

Are your MRI contrast agents cost-effective?

Learn more about generic Gadolinium-Based Contrast Agents.



FRESENIUS  
KABI

caring for life

**AJNR**

**MR Imaging Features of Adult-Onset  
Neuronal Intranuclear Inclusion Disease May  
Be Indistinguishable from Fragile X–  
Associated Tremor/Ataxia Syndrome**

I.G. Padilha, R.H. Nunes, F.A. Scortegagna, J.L. Pedroso,  
V.H. Marussi, M.R. Rodrigues Gonçalves, O.G.P. Barsottini  
and A.J. da Rocha

This information is current as  
of April 17, 2024.

*AJNR Am J Neuroradiol* 2018, 39 (9) E100-E101

doi: <https://doi.org/10.3174/ajnr.A5729>

<http://www.ajnr.org/content/39/9/E100>

# MR Imaging Features of Adult-Onset Neuronal Intranuclear Inclusion Disease May Be Indistinguishable from Fragile X–Associated Tremor/Ataxia Syndrome

We have read with great interest the article by Sugiyama et al<sup>1</sup> published in the November 2017 edition of the *American Journal of Neuroradiology*, which described typical brain MR imaging findings in adult-onset neuronal intranuclear inclusion disease (NIID). The authors highlighted the paravermal signal changes on FLAIR sequences as well as the high-intensity signal on DWI along the corticomedullary junction as typical neuroimaging findings in NIID.

According to Sone et al,<sup>2</sup> who described clinical features, MR imaging findings, and pathologic features in a larger series of fragile X-associated tremor/ataxia syndrome (FXTAS) and NIID overlaps, and it is not reliable for distinguishing both disorders based on either clinical presentation, imaging findings, or family history. Moreover, pathologic changes are also similar because FXTAS and NIID usually present with intranuclear eosinophilic inclusions. The study of Sugiyama et al<sup>1</sup> did not evaluate the CGG repeat length of the *FMR1* gene in their patients, which is a crucial step to support their conclusions. When we reviewed our institutional records, 3 recent patients were clinically evaluated and genetic studies confirmed FXTAS. Most interesting, our 3 patients with FXTAS presented with imaging findings (Figs 1 and 2) very similar to the those in patients described by Sugiyama et al, whose diagnoses were NIID.

Research in neuroradiology must concentrate on predicting specific neurologic disorders, identifying either radiophenotypes and/or useful algorithms for clinical practice. Considering that FXTAS seems to be more common than adult-onset NIID, it is reasonable that the algorithm using genetic studies (*FMR1* gene premutation) proposed by Sone et al<sup>2</sup> remains unpredictable because the pathologic changes and the imaging findings reported by Sugiyama et al<sup>1</sup> may occur in both disorders.

Disclosures: Orlando G.P. Barsottini—UNRELATED: Employment: Federal University of Sao Paulo, Brazil, Comments: Professor of Neurology.

## REFERENCES

1. Sugiyama A, Sato N, Kimura Y, et al. MR imaging features of the cerebellum in adult-onset neuronal intranuclear inclusion disease: 8 cases. *AJNR Am J Neuroradiol* 2017;38:2100–04 CrossRef Medline
2. Sone J, Mori K, Inagaki T, et al. Clinicopathological features of adult-onset neuronal intranuclear inclusion disease. *Brain* 2016; 139:3170–86 CrossRef Medline


 I.G. Padilha

 R.H. Nunes


 F.A. Scortegagna

Division of Neuroradiology  
Santa Casa de Sao Paulo School of Medical Sciences  
Sao Paulo, Brazil

Division of Neuroradiology  
Diagnosticos da America SA  
Sao Paulo, Brazil

 J.L. Pedroso

Ataxia Unit, Department of Neurology  
Universidade Federal de São Paulo  
Sao Paulo, Brazil

