Case Study

Spinocerebellar Ataxia Type 1 in a Filipino Boy

Joanna Lissa F. Payuran-Gatchalian [1], Maria Antonia Aurora Moral-Valencia [1]

Author's Affiliation:
1- Section of Child Neurology, Department of Neuroscience and Behavioral Medicine & Department of Pediatrics University of Santo Tomas Hospital, Manila, Philippines

Correspondence:
Joanna Lissa F. Payuran-Gatchalian, Email: joannapayurangatchalian@gmail.com

Received on: 29-Sep-2025 Accepted for Publication: 12-Mar-2026

INTRODUCTION

Spinocerebellar ataxias (SCA) are relatively rare with an incidence of 1-5 per 100,000. [(1)] They are dominantly inherited, progressive, neurodegenerative, and heterogenous group of diseases that mainly affects the cerebellum. [(2)] The disease is characterized by premature cerebellar neuronal loss, with some types involving additional structures such as the optic nerve, basal ganglia, brainstem, and spinal cord. The age of onset varies significantly depending on the type and phenotype with an average onset at the 4th decade of life. [(3)] They are also known as Autosomal Dominant Cerebellar Ataxias (ADCAs) and can be classified according to the constellation of clinical signs using the Harding Classification System. [(4)] Currently, there is no available local data on the incidence of the different types of SCA in the Philippines. There are a few published reports on Filipino patients including SCA type 13 in Filipino kindred in 41 patients [(5)], SCA type 7 in a 22-year-old male [(7)], and SCA type 2 in Filipino kindred involving 10 patients. [(8)] This case report aims to add to the scarcity in literature regarding SCA in the Philippines.

CASE REPORT

A 12-year-old Filipino boy from San Mateo, Rizal, born of nonconsanguineous marriage, typically developing and previously healthy, was seen presenting with chronic (1 year) progressive ataxia eventually with dysarthria, scanning speech, dysmetria, dysdiadochokinesia, titubation, wide based gait, and impaired memory. This was also accompanied by incoordination of both hands causing difficulty in performing activities of daily living including writing, eating, and bathing. His past medical history was otherwise unremarkable. There was no history of exposure to alcohol, drugs, or toxins. His father, paternal grandfather, two paternal uncles, two paternal aunts, and two paternal female cousins also showed similar symptoms. Upon physical examination, he had normal vital signs and general examination. Mental status exam showed scanning speech and problems with delayed recall and serial subtraction but had good insight and judgment. Cranial nerve examination revealed tongue fasciculations. Motor examination showed normal bulk, tone, and power. Deep tendon reflexes were normal on all extremities. Sensory examination was intact for all modalities. He had cerebellar signs including

bilateral dysmetria, bilateral dysdiadochokinesia and displayed a wide based gait with difficulty performing tandem walk. Saccades and smooth pursuit were intact.

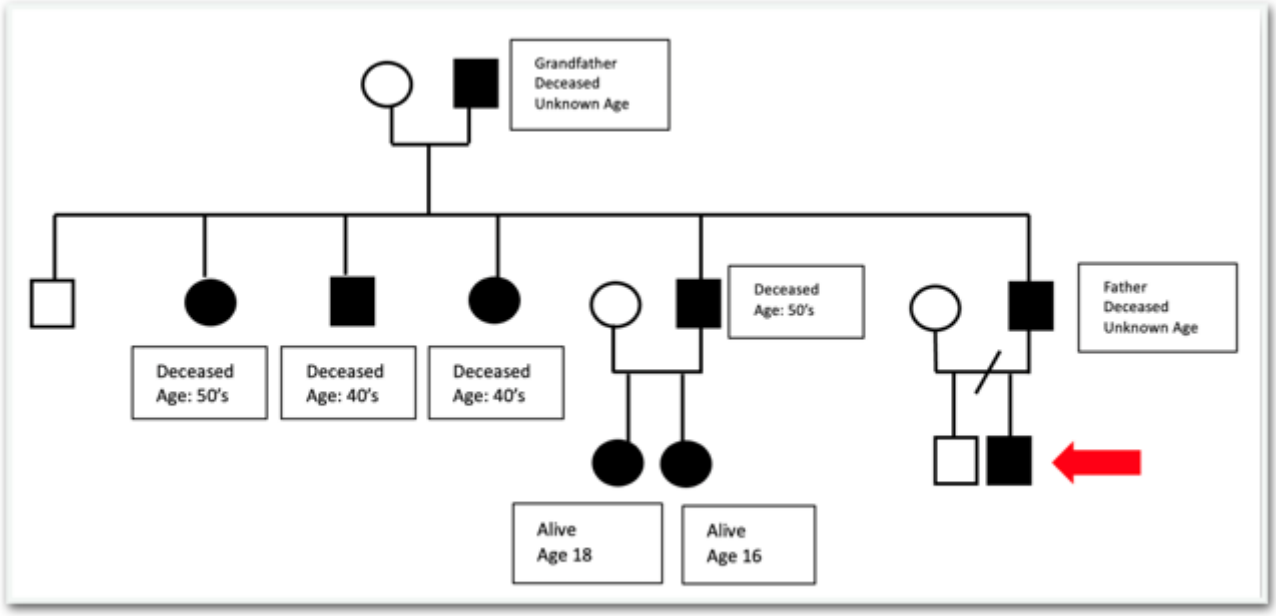

Figure 1: Genogram showing three generations and the affected family members

Investigation with cranial MRI with contrast showed cerebellar atrophy while whole exome sequencing revealed a heterozygous pathogenic expanded allele (~71 CAG) in the full penetrance range identified in the ATXN1 gene consistent with a diagnosis of Spinocerebellar ataxia type 1.

