Teaching NeuroImages: Alexander disease with features of both frontal and bulbospinal involvement

Tai-Seung Nam, MD, PhD, Kyung-Wook Kang, MD, Seok-Yong Choi, MD, PhD, and Myeong-Kyu Kim, MD, PhD

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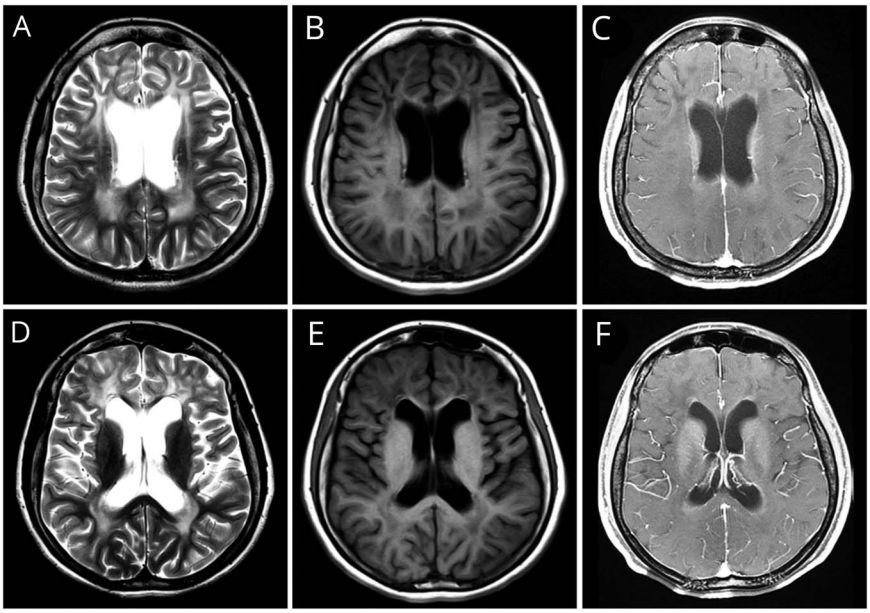
Correspondence
Dr. Kim
mkkim@chonnam.ac.kr

Figure 1 MRI of the supratentorial brain



Axial T2-weighted (A) and T1-weighted (B) images show cerebral white matter abnormalities with frontal predominance, and periventricular rim with T2 hypointensity and T1 hyperintensity. Contrast-enhanced axial T1-weighted image (C) shows symmetrical enhancement of periventricular regions. Axial T2-weighted (D) and T1-weighted (E) images show signal changes in basal ganglia and periventricular white matter, with focal target-shaped lesions around the tip of anterior horns of lateral ventricle. Contrast-enhanced axial T1-weighted image (F) shows contrast-enhancing lesions in basal ganglia. These radiologic findings are suggestive of type I Alexander disease.

A 28-year-old woman, who was considered to have Alexander disease (AxD) at 14 months of age, presented with bulbospinocerebellar symptoms from the age of 22 years. Brain MRI showed frontal white matter abnormality with contrast enhancement (figure 1) suggesting type I AxD,[1] and atrophy in medulla oblongata and cervical cord as well as pial fluid-attenuated inversion recovery hyperintensities in brainstem (figure 2) suggesting type II.[1] *GFAP* sequencing revealed a heterozygous p.Arg88Cys mutation.

AxD is classified into 2 subtypes based on onset age and CNS involvement.[1] This case suggests AxD can be presented as intermediate form with features of both types I and II.[2]

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Author contributions

T.-S. Nam conceived and designed the study. T.-S. Nam and K.-W. Kang enrolled the subject and collected data. T.-S. Nam and M.-K. Kim analyzed and interpreted the clinical and radiologic data. S.-Y. Choi interpreted the genetic data. M.-K. Kim supervised the study. T.-S. Nam and M.-K. Kim revised the manuscript critically.

From the Departments of Neurology (T.-S.N., K.-W.K., M.-K.K.) and Biomedical Sciences (S.-Y.C.), Chonnam National University Medical School, Gwangju, Korea.

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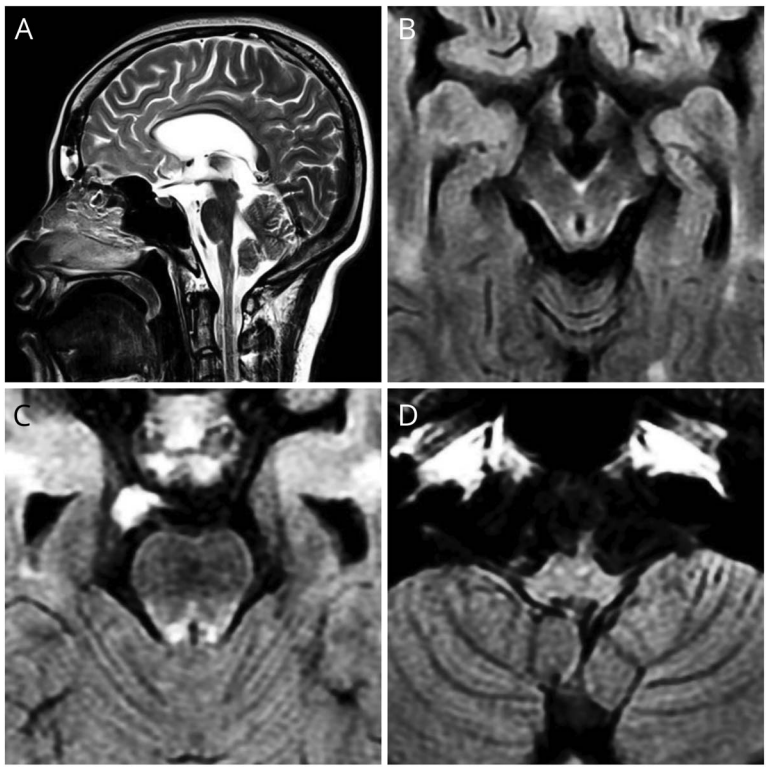
The study protocol was approved by the Institutional Review Board at Chonnam National University Hospital (CNUH-2014-066). The subject consented to the publication of her case.

Figure 2 MRI of the infratentorial brain



Sagittal T2-weighted image (A) shows mild atrophy and hyperintensities in medulla oblongata, cerebellum, and upper cervical spinal cord. Axial fluid-attenuated inversion recovery (FLAIR) images show pial FLAIR signal changes in midbrain (B), pons (C), and medulla oblongata (D). These radiologic findings are suggestive of type II Alexander disease.

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Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

References

1. Prust M, Wang J, Morizono H, et al. GFAP mutations, age at onset, and clinical subtypes in Alexander disease. Neurology 2011;77:1287–1294.
2. Yoshida T, Sasaki M, Yoshida M, et al. Nationwide survey of Alexander disease in Japan and proposed new guidelines for diagnosis. J Neurol 2011;258:1998–2008.