

Sleep Difficulties in Patients With Spinocerebellar Ataxia

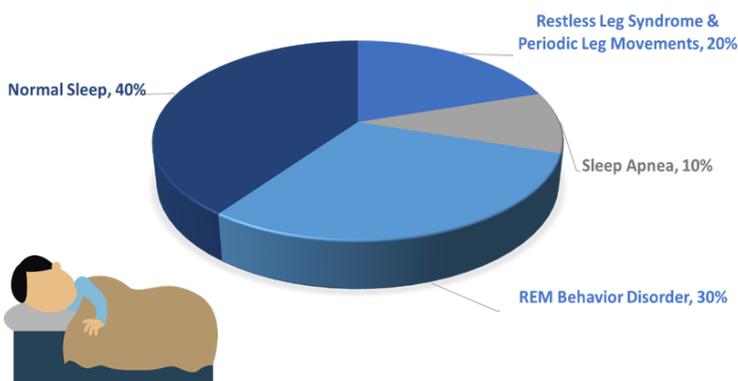
How does SCA have an impact on sleep?

The cerebellum is directly involved in position, movement, and muscle tone during rest and sleep, which can explain the high prevalence of sleep dysfunctions in the Spinocerebellar Ataxias (SCA).¹ More than 50% of individuals with SCA report sleeping difficulties, which includes conditions like REM sleep behavior disorder, restless leg syndrome, atypical periodic limb movement in sleep and sleep apnea.²

Individuals with reduced quantity and quality of sleep experience impairments in cognitive function, mood regulation and body coordination.^{3,4,5} Thus, sleep deprivation can exacerbate symptoms and decrease the effectiveness of rehabilitation therapies. Some research suggests links between specific sleep disorders and subtypes of SCA, but more study is needed to verify these associations.^{6,7,8}

Bursts of electrical activity, known as *sleep spindles*, are believed to mediate many sleep-related functions, including memory consolidation, cortical development, arousal thresholds during sleep and cellular plasticity.^{8,9} Spindle activity comprises about 60% of healthy sleep; however, individuals with SCA may experience little to no spindle activity.⁸ Sleep problems may precede the onset of SCA symptoms and diagnosis of cerebellar disease by up to 10 years.¹⁰ For these reasons, sleep deprivation is gaining attention among medical professionals working to improve the quality of life for individuals with ataxia syndromes.

SELF-REPORTED FREQUENCY OF SLEEP DISORDERS IN PATIENTS WITH SCA^{2,6,7,11,12}



Commonly reported types of sleep disorders in SCA

REM Behavior Disorder (RBD): rapid eye movement (REM) sleep behavior disorder (RBD) is a condition in which a person may physically act out the content of their dreams.¹³ It is classified as a parasomnia, i.e., an abnormal behavior superimposed on and disrupting sleep. Other examples of parasomnia include nightmares or terrors and sleep walking.

Treatment: Patients with this condition and their bedpartners should attend counseling for appropriate steps to minimize injury, such as removing harmful objects and placing protective barriers adjacent to the bed, like a mattress or foam padding.^{13,14} Talk with your physician or a sleep specialist to determine the best lifestyle changes and medications for your situation.

Restless Leg Syndrome (RLS): uncomfortable sensations in the legs with an irresistible urge to move. Symptoms often worsen when a person is inactive. Periodic limb movements are also common.¹⁵

Treatment: Moving affected limbs may provide temporary relief. Decreasing alcohol and tobacco consumption, maintaining a regular sleep pattern and moderate exercise program, massaging legs, taking warm baths, and using heat or ice packs may reduce symptoms. Talk with your physician about whether iron supplements, anti-seizure medications, or dopaminergic agents would be appropriate.^{15,16}

Periodic Limb Movements (PLMS): involuntary leg or arm twitching.¹⁷ A sleep study confirmed that some individuals with SCA experience as many as 59-98 PLMS per night, whereas 4 to 5 per night are considered typical.^{17, 18}

Treatment: Ask your bed partner if you experience PLMS in your sleep, as a partner often has a keener awareness of the frequency and duration.

