

# 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 as respiratory abnormalities, periodic legs movements (PLM) and paroxysmal motor arousals (PMA) 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 Rubenski. 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 is 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,<sup>1</sup> 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.<sup>2</sup> There is a hallmark

# Sleep And Respiration

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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 medicine specialists this one of a kind reference guides you through every aspect of this complex field with authoritative advice from Dr Meir Kryger and other experts. Vibrantly illustrated in full color it thoroughly covers the types of patients you're most likely to see in the outpatient setting and brings you up to date with the latest treatment methods both surgical and non surgical. Consult this title on your favorite e reader conduct rapid searches and adjust font sizes for optimal readability. Complete coverage of the patients you're most likely to encounter including special populations such as those with COPD, obesity, hypoventilation, pregnancy, neuromuscular disorders, stroke and more. Specific information on topics of interest to a wide variety of physicians heart failure and sleep disordered breathing surgical treatments including hypoglossal nerve stimulation, oral appliances and the treatment of sleep apnea home sleep testing and different types of PAP devices. Ideal for pulmonologists, otolaryngologists, cardiologists, primary care physicians, dentists and other healthcare providers who encounter patients with sleep and breathing disorders. Full color illustrations throughout including a profusely illustrated chapter on physical diagnosis.

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[Sleep and Respiration](#) John E. Remmers, Paul Suratt, 2000    **Sleep Research**, 1994    **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 vis a viz control of ventilation into an exciting understandable clinician language

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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 Principles to Clinical Practice summarizes the current state of knowledge regarding the epidemiology physiology and treatment of obesity hypoventilation syndrome OHS Currently the identification and management of OHS is suboptimal especially in the acute setting hence the misdiagnosis or mislabeling of the problem has a significant impact on patient outcomes This volume brings together all aspects of assessment and management into a main resource for understanding the complex physiological and clinical consequences of this condition Provides one page chapter summaries that cover epidemiology physiology and treatment options Presents an easy to use reference on obesity hypoventilation syndrome including symptoms Contains chapters with detailed discussions of topics including color images graphs and tables that summarize current research

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**Regulation of Respiration During Sleep and Anesthesia** Robert Fitzgerald, 2012-11-26 During the 1976 Fall Meeting of the American Physiological Society Dr Lahiri and I learned that no plans were being formulated for holding a symposium on Respiratory Control during the 1977 International Congress. Not to hold such a symposium we felt would be the loss of a unique opportunity for us regulationists to exchange ideas *viva voce* with our international colleagues. It would also break a tradition most recently enjoyed at Srinagar India in 1974 and at Warsaw Poland in 1971. After a time consuming false start we had the good fortune to get advice from Dr Pierre Dejours to make our plans known to Dr Henry Gautier. This we did. There resulted an excellent three days of discussion and hospitality at the Faculte de Medecine Saint Antoine in Paris immediately preceding the International Congress. The aim of the co chairmen was to gather an international mixture of both the younger and more senior experts to discuss, argue and maybe even agree on a point or two arising from their current investigations. We wanted to feature however recent research trying to determine the influence of sleep and anesthesia on the regulation of respiration. That the sessions were lively and presumably profitable was never more in evidence than when on the second day at 6:15 p.m. after fully nine hours of discussion, argumentation and some agreement I counted 78 participants still participating. Saint Bartholomew's Hospital Reports ... London (England). Saint Bartholomew's Hospital, 1893 Includes Statistical tables of patients under treatment. *The medical record*, 1880. *Index Medicus*, 2002-07 Vols for 1963 include as pt 2 of the Jan issue Medical subject headings.

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