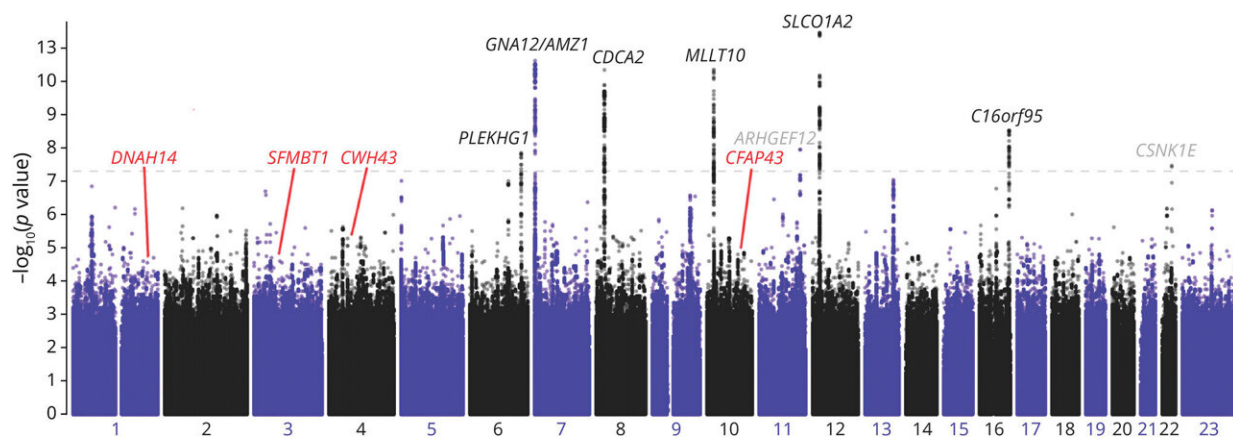


Pioneering study identifies six genetic regions associated with normal pressure hydrocephalus

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Manhattan Plot of the NPH genome-wide association study. Credit: *Neurology* (2024). DOI: 10.1212/WNL.0000000000209694

A new study by the University of Eastern Finland and partners identified new genetic variants associated with normal pressure hydrocephalus (NPH). Published in *Neurology*, the [study](#) is the first large-scale genome-wide association study related to NPH in the world.

The results provide new insights into the genetic background of NPH and provide a basis for further research into the specific biological mechanisms underlying the condition.

NPH is a chronic neurological syndrome in the elderly that affects gait, memory and bladder control. The current treatment for NPH is shunt surgery.

The disease mechanisms of NPH remain unclear, and its genetics have been studied only in a limited context. However, the idiopathic form of NPH has been observed to occur in families to some extent, and previous genetic studies have identified individual variants that increase the risk of NPH.

The study was based on the Finnish FinnGen research project, which includes genomic data from Finnish participants combined with health registry information. The [genome-wide association study](#) included 1,522 NPH patients and more than 450,000 controls. The replication cohort was obtained from the UK Biobank.

"We are extremely grateful to the NPH patients and their families participating in the study," says Professor Ville Leinonen of the University of Eastern Finland. The recruitment of the NPH patient cohort began in Kuopio, Finland, more than 15 years ago.

The NPH patient cohort was augmented through national neurosurgical collaboration, but the present study was made possible only after more recent collaboration with the FinnGen project.

The study identified significant variants associated with NPH in six different gene loci. Some of these genes have previously been found to associate with the structures or functions of brain areas significant for NPH.

These include roles in the blood-cerebrospinal fluid barrier and the [blood-brain barrier](#) as well as associations with increased size of the brain lateral ventricles in the general population, which is also a key

finding in NPH.

"The newly identified gene loci provide a biologically relevant foundation for further investigations into disease mechanisms in NPH," says Professor Mikko Hiltunen, whose research group collaborates closely with Professor Ville Leinonen's NPH research group at the University of Eastern Finland.

The findings support previous assumptions of NPH being a multifactorial disease.

"A genome-wide study of this scale in NPH is extremely unique, and our findings are highly essential for further research," says Joel Räsänen, a Doctoral Researcher at the University of Eastern Finland and the NPH research group.

More information: Joel Räsänen et al, Risk Variants Associated With Normal Pressure Hydrocephalus, *Neurology* (2024). [DOI: 10.1212/WNL.0000000000209694](https://doi.org/10.1212/WNL.0000000000209694)

Provided by University of Eastern Finland

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