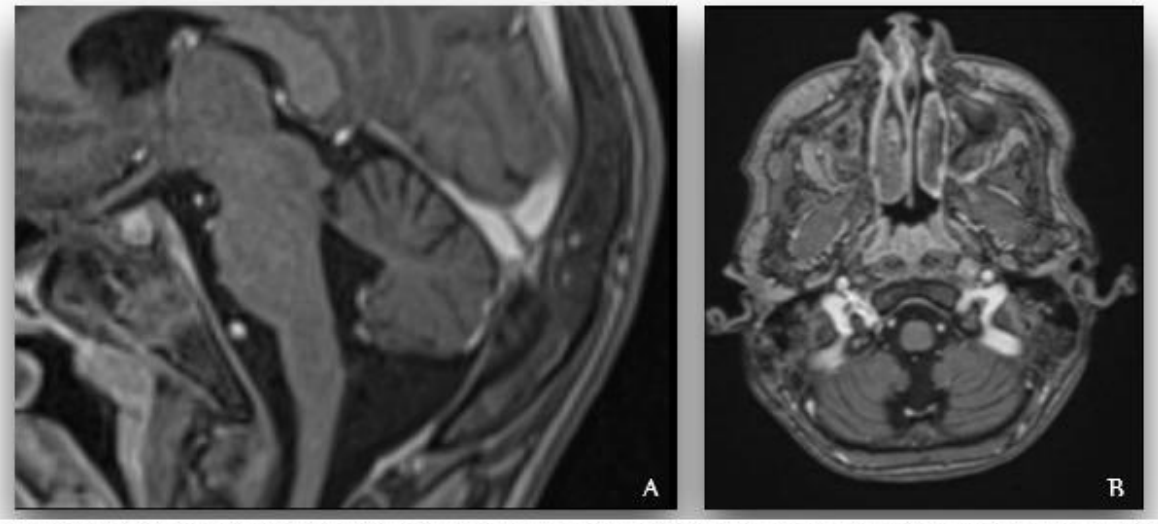

Figure 2: (A) cranial MRI in contrast enhanced T1W in the sagittal view showing cerebellar atrophy with mild hypoplasia of the cerebellar vermis (B) cranial MRI contrast enhanced T1W in the axial view showing slight prominence with a keyhole appearance of the fourth ventricle suggestive of cerebellar atrophy

The patient was referred to a geneticist and genetic counseling was given to the patient and his family. Supportive management was also provided including referral to nutrition for possible initiation of enteral nutrition, referral to rehabilitation medicine for physical and occupational therapy. Since this is a life limiting illness, referral to family medicine was also done for family counseling and palliative care. Monthly online follow up was also scheduled to check on the patient's condition and progression of symptoms. In addition, a hotline is

made available 24 hours a day, 7 days a week for the family for any inquiry or concern they might have in the future.

DISCUSSION

Spinocerebellar ataxia type 1 (SCA 1) is characterized by progressive cerebellar ataxia, dysarthria, and eventual deterioration of bulbar function. Age of onset is typically in the 3rd or 4th decade although with reported childhood-onset and late-adult onset. Those with onset of later than 60 years may manifest a pure cerebellar phenotype with juvenile onset SCA 1 presenting with a more rapid progression and more severe disease, and they usually die before 16 years old.[3] Phenotypic manifestations are not specific, and no formal diagnostic criteria exist but SCA1 should be suspected in individuals with the following: progressive cerebellar ataxia, dysarthria, eventual deterioration of bulbar functions and a positive family history consistent with autosomal dominant inheritance.[6]

Other reports on SCA 1 have estimated its incidence to be at 1-2 per 100,000 population.[6] In Europe, two Polish studies reported the highest relative frequency of SCA1 among European countries, with percentages of 42% and 68%, respectively[10,11]. In one study in Asia involving China, India, Japan, Singapore, South Korea, Thailand, and Taiwan, reported median frequencies of SCA1 ranged from 5.4% to 32.4% with some geographic variation and possibly owing to founder effect. [9]

Genetically, SCA fall into 2 major groups: polyglutamine SCAs (SCA 1, SCA 2, SCA 3/Machado-Joseph Disease, SCA 6, SCA 7, SCA 17) caused by translated CAG repeat expansion mutations that encode stretches of pure glutamine in the respective disease proteins, to which our patient belongs to. The other major group are those caused by conventional, non-repeat mutations, nonsense mutations, insertion, or deletions. Clinically important is that polyglutamine SCAs demonstrate the clinical anticipation phenomenon wherein the diseases causing the repeats tend to lengthen upon transmission and the longer are the repeats, the greater is the severity of the disease and the earlier the onset. Hence, the disease symptoms tend to worsen from generation to generation in a family. Clinical anticipation occurs in the most common SCAs including SCA 1, SCA 2, and SCA 7.[1] Evident in our patient's genogram (Figure 1) is the decreasing age of onset of this disease in the succeeding generations. Recognition of anticipation in polyglutamine disorders like SCA1 is critical for pediatricians managing affected families. Genetic counseling must be sensitive to familial dynamics, inheritance risks, and future planning. Ideally, for our patient, further investigation and genetic testing can be done in all living symptomatic individuals in the patient's family. However, no consent was given.

