

Sleep, Respiration and Nocturnal Paroxysmal Events in Joubert Syndrome: A Case Report

Rosa Peraita-Adrados 

Sleep and Epilepsy Unit- Clinical Neurophysiology Service, University General Hospital and Research Institute Gregorio Marañón, University Complutense of Madrid (UCH), Madrid, Spain

Correspondence: Rosa Peraita-Adrados, Email rosa-peraita@telefonica.net

Introduction: Joubert syndrome is a rare disorder, characterized by a complex midbrain malformation caused by defects in the structure and/or function of the primary cilium.

Case Report: A 15-year-old boy with mild intellectual disability, hypotonia, mild ataxia, and abnormal eye movements diagnosed as having Joubert Syndrome since childhood, was referred to the Sleep Unit because spells of apnea while sleeping. He did not complain of snoring or daytime somnolence. The macro and microstructure of sleep and the comorbidities such respiratory abnormalities, periodic legs movements (PLM) and paroxysmal motor arousals (PA) and minimal motor events (MME) are described for the first time in Joubert syndrome.

Results: EEG was normal. Video-polysomnography revealed a nocturnal disturbed sleep and periods of hyperpnea accompanied by body movements and followed by a periodic breathing lasting several minutes with no oxygen desaturation. The arousals provoked by apneas triggered paroxysmal motor events with dystonic movements in the hand and right foot accompanied by a spontaneous Babinski. Brain MRI showed the typical “molar tooth sign”.

Conclusion: Joubert syndrome is a heterogeneous disease. Epileptic seizures have been reported in some cases. Video-PSG is mandatory for the identification of nocturnal breathing abnormalities and sleep-related motor paroxysmal episodes.

Keywords: Joubert syndrome, molar tooth sign, hyperpnea and central periodic breathing, nocturnal paroxysmal motor events, sleep fragmentation and arousals

Plain Language Summary

Joubert syndrome is a clinically heterogeneous disease. Significantly, variability in phenotype and genetics has been described.

Joubert syndrome is under-diagnosed and should always be considered in children with non-progressive ataxia, intellectual disability and episodes of hyperpnea and apnea.

The presence of molar tooth sign its present in 85% of the cases of Joubert syndrome.

The epileptic seizures have been reported with lack of details regarding the electro-clinical semiology and without a clear-cut circadian presentation schedule.

The possible physiological mechanism triggering the sleep paroxysmal motor events has been described such as respiratory arousals and motor arousals (PLM) as the usual triggers in nocturnal frontal epileptic seizures and in parasomnias.

A Video-PSG is mandatory for the identification of breathing abnormalities during sleep as well as comorbidities with PLM, parasomnias and related motor seizures during sleep.

Introduction

Joubert Syndrome (JS) first described in 1969,¹ is a rare, autosomal recessive disorder, clinically heterogeneous that combine neurological signs: generally poorly controlled movements and mild to moderate intellectual disability, with variable multiorgan involvement, mainly of retina, kidneys, liver and skeleton. Brain MRI is characterized by a complex midbrain-hindbrain malformation with the absence or underdevelopment of the cerebellar vermis and a broad spectrum of other phenotypic findings caused by defects in the structure and/or function of the primary cilium.² There is a hallmark



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Nic Butkov, Teofilo L. Lee-Chiong



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 - The Rise of Digital Reading Sleep And Respiration
 - Advantages of eBooks Over Traditional Books
2. Identifying Sleep And Respiration
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Sleep And Respiration
 - User-Friendly Interface
4. Exploring eBook Recommendations from Sleep And Respiration
 - Personalized Recommendations
 - Sleep And Respiration User Reviews and Ratings
 - Sleep And Respiration and Bestseller Lists
5. Accessing Sleep And Respiration Free and Paid eBooks
 - Sleep And Respiration Public Domain eBooks
 - Sleep And Respiration eBook Subscription Services
 - Sleep And Respiration Budget-Friendly Options
6. Navigating Sleep And Respiration eBook Formats

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- Sleep And Respiration Compatibility with Devices
- Sleep And Respiration Enhanced eBook Features
- 7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Sleep And Respiration
 - Highlighting and Note-Taking Sleep And Respiration
 - Interactive Elements Sleep And Respiration
- 8. Staying Engaged with Sleep And Respiration
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Sleep And Respiration
- 9. Balancing eBooks and Physical Books Sleep And Respiration
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Sleep And Respiration
- 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
- 11. Cultivating a Reading Routine Sleep And Respiration
 - Setting Reading Goals Sleep And Respiration
 - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Sleep And Respiration
 - Fact-Checking eBook Content of Sleep And Respiration
 - Distinguishing Credible Sources
- 13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development
 - Exploring Educational eBooks
- 14. Embracing eBook Trends
 - Integration of Multimedia Elements
 - Interactive and Gamified eBooks

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