rate of less than 10 years. Through accurate diagnosis by the Third Department of Internal Medicine (Neurology), patients were referred to the ophthalmology medical team of Shinshu University. Doctor Teruyoshi Miyahara examined many of the patients over the years.

Small gauge vitrectomy, or surgery on the eyes improved eyesight which was maintained until the last visit with the ophthalmologist when intraocular pressure control was adequate. However, subsequent glaucoma surgeries were needed if the intraocular pressure was not controlled, because pressure rapidly increases after vitrectomy and advanced the progression of glaucoma.

This is a very rare disease, but there are patients around the world who suffer from hereditary transthyretin amyloidosis. Doctors at Shinshu University are hopeful that their experience can be utilized to help protect the eyesight of people around the world.

They hope to continue research to see how they can further improve the eyes of people with familial amyloid polyneuropathy. This research is about a very rare disease, but many people suffer from glaucoma. The doctors hope to employ their understanding by continuing research of FAP as well as glaucoma.

More information: Shinji Kakihara et al, Small gauge vitrectomy for vitreous amyloidosis and subsequent management of secondary glaucoma in patients with hereditary transthyretin amyloidosis, *Scientific Reports* (2020). DOI: 10.1038/s41598-020-62559-x

Provided by Shinshu University

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