Management following diagnosis of SCA1 involves the following recommended evaluation to establish the extent of disease: detailed neurologic examination, video esophagogram for those presenting with dysphagia, oral feeding assessment by a therapist, formal ophthalmologic examination, cognitive assessment, assessment for

pain secondary to muscle cramps, genetic counseling, as well as assessment for presence of community resources, social support system, and possible referral to a home nursing.[6] No treatment is available yet to delay or halt the progression of SCA1, and management is largely individualized and catered each patient.

To our knowledge, this is the first reported pediatric case of SCA1 in the Philippines, highlighting the need for regional data to better understand its epidemiology and guide future screening efforts.

In conclusion, while disease-modifying treatments are lacking, recognizing and understanding this disease is imperative for pediatricians because pediatricians play a central role in the multidisciplinary care to support quality of life, functional independence, and emotional well-being.

ETHICS APPROVAL

This case report was originally written, and informed consent and assent obtained in compliance with the National Ethical Guidelines for Research Involving Human Participants NEGRIHP 2022 and has been submitted to and approved by the University of Santo Tomas Research Ethics Committee as required for presentation and publication of this case report.

REFERENCES

1. Klockgether, T., Mariotti, C. & Paulson, H.L. Spinocerebellar ataxia. Nat Rev Dis Primers 5, 24 (2019). https://doi.org/10.1038/s41572-019-0074-3
2. Bhandari J, Thada PK, Samanta D. Spinocerebellar Ataxia. [Updated 2023 Sep 15]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK557816/#
3. Swaiman, K. F., Ashwal, S., Ferriero, D. M., Schor, N. F., Finkel, R. S., Gropman, A. L., Pearl, P. L., & Shevell, M. I. (2017). Swaiman's Pediatric Neurology: Principles and Practice: Sixth Edition. Elsevier Inc.
4. Teive HA, Munhoz RP, Arruda WO, Lopes-Cendes I, Raskin S, Werneck LC, Ashizawa T. Spinocerebellar ataxias: genotype-phenotype correlations in 104 Brazilian families. Clinics (Sao Paulo). 2012;67(5):443-9. doi: 10.6061/clinics/2012(05)07. PMID: 22666787; PMCID: PMC3351252.
5. Subramony SH, Advincula J, Perlman S, Rosales RL, Lee LV, Ashizawa T, Waters MF. Comprehensive phenotype of the p.Arg420his allelic form of spinocerebellar ataxia type 13. Cerebellum. 2013 Dec;12(6):932-6. doi: 10.1007/s12311-013-0507-6. PMID: 23912307; PMCID: PMC3824261.
6. Opal P, Ashizawa T. Spinocerebellar Ataxia Type 1. 1998 Oct 1 [Updated 2023 Feb 2]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024.
7. Jamora, R., & Ocampo, C. (2006). A rare case of cerebellar ataxia with macular degeneration. Philippine Journal of Neurology, 10(2), 67.
8. Matibag, J. L. R., Uematsu, E., & Diesta, C. C. (2020). A benchmark study on the correlation of CAG trinucleotide repeat length with SARA score, age of onset, and disease duration of genetically-positive spinocerebellar ataxia 2 Filipino family. International Journal of Neurodegenerative Disorders, 3, Article 015. https://doi.org/10.23937/2643-4539/1710015Amanote Research
9. Teije van Prooije, Norlinah Mohamed Ibrahim, Shahrul Azmin, Bart van de Warrenburg, Spinocerebellar ataxias in Asia: Prevalence, phenotypes and management, Parkinsonism & Related Disorders, Volume 92, 2021, Pages 112-118, ISSN 1353-8020, https://doi.org/10.1016/j.parkreldis.2021.10.023. (https://www.sciencedirect.com/science/article/pii/S1353802021003862)

10. Sułek-Piątkowska A, Zdzienicka E, Rakowicz M, et al. The occurrence of spinocerebellar ataxias caused by dynamic mutations in Polish patients. Neurol Neurochir Pol. 2010;44(3):238–45. https://doi.org/10.1016/S0028-3843(14)60037-2.

11. Krysa W, Sulek A, Rakowicz M, Szirkowiec W, Zaremba J. High relative frequency of SCA1 in Poland reflecting a potential founder effect. Neurol Sci. 2016;37(8):1319–25. https://doi.org/10.1007/s10072-016-2594-x.