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Editorial

Familial Normal Pressure Hydrocephalus: A Novel Subgroup

Takeo Kato*, Yoshimi Takahashi, and Toru Kawanami

Department of Neurology, Hematology, Metabolism, Endocrinology and Diabetology, Yamagata University Faculty of Medicine, Yamagata 990-9585, Japan

Corresponding author

Takeo Kato, Department of Neurology, Hematology, Metabolism, Endocrinology and Diabetology, Yamagata University Faculty of Medicine, 2-2-2 Iida-nishi, Yamagata 990-9585, Japan, Tel: +81-23-628-5316; Fax: +81-23-628-5318; E-mail: tkato@med.id.yamagata-u.ac.jp

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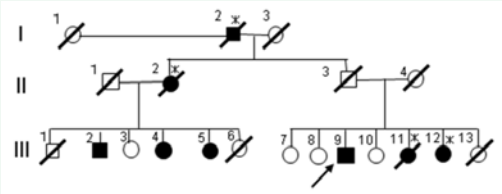
EDITORIAL

Normal pressure hydrocephalus (NPH) was first reported by Hakim and Adams in 1965 [1,2] and is characterized by the clinical triad (cognitive impairment, gait disturbance, and urinary incontinence), ventricular dilatation of the brain, normal pressure of the cerebrospinal fluid (CSF), and symptomatic improvement by CSF shunting [1-4]. NPH is classified into two subgroups: idiopathic NPH (iNPH) and secondary NPH (sNPH) of known etiology. sNPH occurs several weeks or months after subarachnoid hemorrhage, meningitis, traumatic brain injuries, and certain other illnesses. iNPH occurs without any preceding diseases, and its cause remains undetermined. Most textbooks and guidelines for NPH describe iNPH and sNPH. However, little has been described about familial occurrences of NPH. Recently, cases of familial NPH have become known. Therefore, we prepared a review on familial NPH.

The first report on familial NPH was described by Portenoy et al. (1984), who showed two sibling cases (a 67-year-old man and his 74-year-old sister) with NPH [5]. In both patients, the ventricles of the brain were dilated. In each case, neurological symptoms were improved by a CSF shunt operation. In 2011, Cusimano et al. also reported two siblings (a 71-year-old woman and her 73-year-old sister) with NPH, who were shunt responsive [6]. Occurrence of NPH in siblings does not always indicate the presence of genetic predisposition in the etiology and/or pathogenesis of NPH. It also suggests a yet-unknown, common environmental factor causing the disease. In 2011, we reported decisive evidence on this matter by showing a large family that included 8 patients with elderly-onset NPH in three generations, which is consistent with an autosomal-dominant inheritance (Figure 1). Each affected member of the family had neurological symptoms and brain MRI features that were indistinguishable from those of iNPH [7]. In 2012, McGirr et al. also reported 4 cases (proband, uncle, and cousins) of NPH in two generations [8]. At the present time, eight additional families with NPH have been identified in Japan, suggesting the possibility that a considerable number of patients with familial NPH would be detectable if a family history was carefully examined.



○/□: normal woman/man, ●/■: woman/man with NPH, /: dead

Figure 1 Pedigree of our NPH family. Four patients (III-2, III-4, III-5, and III-9) were examined both neurologically and on brain MRI, and were diagnosed as having NPH. III-2 and III-5 underwent a CSF tap test and their neurological symptoms improved. III-2 underwent a CSF shunt operation, resulting in improvement of his neurological symptoms. In the remaining four patients (*), NPH was suspected on the basis of medical interview. The arrow (III-9) indicates the proband.

Many diseases are classified into "idiopathic," "secondary," and "familial." For example, parkinsonism is classified into idiopathic parkinsonism (Parkinson's disease), secondary parkinsonism, and familial parkinsonism. We now know that a novel subgroup of NPH, familial NPH (fNPH), exists. Therefore, NPH should be classified into three subgroups: iNPH, sNPH, and fNPH.

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