Familial idiopathic normal pressure hydrocephalus

Citation

Huovinen, J., S. Kastinen, S. Komulainen, M. Oinas, C. Avellan, J. Franzen, J. Rinne, et al. 2015. "Familial idiopathic normal pressure hydrocephalus." *Fluids and Barriers of the CNS* 12 (Suppl 1): 043. doi:10.1186/2045-8118-12-S1-043. <http://dx.doi.org/10.1186/2045-8118-12-S1-043>.

Published Version

doi:10.1186/2045-8118-12-S1-043

Permanent link

<http://nrs.harvard.edu/urn-3:HUL.InstRepos:22857083>

Terms of Use

This article was downloaded from Harvard University's DASH repository, and is made available under the terms and conditions applicable to Other Posted Material, as set forth at <http://nrs.harvard.edu/urn-3:HUL.InstRepos:dash.current.terms-of-use#LAA>

Share Your Story

The Harvard community has made this article openly available.
Please share how this access benefits you. [Submit a story](#).

[Accessibility](#)

ORAL PRESENTATION

Open Access

Familial idiopathic normal pressure hydrocephalus

Joel Huovinen^{1*}, Sami Kastinen¹, Simo Komulainen¹, Minna Oinas², Cecilia Avellan², Janek Franzen³, Jaakko Rinne³, Antti Ronkainen⁴, Mikko Kauppinen⁵, Kimmo Lönnroth⁶, Markus Perola⁷, Okko T Pyykkö¹, Anne M Koivisto⁸, Anne M Remes⁸, Mikko Hiltunen^{8,9}, Seppo Helisalmi⁸, Mitja Kurki^{1,10}, Juha E Jääskeläinen¹, Ville Leinonen¹

From Hydrocephalus 2015
Banff, Canada. 18-21 September 2015

Introduction

Idiopathic normal pressure hydrocephalus (iNPH) is a late-onset, surgically alleviated, progressive neurodegenerative disease with unknown etiology. There are few studies describing pedigrees with multiple affected relatives. Our aim was to identify and characterize a potential familial subgroup of idiopathic normal pressure hydrocephalus in a nation-wide Finnish cohort

Methods

Overall 375 iNPH-patients operated between 1993 and 2014 were questioned and phone interviewed, whether they have relatives with either diagnosed iNPH or disease-related symptomatology. Genograms of families with such findings were drawn.

Results

60 patients (16 %) had potential familial iNPH. 18 patients from 12 separate pedigrees had at least one relative shunted due to iNPH. Patients with familial iNPH reported a complete triad of NPH-symptoms ($p=0.03$) and memory problems ($p=0.014$) more often than sporadic cases. Both shunted and symptomatic relatives were mainly first-degree.

According to age-adjusted multivariate logistic regression analysis diagnosed dementia (odds ratio [OR] 2.9; 95% confidence interval [CI], 1.5–5.4) and nonarthritic rheumatoid etiologies (OR, 4.4; 95% CI, 1.6–11.7) were more frequent in familial than sporadic patients. Geographical variation in the occurrence of iNPH was observed, the incidence being highest in Eastern Finland. Frequency of APOE epsilon 4 as well as diagnosed Alzheimer's

disease (AD) and AD-medication were similar in familial and sporadic iNPH-patients.

Conclusions

This study indicates a familial entity of iNPH offering a novel approach to discover the potential genetic characteristics of iNPH. Furthermore, these pedigrees offer an intriguing opportunity to conduct longitudinal studies focusing on potential preclinical signs of iNPH. Our findings support iNPH as a specific neurodegenerative disease.

Authors' details

¹Department of Neurosurgery, Kuopio University Hospital and Neurosurgery, Institute of Clinical Medicine, University of Eastern Finland, Kuopio, Finland. ²Department of Neurosurgery, University of Helsinki and Helsinki University Hospital, Finland. ³Clinical Neurosciences, Department of Neurosurgery, Turku University Hospital, Turku, Finland. ⁴Department of Neurosurgery, Tampere University Hospital, Tampere, Finland. ⁵Department of Neurosurgery, Oulu University Hospital, Oulu, Finland. ⁶South Ostrobothnia Central Hospital, Seinäjoki, Finland. ⁷National Institute for Health and Welfare, Finland and University of Helsinki, Helsinki, Finland. ⁸Unit of Neurology, Institute of Clinical Medicine, University of Eastern Finland, Kuopio, Finland and Department of Neurology, Kuopio University Hospital, Kuopio, Finland. ⁹Institute of Biomedicine, University of Eastern Finland, Kuopio, Finland. ¹⁰Analytical and Translational Genetics Unit, Department of Medicine, Massachusetts General Hospital, USA, Program in Medical and Population Genetics, Broad Institute of MIT and Harvard, USA Stanley Center for Psychiatric Research.

Published: 18 September 2015

doi:10.1186/2045-8118-12-S1-O43

Cite this article as: Huovinen et al.: Familial idiopathic normal pressure hydrocephalus. *Fluids and Barriers of the CNS* 2015 **12**(Suppl 1):O43.

* Correspondence: jhuovine@student.oulu.fi

¹Department of Neurosurgery, Kuopio University Hospital and Neurosurgery, Institute of Clinical Medicine, University of Eastern Finland, Kuopio, Finland
Full list of author information is available at the end of the article