PLMS is not typically treated unless a person is also diagnosed with RLS, for which treatment is often the same. Walking or stretching may temporarily relieve symptoms.^{16,17} Talk with your physician or request to speak with a sleep professional for more information on management of PLMS.

Sleep Apnea: characterized by snoring or gasping during sleep, reduced or absent breathing, and sleepiness caused by an obstructed airway or a lack of signals from the brain to facilitate breathing.¹⁶

Treatment: Lifestyle changes include heart-healthy eating, physical activity, aiming for a healthy weight, and maintaining a healthy sleep routine. Therapy is also available for mouth and facial muscles. Devices can also be used to improve night-time breathing, such as a mouth piece or implant, or a continuous positive air pressure (CPAP) machine.^{14,16}

What should I do if I have difficulty sleeping?

If you have concerns about your quality of sleep, talk with your physician or other qualified professional to determine the best course of management or treatment. Sleep is a daily function that is essential for human health and well-being, talk with your physician about seeing a sleep specialist if you have concerns, such as a sleep neurologist. Treatments may vary depending on the proper diagnosis of a sleep condition through the management of medications, sleep routines and behaviors, and/or machines (e.g. CPAP machines for sleep apnea).^{16,19}

The physiological mechanisms of sleep disturbances among individuals with cerebellar ataxias are largely unknown. Therefore, a one-size fits all approach is not available. Treatment plans are individualized and are developed through a consultation with the medical team—with the goal of an optimal pharmacological and physiotherapy regimen for the specific problems¹⁹

How does the National Ataxia Foundation help?

The National Ataxia Foundation (NAF) is committed to providing information and education about Ataxia, support groups for those affected by Ataxia, and promoting and funding research to find the cause for the various forms of Ataxia, better treatments, and, hopefully someday, a cure. NAF has been at the forefront funding promising worldwide research to find answers.

As Ataxia research moves into the clinical phase, pharmaceutical companies will begin recruiting participants for clinical trials. Individuals with Ataxia or who are at-risk for Ataxia are encouraged to enroll in the CoRDS Ataxia Patient Registry. To access the Registry, go to NAF's website www.ataxia.org and click on the "Enroll in the Patient Registry" tab and follow the directions on the CoRDS website.

NAF provides accurate information for you, your family, and your physician about Ataxia. Please visit the NAF website at www.ataxia.org for additional information, including a listing of ataxia support groups, physicians who treat Ataxia, social networks, and more. For questions contact the NAF directly at 763/553-0020 or naf@ataxia.org.

Jessica J. Rilee, OTD, Chiadi U. Onyike, MD, Liana S. Rosenthal, MD, and Cherie L. Marvel, PhD

The Johns Hopkins University School of Medicine in
Collaboration with the National Ataxia Foundation

References:

