pii: jc-18-00240 https://dx.doi.org/10.5664/jcsm.7594

JCSM
Journal of Clinical Sleep Medicine

CASE REPORTS

Sleep-Related Rhythmic Movement Disorder in Triplets: Evidence for Genetic Predisposition?

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Sleep-related rhythmic movement disorder (RMD) is common in very young children but rarely persists beyond childhood. Despite its high frequency, the underlying pathophysiology remains unclear. Familial occurrence is rare. Here we present monozygotic female triplets, all of them being affected by body rolling in terms of RMD. Furthermore, they all present with an additional genetic disease, cystic fibrosis, with the same documented mutation of the cystic fibrosis transmembrane conductance regulator gene (F508del-CFTR). Because all three monozygotic siblings are concordant for RMD, genetic factors may contribute to the time course of the disorder.

Keywords: familial form, genetic predisposition, sleep-related rhythmic movement disorder

Citation: Hayward-Koennecke HK, Werth E, Valko PO, Baumann CR, Poryazova R. Sleep-related rhythmic movement disorder in triplets: evidence for genetic predisposition? *J Clin Sleep Med.* 2019;15(1):157–158.

INTRODUCTION

Sleep-related rhythmic movement disorder (RMD) is a benign condition that comprises a variety of disorders characterized by repetitive, stereotyped, rhythmic movements of the large muscle groups of different parts of the body associated with sleep.[1] RMD usually emerges during transition from wakefulness to sleep but also during consolidated sleep. The condition affects mostly infants and toddlers, with a frequency drop from 59% at 9 months to 5% at 5 years.[2,3] RMD rarely persists into adulthood.[4]

The etiology of RMD remains unknown. Self-soothing behavior, vestibular self-stimulation, or anxiety relief have been suggested, and RMD can be associated with psychiatric comorbidities or developmental disabilities.[4,5] Other conditions, possibly mimicking RMD, need to be ruled out: sleep-related seizures, parasomnias, periodic limb movement disorder, rapid eye movement sleep behavior disorder, and sleep myoclonus or tics. Familial occurrence seems to be rare.[6] Walen reported on a pair of twins with jactatio capitis,[7] but other RMD cases in twins, to our knowledge, have not yet been published.

REPORT OF CASE

Monozygotic female triplets, aged 21 years, were referred for assessment of suspected parasomnia. All three, but especially the two younger girls of the triplets, showed repetitive, rhythmic movements at night from early childhood on, occurring on a daily basis at sleep onset and during sleep. The nocturnal episodes had been observed by the partners of the three young women, whereas the patients themselves could usually not remember these nocturnal events on awakening. The patients complained mainly about nonrestorative sleep and daytime sleepiness (Epworth Sleepiness Scale score 12/14 and 18/24 respectively). Prenatal and perinatal histories were unremarkable and psychomotor milestones were normal, but cystic fibrosis (CF) was diagnosed in all triplets at age 12 years, with all three showing a mutation of the cystic fibrosis transmembrane conductance regulator gene (F508del-CFTR). Triplet 1 had mild primary nocturnal enuresis. Otherwise medical history was negative for tics, obsessive compulsive disorders, or other sleep-wake disorders. Seven half brothers and sisters, who lived together with the triplets, were not affected by RMD or any other sleep disorder. The patients denied alcohol or substance abuse. The neurological examination was unremarkable. Two of the triplets underwent video polysomnography (PSG) (**Video 1** and **Video 2**). The PSG of one triplet showed a 16-minute period of body rolling before falling asleep and shorter episodes of body rolling following arousals from stage N1 and N2 sleep. The second triplet showed a short episode of body-rolling movements during stage N2 sleep. No other sleep pathology, especially no respiratory events, epileptic activity, or tonic or phasic electromyographic abnormalities during REM sleep, could be detected (**Table 1**). The third triplet was only mildly affected and did not wish to undergo PSG. Sleep-modulating therapy was discussed with the triplet with long body-rolling episodes but was not wanted.

DISCUSSION

The presence of RMD in triplets is remarkable, as monozygotic RMD cases have only rarely been reported.[1] In addition, a genetic disease, CF, affected all triplets. The association of RMD and CF could be explained by pure chance alone, although poor

Table 1—Findings of video polysomnography in two triplets with sleep-related rhythmic movement disorder.

Polysomnography Findings	Triplet 1	Triplet 2
Latency to stage N2 sleep (minutes)	47.0	28.0
Sleep efficiency (%)	86.6	92.6
Stage N1 sleep (% TST)	13.6	4.4
Stage N2 sleep (% TST)	55.6	52.7
Stage N3 sleep (% TST)	18.8	21.0
Stage R sleep (% TST)	12.0	21.9
Arousal index (events/h)	6.7	5.0
Periodic limb movement in sleep index (events/h)	1.8	0.8
Apnea-hypopnea index (events/h)	0.2	0.0
Oxygen desaturation index (events/h)	1.8	0.5
Mean oxygen saturation in sleep (%)	94.8	95.1
Total number of sleep-related rhythmic movements	One episode of body rolling (after an arousal from stage N2 sleep), 14 episodes of rocking leg and pelvic movements (mostly after arousals from stage N1 and N2 sleep, once out of stage N2 sleep without an arousal, once after an arousal from stage N3 sleep)	Four episodes of body rolling (one at sleep onset, lasting 16 minutes, one from stage N1 sleep, and two from stage N2 sleep, each with a short arousal over a few seconds, yet without awakening, each lasting approximately 1 minute)
Epworth Sleepiness Scale score	12/24	18/24
Fatigue Severity Scale score	3.4/7	5.5/7

TST = total sleep time.

sleep quality is characteristic for both conditions. In addition, daytime sleepiness has been described in patients with CF and severe lung disease, and is typically associated with reduced SaO_2, even in the absence of apneas.[8] Nocturnal hypoxemia in CF might enhance the likelihood of RMD by facilitating arousal-related mechanisms,[9,10] and treatment with continuous positive airway pressure may resolve RMD.[9] In the triplets in this study, however, PSG revealed normal breathing parameters, and sleep appeared normally consolidated. The potential etiological link between CF and RMD remains therefore elusive, at least in view of the PSG findings. Although earlier studies advocated psychological or behavioral mechanisms underlying RMD,[4] our observation now suggests that genetic factors may also be involved, particularly when RMD persists beyond childhood.

ABBREVIATIONS

CF, cystic fibrosis
PSG, polysomnography
RMD, rhythmic movement disorder

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SUBMISSION & CORRESPONDENCE INFORMATION

Submitted for publication May 2, 2018
Submitted in final revised form August 21, 2018
Accepted for publication October 1, 2018

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DISCLOSURE STATEMENT

All authors have seen and approved the manuscript. The authors report no conflicts of interest.