Sleep Disorders in Patients with Myotonic Dystrophies

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Abstract

Myotonic dystrophies (DM) are multisystem autosomal dominant neuromuscular disorders characterized by myotonia, progressive muscle weakness, and systemic involvement. The main forms are myotonic dystrophy type 1 (DM1), caused by CTG repeat expansion in the *DMPK* gene, and myotonic dystrophy type 2 (DM2), caused by CCTG repeat expansion in the *CNBP* gene. Sleep disorders, particularly excessive daytime sleepiness (EDS), are among the most important nonmotor manifestations and significantly impair quality of life. This paper presents a narrative review of clinical, neurophysiological, and polysomnographic studies investigating sleep disorders in DM1 and DM2. The review addresses abnormalities of sleep architecture, sleep-related breathing disorders, neuroimaging findings, and pharmacological treatment outcomes. Excessive daytime sleepiness is highly prevalent in both DM1 and DM2, although it is generally more severe in DM1. Polysomnographic studies in DM1 demonstrate REM sleep dysregulation and NREM sleep abnormalities that may contribute to hypersomnolence. Sleep disturbances in myotonic dystrophies have a multifactorial origin involving both central and peripheral mechanisms. Dysfunction of central sleep–wake regulation, circadian rhythm abnormalities, and alterations in serotonergic and orexinergic pathways appear to play important roles. Sleep-related breathing disorders, including obstructive and central sleep apnea, are also common and may contribute to nocturnal hypoxemia and daytime hypercapnia. Neuroimaging studies provide additional evidence of central nervous system involvement in sleep regulation abnormalities. Pharmacological treatment with modafinil may improve excessive daytime sleepiness, while mexiletine can reduce myotonia, although its potential cardiac effects require appropriate assessment and monitoring. Early recognition and multidisciplinary management of sleep disorders are essential for improving functional outcomes and quality of life in patients with myotonic dystrophies.

Keywords: myotonic dystrophy type 1; myotonic dystrophy type 2; sleep disorders; excessive daytime sleepiness; polysomnography

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Poremećaji spavanja u bolesnika s miotoničnim distrofijama

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Sažetak

Miotonične distrofije (DM) multisistemske su autosomno dominantne neuromuskularne bolesti obilježene miotonijom, progresivnom mišićnom slabošću i zahvaćanjem brojnih organskih sustava. Glavni oblici bolesti su miotonična distrofija tipa 1 (DM1), uzrokovana ekspanzijom CTG ponavljanja u genu *DMPK*, i miotonična distrofija tipa 2 (DM2), uzrokovana ekspanzijom CCTG ponavljanja u genu *CNBP*. Poremećaji spavanja, osobito prekomjerna dnevna pospanost (EDS), među najvažnijim su nemotoričkim manifestacijama bolesti i značajno narušavaju kvalitetu života. Rad predstavlja narativni pregled kliničkih, neurofizioloških i polisomnografskih istraživanja poremećaja spavanja u DM1 i DM2. Obuhvaćeni su poremećaji arhitekture spavanja, poremećaji disanja tijekom spavanja, neuroradiološki nalazi i ishodi farmakološkog liječenja. Prekomjerna dnevna pospanost vrlo je česta u oba oblika bolesti, ali je uglavnom izraženija u DM1. Polisomnografska istraživanja u DM1 pokazuju poremećaje regulacije REM spavanja i promjene NREM spavanja koje mogu pridonijeti hipersomnolenciji. Poremećaji spavanja u miotoničnim distrofijama imaju multifaktorijalnu podlogu koja uključuje središnje i periferne mehanizme. Važnu ulogu imaju poremećaji središnje regulacije ciklusa spavanje–budnost, cirkadijanog ritma te serotonergičkih i oreksinergičkih putova. Česti su i poremećaji disanja tijekom spavanja, uključujući opstruktivnu i centralnu apneju, koji mogu pridonijeti noćnoj hipoksemiji i dnevnoj hiperkapniji. Neuroradiološka istraživanja pružaju dodatne dokaze o zahvaćenosti središnjega živčanog sustava u poremećajima regulacije spavanja. Farmakološko liječenje modafinilom može ublažiti prekomjernu dnevnu pospanost, dok meksiletin može smanjiti miotoniju, uz potrebu za odgovarajućom procjenom i praćenjem mogućih srčanih učinaka. Rano prepoznavanje i multidisciplinarno liječenje poremećaja spavanja ključni su za poboljšanje funkcionalnih ishoda i kvalitete života bolesnika s miotoničnim distrofijama.

Ključne riječi: miotonična distrofija tipa 1; miotonična distrofija tipa 2; poremećaji spavanja; prekomjerna dnevna pospanost; polisomnografija

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