- [1] DelRosso, L. M., & Hoque, R. (2014). The cerebellum and sleep. *Neurologic Clinics*, 32, 893-900. doi:10.1016/j.ncl.2014.07.003
- [2] Pedroso, J. L., Braga-Neto, P., Felicio, A., Aquino, C., Fernandes do Prado, L., Fernandes do Prado, G., & Barsottini, O. (2011). Sleep disorders in cerebellar ataxias. *Arquivos de Neuro-Psiquiatria*, 69(2-A), 253-257.
- [3] Bobic, T. T., Secic, A., Zavoreo, I., Matijevic, V., Filipovic, B., Kolak, Z., . . . Sajkovic, D. (2016). The impact of sleep deprivation on the brain. *Acta Clinica Croatica*, 55(3), 469-473. doi:10.20471/acc.2016.55.03.17
- [4] Boonstra, T. W., Stins, J. F., Daffertshofer, A., & Beek, P. J. (2007). Effects of sleep deprivation on neural functioning: an integrative review. *Cellular and Molecular Life Sciences*, 64(8), 934-946. doi:10.1007/s00018-0076457-8
- [5] Kempler, L., & Richmond, J. (2012). Effect of sleep on gross motor memory. *Memory*, 20(8), 907-914. doi:10.1080/09658211.2012.711837
- [6] Abele, M., Burk, K., Laccone, F., Dichgans, J., & Klockgether, T. (2001). Restless leg syndrome in spinocerebellar ataxia types 1, 2, and 3. *Journal of Neurology*(248), 311-314.
- [7] Schols, L., Haan, J., Riess, O., Amoiridis, G., & Przuntek, H. (1998). Sleep disturbance in spinocerebellar ataxias: Is the SCA3 mutation a cause of restless legs syndrome? *Neurology*, 1603-1607.
- [8] Seshagiri, D. V., Botta, R., Sasidharan, A., Pal, P. K., Jain, S., Yadav, R., & Kutty, B. M. (2018). Assessment of sleep spindle density among genetically positive spinocerebellar ataxias types 1, 2, and 3 patients. *Annals of Neuroscience*, 25, 106-111. doi:10.1159/000484516
- [9] Andrillon, T., Nir, Y., Staba, R., Ferrarelli, F., Cirelli, C., Tononi, G., & Fried, I. (2011). Sleep spindles in humans: insights from intracranial EEG and unit recordings. *Journal of Neuroscience*, 17821-17834. doi:10.1523/JNEUROSCI.2604-11.2011
- [10] Perlman, S. L. (2016). *Evaluation and Management of Ataxic Disorders: An Overview for Physicians*. Minneapolis, MN: National Ataxia Foundation.
- [11] Brusse, E., Brusse-Keizer, M. G., Duivenvoorden, H. J., & Swieten, J. v. (2011). Fatigue in spinocerebellar ataxia: Patient self-assessment of an early and disabling symptom. *Neurology*, 76, 953-959.
- [12] Sonni, A., Kurdziel, L. B., Baran, B., & Spencer, R. (2014). The effects of sleep dysfunction on cognition, affect, and quality of life in individuals with cerebellar ataxia. *Journal of Clinical Sleep Medicine*, 10(5), 535-543. doi:10.5664/jcsm.3706
- [13] National Sleep Foundation. (2018, November 2). *REM Sleep Behavior Disorder*. Retrieved from National Sleep Foundation, Sleep Disorders: <https://www.sleepfoundation.org/sleep-disorders-problems/rem-behavior-disorder>

[14] National Heart, Lung, and Blood Institute. (2018, November 2). *Sleep Apnea*. Retrieved from Fact Sheet: <https://www.nhlbi.nih.gov/health-topics/sleep-apnea>

[15] National Institute of Neurological Disorders and Stroke. (2018, November 2). *Restless Legs Syndrome*. Retrieved from Fact Sheet: <https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Restless-Legs-Syndrome-Fact-Sheet>

[16] American Occupational Therapy Association. (2014). Occupational Therapy Practice Framework: Domain & Process (3rd Edition). *American Journal of Occupational Therapy, 68*, S1-S48, doi:10.5014/ajot.2014.682006.

[17] [National Sleep Foundation. \(2018, November 2\). *Periodic Limb Movements in Sleep*. Retrieved from National Sleep Foundation, Sleep Disorders: https://www.sleepfoundation.org/sleep-disorders-problems/sleep-related-movement-disorders/periodic-limb-movement-disorder](https://www.sleepfoundation.org/sleep-disorders-problems/sleep-related-movement-disorders/periodic-limb-movement-disorder)

[18] Boesch, S. M., Frauscher, B., Brandauer, E., Wenning, G. K., Hogl, B., & Poewe, W. (2006). Disturbance of rapid eye movement sleep in spinocerebellar ataxia type 2. *Movement Disorders, 21*(10), 1751-1754.

[19] Driver-Dunckley, E. D., & Adler, C. H. (2012). Movement disorders and sleep. *Neurologic Clinics, 1345-1358*. doi:10.1016/j.ncl.2012.08.019