Spino-cerebellar ataxia type 10 (SCA10; OMIM #603516) is an autosomal dominant cerebellar ataxia with variably associated extracerebellar signs. SCA10 is caused by an expanded noncoding pentanucleotide repeat in ATXN10, which normally ranges from 9 to 32 repeats; pathogenic alleles have as many as 4,500 repeats. To date, SCA10 has been found exclusively in the American continents. The clinical presentation of this family is complex and closely resembles “Mexican” SCA10 (with seizures). The later onset of disease in these individuals is likely a reflection of the short expansion length. Absence of cerebellar atrophy in 2 of the individuals is not entirely surprising given their young age.

**History and examination.** The family in this case report lives in a northeastern province of China. The pedigree is noted in the figure, A.

**Proband (IV-2).** This 45-year-old woman first noted gait imbalance at 40 years of age with accompanying dysgraphia, dysarthria, diplopia, short-term memory loss, anxiety, irritability, occasional bouts of insomnia, and recurrent headaches. She does not have a history suggestive of seizures.

On examination, she had horizontal and vertical nystagmus, fragmented ocular pursuit, ocular dysmetria, scanning speech, mild limb and gait ataxia with a widened base, slow speed, and inability to perform tandem walking. She had no Romberg sign, sensory loss, or pyramidal or extrapyramidal signs. Mental and cognitive status were within normal limits. Brain MRI scan was normal. She had no dysphagia, seizures, or other neurologic deficits except mild memory loss.

**Mother of the proband (III-2).** This 72-year-old female began noticing unsteady gait and slurred speech at 60 years of age. She noted no dysphagia, seizures, or other neurologic deficits except mild memory loss.

Her examination showed fragmented ocular pursuit, vertical nystagmus, scanning speech, limb and gait ataxia, and cognitive impairment (MMSE score 25/30, MoCA score 19/30) but no pyramidal, extrapyramidal, or sensory disturbances. Brain MRI was normal. EEG reportedly showed a single moderate-voltage 16- to 20-Hz spike-like wave, a few low-to-moderate voltage 4- to 7-Hz theta waves, and 2.5- to 3.5-Hz delta waves in bilateral anterior and middle temporal regions during wakeful state.

**SCA10 expansion testing.** Following informed consent and in accordance with institutional review panels at the China-Japan Friendship Hospital, genomic DNA was extracted from peripheral blood leukocytes and tested for the SCA10 expansion by repeat-primed PCR (RP-PCR) following negative results for SCA1, 2, 3, 6, 7, 8, 12, and 17 and dentatorubral-pallidolysian atrophy. Further molecular analysis was performed on anonymized samples at the University of Florida after institutional review board approval. The proband (IV-2) tested positive for an SCA10 expansion by RP-PCR, as did her mother (III-2), maternal aunt (III-4), and asymptomatic sister (IV-3). The SCA10 expansion was sized by Southern blot and found to be in the pathogenic range (figure, C). Haplotype analysis showed the “G-expansion-G-G” haplotype shared by other SCA10-positive individuals (figure, D).

**Discussion.** We report SCA10 in a family outside the American continents. The clinical presentation of this family is complex and closely resembles “Mexican” SCA10 (with seizures). The later onset of disease in these individuals is likely a reflection of the short expansion length. Presence of cerebellar atrophy in 2 of the individuals is not entirely surprising given their young age.

---

**References**


2. Kang Wang, MD* Karen N. McFarland, PhD*

3. Desmond Zeng, BS

4. Jilin Liu, MS, MD

5. Karen N. McFarland, PhD*

6. Kang Wang, MD*
late disease onset and comparatively short expansion allele.

The presence of SCA10 in a Chinese Han family suggests that the original SCA10 mutation may have occurred before the divergence of Proto-Amerinds from ancestral Asians. However, as the shared SCA10 haplotype is relatively common, we cannot rule out alternative hypotheses, including (1) independent expansion events on a common haplotype but in separate geographic locations by chance, or...
backflow of human migration from Beringia carrying SCA10 expansions into Asia. While SCA10 may be a rare cause of autosomal dominant cerebellar ataxia in China, the observation of SCA10 in this family suggests that SCA10 should be included in the differential diagnosis of ataxia in patients with Asian origins.

*These authors contributed equally to the manuscript.

From the Department of Neurology (K.W., Y.H., M.J., W.G.), China-Japan Friendship Hospital, Chaoyang, Beijing, China; and Department of Neurology and McKnight Brain Institute (K.N.M., J.L., D.Z., I.L., G.X., T.A.) and Department of Anthropology and Genetics Institute (C.J.M.), University of Florida, Gainesville.


Acknowledgment: The authors thank this family for their cooperation and participation in this study.

Study funding: This work was supported by NIH R01 NS083564 to T.A. and by a grant from the Ministry of Health of China to W.G. The funders of this study had no role in study design or results interpretation.

Disclosure: Dr. Ashizawa receives patent royalty from Baylor College of Medicine for the SCA10 diagnostic assay. Dr. Ashizawa also receives a book chapter honorarium from Elsevier. All remaining authors report no disclosures. Go to Neurology.org/ng for full disclosure forms.

This is an open access article distributed under the terms of the Creative Commons Attribution-NonCommercial-NoDerivatives License 4.0 (CC BY-NC-ND), which permits downloading and sharing the work provided it is properly cited. The work cannot be changed in any way or used commercially.

Received June 19, 2015. Accepted in final form September 1, 2015.

Correspondence to Dr. Ashizawa: tetsuo.ashizawa@neurology.ufl.edu

Spinocerebellar ataxia type 10 in Chinese Han

*Neurol Genet* 2015;1;
DOI 10.1212/NXG.0000000000000026

This information is current as of October 8, 2015

**Updated Information & Services**

including high resolution figures, can be found at:
[http://ng.neurology.org/content/1/3/e26.full.html](http://ng.neurology.org/content/1/3/e26.full.html)

**References**

This article cites 7 articles, 0 of which you can access for free at:
[http://ng.neurology.org/content/1/3/e26.full.html##ref-list-1](http://ng.neurology.org/content/1/3/e26.full.html##ref-list-1)

**Subspecialty Collections**

This article, along with others on similar topics, appears in the following collection(s):

- **All Epilepsy/Seizures**
  [http://ng.neurology.org//cgi/collection/all_epilepsy_seizures](http://ng.neurology.org//cgi/collection/all_epilepsy_seizures)
- **Cerebellum**
  [http://ng.neurology.org//cgi/collection/cerebellum](http://ng.neurology.org//cgi/collection/cerebellum)
- **Spinocerebellar ataxia**
  [http://ng.neurology.org//cgi/collection/spinocerebellar_ataxia](http://ng.neurology.org//cgi/collection/spinocerebellar_ataxia)

**Permissions & Licensing**

Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
[http://ng.neurology.org/misc/about.xhtml#permissions](http://ng.neurology.org/misc/about.xhtml#permissions)

**Reprints**

Information about ordering reprints can be found online:
[http://ng.neurology.org/misc/addir.xhtml#reprintsus](http://ng.neurology.org/misc/addir.xhtml#reprintsus)

*Neurol Genet* is an official journal of the American Academy of Neurology. Published since April 2015, it is an open-access, online-only, continuous publication journal. Copyright © 2015 American Academy of Neurology. All rights reserved. Online ISSN: 2376-7839.