

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

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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



Sleep And Respiration

Nicholas A. Saunders, Colin E. Sullivan



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Fundamentals of Sleep Technology Nic Butkov, Teofilo L. Lee-Chiong, 2007 This text provides a thorough understanding of the use of polysomnography and other technologies in the evaluation and management of sleep disorders Coverage includes in depth reviews of the neurophysiology and cardiopulmonary aspects of sleep and the pathophysiology of sleep disorders Detailed sections on polysomnography include recording procedures identifying and scoring sleep stages and sleep related events and report generation Chapters discuss therapeutic interventions including positive airway pressure supplemental oxygen surgical and pharmacologic treatments and patient education A section focuses on pediatric sleep disorders and polysomnography Also included are chapters on establishing and managing a sleep center and accrediting a sleep program Sleep and Breathing Disorders E-Book Meir H. Kryger, 2016-07-01 Written specifically for non sleep

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Control of Breathing during Sleep Susmita Chowdhuri,M. Safwan Badr,James A Rowley,2022-08-16 This book describes control of ventilation during sleep in both health and disease states The topics are presented in a fashion that can be easily comprehended with many figures to illustrate complex concepts Thus a wide range of topics starting from the site of normal respiratory rhythm generation to chemoreceptor control of sleep apnea description of the apneic threshold pathophysiology of upper airway closure novel techniques to measure control of breathing effect of cerebral blood flow on breathing effect of opioids on ventilation effect of heart failure on ventilation genetic aspects of breathing disorders age and gender differences and various therapies are discussed Key Features Helps to bridge the gap between straight forward physiology and clinical practice through a range of topics and use of case vignettes Explores various aspects of clinical management and control which is beneficial to sleep clinicians respiratory physiologists intensivists trainees and researchers Distills complex concepts into understandable language and figures providing helping resource to the clinicians that transforms a dry topic viz control of ventilation into an exciting understandable clinician language *Sleep and Respiration in Aging Adults* Samuel T. Kuna,Paul M. Suratt,John E. Remmers,1991 *National Institutes of Health* Linda T. Kohn,2011 This is a print on demand edition of a hard to find publication The Amer Recovery and Reinvestment Act of 2009 included 10 4 billion in funding for NIH Of the NIH Recovery Act funding 8 2 billion was to be used to support additional scientific research and 400 million for comparative effectiveness research including extramural research at universities and research institutions This report examined how NIH awarded Recovery Act funds for scientific research and the information that NIH made available about the award of these funds It describes the 1 process and criteria NIH used to award extramural grants using Recovery Act funding and 2 characteristics of Recovery Act extramural grants and the information made publicly available about these grants Illus *Sleep and Breathing* Nicholas A. Saunders,Colin E. Sullivan,1984 Providing up to date information on the neurochemistry and physiology of sleep and breathing in the normal state the comprehensive 2nd edition of this monograph details current understanding of the epidemiology pathophysiology clinical features and treatment of disordered breathing during sleep in various syndromes occurring from birth to old age offering perspectives on sleep specific abnormalities as well as the effects of sleep on other disorders such as chronic lung disease Annotation copyright by Book News Inc Portland OR *Obesity Hypoventilation Syndrome* Aiman Tulaimat,2020-07-26 Obesity Hypoventilation Syndrome From Physiologic

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