P 3-112

Dysphagia in Wilson's Disease: a case report of one year follow-up

Doyoung Kim[1*], Juntaek Hong[2], Jun Min Cha[3], Jung Hyun Park[1], Yoon Ghil Park[1], Jinyoung Park[1†]

Department of Rehabilitation Medicine, Gangnam Severance Hospital, Rehabilitation Institute of Neuromuscular Disease, Yonsei University College of Medicine, Seoul, Korea[1], Department of Rehabilitation Medicine, Severance Hospital, Yonsei University College of Medicine, Seoul, Korea[2], Department of Physical Medicine and Rehabilitation, National Health Insurance Service Ilsan Hospital[3]

Introduction

Wilson's disease (WD) is an autosomal recessive disease caused by mutations in the ATP7B gene. Impaired copper metabolism due to this mutation leads to metal accumulation in various tissues and organs, including the central nervous system. Although dysphagia is the earliest manifestation of the neurologic dysfunction in this disease, the swallowing dynamics remains unclear. In this report, we present a case of WD with associated dysphagia and its one year follow-up after treatment.

Case report

A 17-year-old man visited our hospital with a complaint of difficulty in swallowing and general weakness. A general weakness developed about 1 month before, and aphasia and dysphagia occurred in the last 5 days. After the admission to the Department of Pediatrics, he was diagnosed with Wilson's disease by mutation of the ATP7B gene. The physical examination showed that the cognitive function was normal while dysarthria and dysphagia was evident. Cerebellar function tests showed severe tremor in both upper extremities and dysdiadochokinesia in both finger to nose test. The initial manual muscle test showed good grade in all extremities without muscle atrophy. Low serum copper and ceruloplasmin level (28.4 ug/dL and < 4 mg/dL) were consistent with WD. The brain magnetic resonance imaging (MRI) on T2WI showed high signal intensity in both the pons and the midbrain (Figure 1A). Videofluoroscopic swallowing study (VFSS) findings were evaluated using videofluoroscopic dysphagia scale (VDS). The initial VDS score was 48. Incomplete lip closure was seen in the oral preparatory phase. Decreased tongue movement and posterior propelling were seen in the oral phase with delayed swallowing reflex. After the VFSS, the patient was treated with 15 sessions of neuromuscular electrical stimulation therapy (NMES) with the electrical pads attached horizontally just above the hyoid bone and thyroid notch. The stimulating intensity was increased from 5 mA to 6.5 mA. Double swallowing and chin tuck posture was educated. The patient continued to take the D-penicillamine(PO, 250mg QID) for the treatment of WD. One year after the symptom onset, brain MRI showed decreased signal intensity in the pons and the midbrain on T2WI (Figure 1B) in compared with initial brain MRI. By follow-up VFSS, the VDS score was improved to 23 (Table 1). Mastication, tongue thrust, and piecemeal deglutition were improved compared to the initial swallowing function. In the pharyngeal phase, the amount of vallecular residue was reduced (Figure 2).

Discussion

Our report shows that WD patients present prolonged oral transit duration and greater percentage of oral residue when compared to age-matched healthy individuals. In this case, the patient underwent NMES with medical treatment of D-penicillamine, showed improved swallowing function both oral and pharyngeal phases. Further studies are necessary to investigate the characteristics of dysphagia and the clinical efficacy of NMES in WD.

Table 1. One year follow up of videofluoroscopic swallowing study (videofluoroscopic dysphagia scale) *, improved item in follow-up VFSS.

	Feburary 12, 2016	January 13, 2017
Lip closure	2	2
Bolus formation	3	3
Mastication*	4	0
Apraxia*	3	0
Tongue to palate contact	5	5
Premature bolus loss*	4.5	3
Oral transit time*	3	0
Triggering of pharyngeal swallow*	4.5	0
Valleular residue	4	4
Laryngeal elevation*	9	0
Pyriform sinus residue	0	0
Coating on the pharyngeal wall	0	0
Pharyngeal transit time	0	0
Aspiration	6	6
Total VDS score	48	23

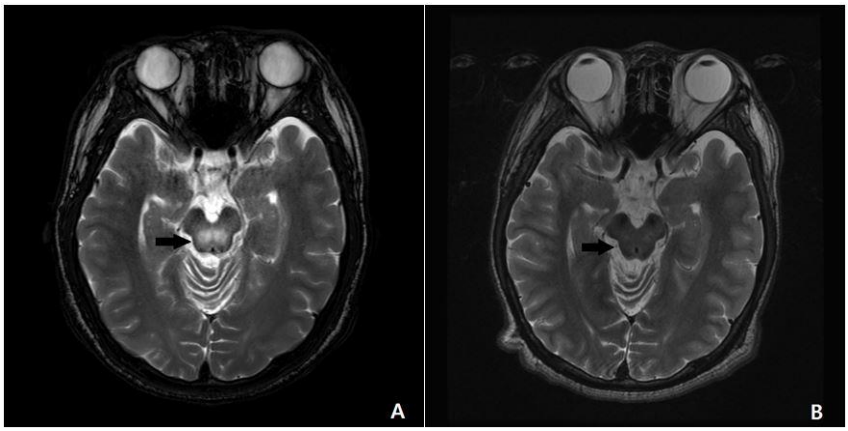

Figure 1. One year follow-up of brain MRI. Initial brain MRI shows high signal intensity (black arrow) in pons and midbrain (A, February 2, 2016). With the treatment of D-penicillamine, 1-year follow-up brain MRI shows decreased signal intensity (black arrow) in the pons and the midbrain (B, January 10, 2017).

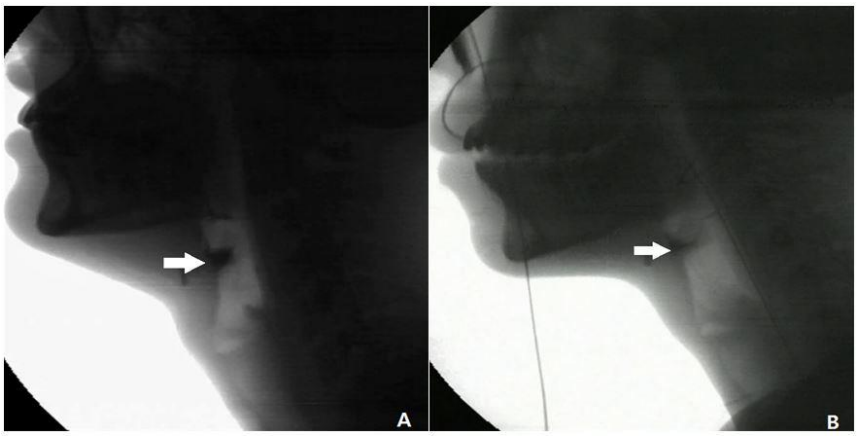

Figure 2. One year follow-up of videofluoroscopic swallowing study (VFSS). The amount of semisolid vallecular residue (B, January 13, 2017) was reduced compared to initial VFSS (A, February 12, 2016). White arrow is the vallecular residue.