

# 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](mailto: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,<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



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**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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**The American Review of Respiratory Disease**, 1991 Includes Abstracts section previously issued separately

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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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