 V.H. Marussi


Division of Neuroradiology  
Med Imagem, Hospital da Beneficencia Portuguesa de Sao Paulo  
Sao Paulo, Brazil

 M.R. Rodrigues Gonçalves

Department of Neurology  
Universidade de São Paulo  
Sao Paulo, Brazil

 O.G.P. Barsottini

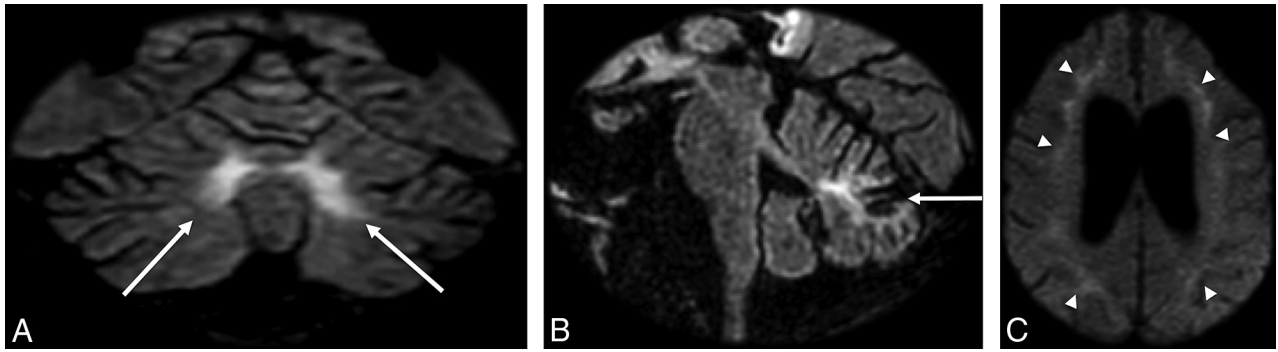
Division of Neuroradiology  
Med Imagem, Hospital da Beneficencia Portuguesa de Sao Paulo  
Sao Paulo, Brazil

 A.J. da Rocha

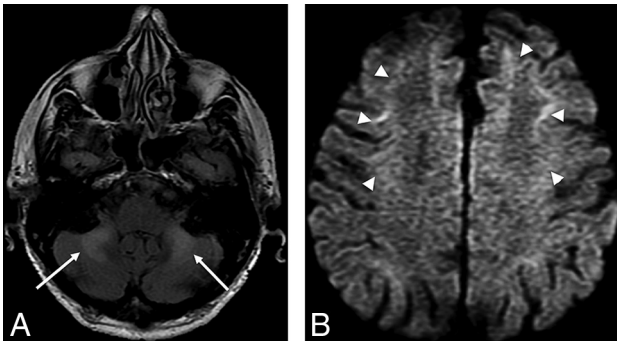
Division of Neuroradiology  
Santa Casa de Sao Paulo School of Medical Sciences  
Sao Paulo, Brazil

Division of Neuroradiology  
Diagnosticos da America SA  
Sao Paulo, Brazil

<http://dx.doi.org/10.3174/ajnr.A5729>



**FIG 1.** Representative case (patient 1). A 49-year-old male patient with a genetically confirmed diagnosis of FXTAS (CGG 115). Coronal (A) and sagittal (B) FLAIR images demonstrate cerebellar atrophy and a high signal intensity in the paravermal area (*arrows*). C, DWI shows abnormal high-intensity signal along the corticomedullary junction (*arrowheads*).



**FIG 2.** Representative case (patient 2). A 69-year-old male patient with slowly progressive ataxia, tremor, and paraparesis. Axial FLAIR image (A) shows bilateral abnormal high signal intensity in the middle cerebellar peduncles (*arrows*). Axial DWI ( $b = 1000 \text{ s/mm}^2$ ) (B) image depicts similar signal changes along the corticomedullary junction (*arrowheads*). Genetic investigation showed 100 CGG repetition (pre-mutation) of the *FMR1* gene, which confirmed FXTAS.