

## OCCURRENCE OF NORMAL PRESSURE HYDROCEPHALUS IN TWO SIBLINGS

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Normal pressure hydrocephalus (NPH) was first described about three decades ago.<sup>1,2</sup> However, its mechanisms are still incompletely understood and possible cause is found in less than 50% of the cases.<sup>1,3,4</sup> A genetic mechanism, at least in some of these cases, was considered when Portenoy et al. described the occurrence of the condition in two siblings four years apart.<sup>5</sup>

We had the opportunity to observe another set of two siblings with NPH managed in our hospital, representing the second familial association to be documented in the literature.

### Case Reports

#### Case 1

A 66-year-old Saudi male was admitted to our hospital in 1993 with a history of urinary incontinence of 12 months' duration, progressive difficulty in walking, and memory disturbances for the previous six months. He complained also of frequent falls in the previous three months. His past history was unremarkable, apart from investigations for a probable urological cause for his incontinence, which were negative. He was not a known hypertensive or diabetic. There was no family history of dementia.

On examination, there was a clear deficit in the three-item registration and recall, as well as in recent memory, but other cognitive domains were normal. He had great difficulties in walking, with poor equilibrium and a wide-based gait. Tandem gait was not possible. He had positive snout, sucking and palmomental reflexes. There were no abnormalities in segmental power, tone, sensations, or tendon reflexes and there were no cerebellar signs or cranial nerve palsy.

Brain CT scan revealed dilated ventricles, while MRI showed no aqueduct stenosis or posterior fossa

abnormality (Figure 1). Usual blood work was normal. The opening pressure at lumbar puncture was 150 mm H<sub>2</sub>O and the CSF was essentially normal. Gait was mildly improved after subtraction of 30 cc of CSF.

A low-pressure ventriculoperitoneal shunt was inserted. When the patient was examined three months later, his gait was much better and he could recall two to three items without difficulty. A follow-up CT scan done on review revealed reduction in the ventricular size.

#### Case 2

This 50-year-old female was the only living sister of the above patient. She was seen in November 1994 with complaints of progressive difficulty in walking for the previous eight months. There was no history of cognitive decline or sphincter disturbances. She had been hypertensive for several years and had been treated by atenolol 100 mg daily. On clinical examination, she had a normal mental state, needed a cane to walk, and her gait was wide-based. Tandem gait was impossible. There were no primitive reflexes. The motor power was normal, with slightly increased reflexes with flexor plantar response. There was no sensory deficit or incoordination.

CT scan of the brain revealed dilated ventricles with minimal cerebral atrophy. MRI did not show evidence of obstruction in the brain stem or aqueduct stenosis. There were no ischemic brain changes. The patient's CSF opening pressure was 170 mm H<sub>2</sub>O with normal glucose and protein and no cells. Her WBC, hemoglobin and serum chemistry were unremarkable. She also had a low pressure shunt inserted with noticeable improvement in gait after three months. Repeat CT scan also showed reduction in ventricular size.

### Discussion

Normal pressure hydrocephalus is a rare disease which is often overdiagnosed, since gait disorders and large cerebral ventricles are quite frequent in elderly people. The diagnosis is, however, easier when the classical clinical triads, including the typical gait disorder, are present and when the condition of the patient improves

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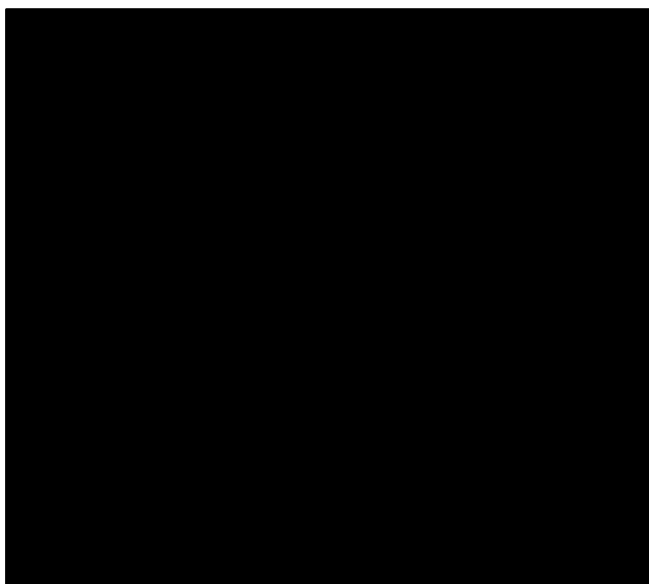


FIGURE 1. MRI of the patient in case 1 showed a moderate cerebellar atrophy with hydrocephalus and normal aqueduct canal.

after ventricular shunting. This was the case in our first patient. The clinical presentation was limited to a gait disorder in his sister, but this presentation is not uncommon. Of 16 cases of NPH improved by shunt and reported by Fisher in 1978,<sup>6</sup> 12 were revealed by isolated gait disturbances.

It is difficult to ascribe familial occurrence of NPH solely to a genetic defect or an inherited disorder with long latency that slowly evolved until the seventh decade. But coincidence is also unlikely in view of the rarity of the disease. It is possible that our patients had long-standing hydrocephalus which was unmasked when neuronal attrition and/or possible neurotransmitter dysfunctions

progressed beyond a critical threshold. In 1984, Portenoy et al. speculated on the possibility of a rare, genetically-determined form of late-onset NPH.<sup>5</sup> To our knowledge, no other such cases have been published since then. Our documentation of a second set of familial cases in their sixties would appear to give this speculation some credence. We attempted to screen other members of the family, but only the oldest son of the second case could be traced and he was completely asymptomatic with normal brain CT scan.

In conclusion, familial NPH seems rare but does exist. Its mechanisms are poorly understood. However, it warrants further studies with the powerful tool of molecular biology.

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