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Florence Renaldo, François Chalard, Stephanie Valence, Lydie Burglen, Diana  
Rodriguez

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### Area Postrema Syndrome as the Initial Presentation of Alexander Disease

**Author(s):**

Florence Renaldo, MD<sup>1</sup>; François Chalard, MD<sup>2</sup>; Stephanie Valence, MD, PhD<sup>1</sup>; Lydie Burglen, MD, PhD<sup>3</sup>; Diana Rodriguez, MD, PhD<sup>4</sup>

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**Corresponding Author:**

Florence Renaldo  
florence.renaldo@aphp.fr

**Affiliation Information for All Authors:** 1. Service de Neuropédiatrie & Centre de Référence Neurogénétique, APHP.SU, Hôpital Trousseau, Paris, France; 2. Department of Pediatric Imaging, APHP.SU, Hôpital Trousseau, Paris, France; 3. Département de génétique médicale, APHP.SU, Hôpital Armand Trousseau; Developmental Brain Disorders Laboratory Imagine Institute, INSERM UMR 1163; Paris, France; 4. Service de Neuropédiatrie & Centre de Référence Neurogénétique, APHP.SU, Hôpital Trousseau; Sorbonne Université, Inserm U1141- FHU I2-D2; Paris, France;

**Contributions:**

Florence Renaldo: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data  
François Chalard: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Analysis or interpretation of data; Additional contributions: F. Chalard analyzed and interpreted the radiologic data.  
Stephanie Valence: Drafting/revision of the manuscript for content, including medical writing for content; Analysis or interpretation of data  
Lydie Burglen: Drafting/revision of the manuscript for content, including medical writing for content; Analysis or interpretation of data; Additional contributions: L. Burglen interpreted the genetic data.  
Diana Rodriguez: Drafting/revision of the manuscript for content, including medical writing for content; Analysis or interpretation of data

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A 16-year-old boy presented with six years evolution of anorexia, failure to thrive, nocturnal vomiting, and dysphagia. He was exhaustively explored. Neurologic examination was normal except hoarse voice. MRI revealed a hyperintensity of the area postrema (figure).

Faced with this chronic and isolated area postrema syndrome we looked for Alexander disease rather than neuromyelitis optica spectrum disorder. GFAP gene sequencing identified a reported, *de novo*, heterozygous variant c.1290C>A;p.Arg430Arg<sup>1</sup>.

Several patients with juvenile onset or type II Alexander disease have been reported with episodic vomiting, anorexia, and MRI lesions which can retrospectively be recognized as an impairment of the area postrema.<sup>2</sup>

#### Appendix 1: Authors

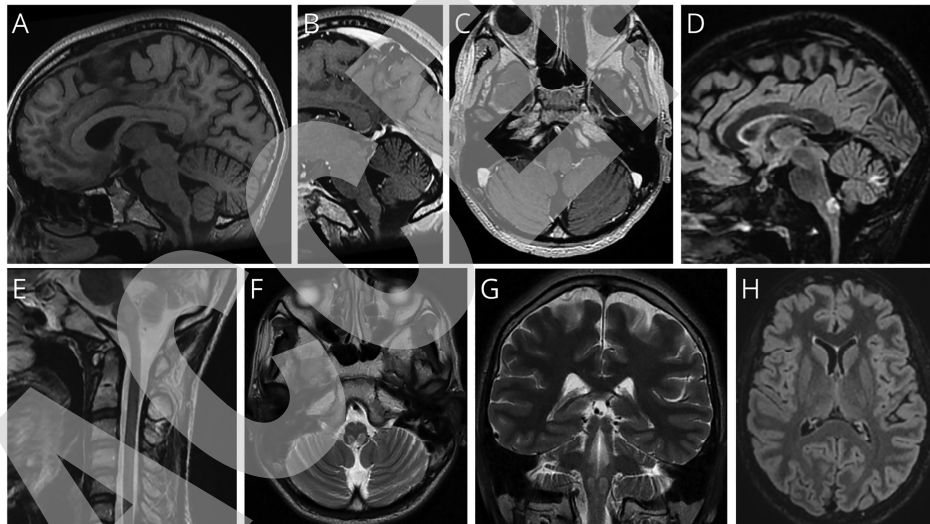
| Name                       | Location  | Contribution  |
|----------------------------|---|---|
| Florence Renaldo, MD       | Service de Neuropédiatrie & Centre de Référence Neurogénétique, APHP.SU, Hôpital Trousseau, Paris, France   | designed the study, collected and analyzed the data, drafted the manuscript |
| François Chalard, MD       | Department of Pediatric Imaging, APHP.SU, Hôpital Trousseau, Paris, France  | analyzed and interpreted the radiologic data                                |
| Stephanie Valence, MD, PhD | Service de Neuropédiatrie & Centre de Référence Neurogénétique, APHP.SU, Hôpital Trousseau, Paris, France   | analyzed the data, critically reviewed and revised the manuscript           |
| Lydie Burglen, MD, PhD     | Département de génétique médicale & Centre de Référence Neurogénétique, APHP.SU, Hôpital Trousseau ; Developmental Brain Disorders Laboratory Imagine Institute, INSERM UMR 1163; Paris, France | interpreted the genetic data.   |
| Diana Rodriguez, MD, PhD   | Sorbonne Université, Service de Neuropédiatrie & Centre de Référence Neurogénétique, APHP.SU, Hôpital Trousseau; Inserm U1141- FHU I2-D2 ; Paris, France  | analyzed the data, critically reviewed and revised the manuscript           |

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Legend to figure:

MRI shows lesion in the area postrema with T1-weighted hypointensity (A), contrast enhancement (B,C) and FLAIR (D) and T2-weighted (E-G) hyperintensity. Apart from a vermian lesion with FLAIR (D) hyperintensity, MRI finds no significant abnormalities of the supratentorial white matter nor basal ganglia (G,H) nor spinal cord (not shown).



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