

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

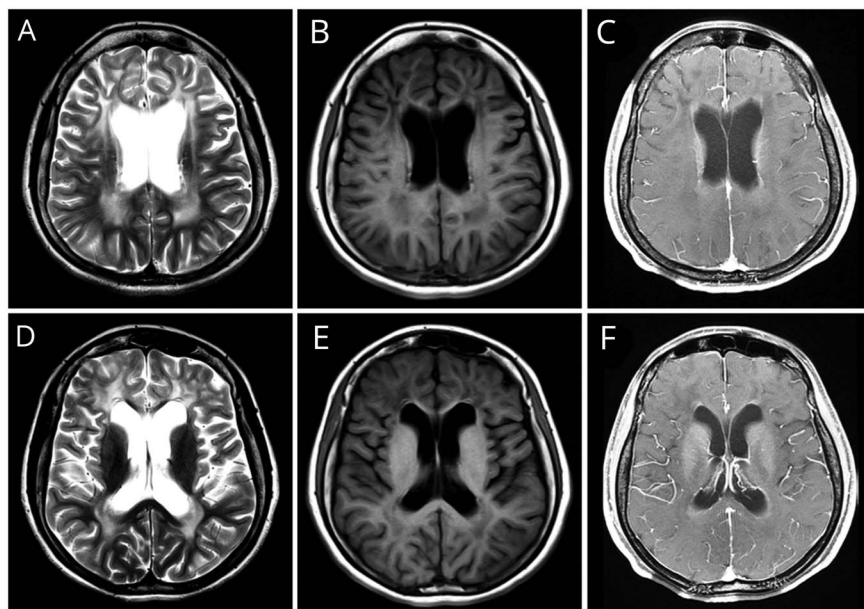
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Figure 1 MRI of the supratentorial brain



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 bulbospino-cerebellar 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

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.

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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.

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