Familial idiopathic normal pressure hydrocephalus

Citation

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

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
Familial idiopathic normal pressure hydrocephalus

Joel Huovinen1*, Sami Kastinen1, Simo Komulainen1, Minna Oinas2, Cecilia Avellan1, Janek Franzen3, Jaakko Rinne3, Antti Ronkainen4, Mikko Kauppinen4, Kimmo Lönnroth5, Markus Perola7, Okko T Pykkö7, Anne M Koivisto8, Anne M Remes8, Mikko Hiltunen8,9, Seppo Helisalmi8, Mitja Kurki1,10, 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 questionnaire 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.