MDS-ES/EAN: Differential diagnosis of sleep related movement disorders - Level 3

Other sleep related movement disorders: Common and rare differential diagnosis based on clinical and PSG features

Federica Provini
Bologna, Italy

Email: federica.provini@unibo.it
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According to the International Classification of Sleep Disorders, sleep related movement disorders are primarily characterized by relatively simple, usually stereotyped, movements that disturb sleep or its onset. They comprise Periodic Limb Movement Disorder, Leg Cramps, Sleep Related Bruxism, Sleep Related Rhythmic Movement Disorder, and movement disorders due to drug or medical conditions. Some of these movements, such as bruxism, might occur both during wakefulness and sleep, but a clear worsening of the symptoms during sleep is necessary in order to include the condition among the Sleep Related Movement Disorder. Sleep Related Movement Disorders might occasionally be present in healthy individuals but the manifestations must disturb sleep with daytime consequences in order to be classified into this group of disorders. Sleep Related Movement Disorders must be distinguished from parasomnias, such as Sleepwalking or Rapid Eye Movement Sleep Behaviour Disorder (RBD), which normally show complex muscular pattern and complex behaviours, that may appear purposeful, although unconscious, too. Other movements, such as Sleep Starts (Hypnic Jerks), might occasionally be present in healthy individuals.

The employment of an all-night video-polysomnography recording is necessary to describe properly the neurophysiologic features of the phenomenon, the time and the stage of occurrence during the night, and the patients’ level of consciousness, in order to reach a correct diagnosis.
This chapter describes the sleep-related salient characteristics and distinguishing features of some movement disorders, ranging from some simple movements up to more complex behaviours classified within epileptic seizures.

**Sleep related rhythmic movement disorder**

Also known as “jactatio capitis nocturna” or “headbanging” or “headrolling”, the term Rhythmic movement disorder (RMD) is preferred as different body areas may be involved in the movement activity. RMD consists of repetitive and stereotyped movements of the head, neck, trunk, and sometimes also legs. The movements are predominantly sleep related, occurring near nap or bedtime, or when the individual appears drowsy or asleep.

These stereotypic movements may last a few or several minutes, repeating at a frequency of 0.5-2 per second. RMD is seen in normal children, but it has also been reported in mentally retarded and autistic patients. In these patients, RMD may persist during wakefulness and into adulthood, not rarely associated with other “stereotypies” i.e. the rhythmic habit pattern.

Serious injuries from sleep-related RMD are rare, but include bone skull injuries.

The association of RMD with long-lasting restless legs syndrome (RLS) is well-known and RMD may also occur in RLS of recent onset.

Rhythmic feet movement, formerly hypnagogic foot tremor (0.5-3 Hz), occurring during pre-sleep wakefulness and light sleep may be considered a new kind of RMD arising in adults, in some cases associated with insomnia, sleep apnoea, periodic limb movements and RLS.
Brief activation of tibialis anterior in one leg alternating with similar activation in the other leg, so-called alternating leg muscles activity (ALMA), has been described. Such activations, similar to rhythmic feet movements while falling asleep, occur at a frequency of 1-1.5 Hz, each lasting up to 0.5 s, with sequences of several to twenty seconds and recurring particularly during arousal. ALMA has been described in patients with sleep apnoea, those taking antidepressant medication, and with periodic limb movements and RLS.

The specific causes and physiologic mechanisms underlying RMD remain uncertain. The absence of organic causes in the majority of cases has led to behavioural and psychological theories. Other hypothesis proposes that RMD is linked to arousal fluctuations and mediated via central motor pattern generators of the brainstem.

Provided that RMD does not substantially affect sleep quality or daytime function, the condition does not require any treatment. For particularly violent forms of RMD, use of protective measures is appropriate. Clonazepam (1 mg nightly), oxazepam (10-20 mg nightly), citalopram, imipramine and behavioural therapies have been reported to produce significant improvement in some cases.

**HYPNIC JERKS also called HYPNAGOGIC JERKS or SLEEP STARTS**

Hypnic jerks, otherwise known as sleep starts or hypnagogic jerks, represent a normal accompaniment of sleep. Hypnic jerks consist in non-periodic myoclonic movements, usually involving asynchronically different and isolated body segments, that occur mainly at sleep onset and/or with K-complexes or an EEG arousal. They are associated with autonomic activation tachypnea and sudomotor activity, and sometimes with a
peculiar sensory feeling of “shock” or “falling into the void”. These non-
stereotyped myoclonic jerks are often triggered by fatigue, stress, sleep
deprivation, vigorous exercise and stimulants like caffeine and nicotine. In
some cases they intensify and are the cause of some concern, occasionally
entering the differential diagnosis of epileptic myoclonic seizures,
especially when recurring repetitively in neurologically impaired children.
When particularly frequent and severe, hypnic jerks have been reported
as a cause for sleep-onset insomnia.
HJs are a frequent sleep-related motor phenomenon in patients with
Parkinson Disease and Atypical Parkinsonisms, appearing since the early
stages of these diseases and unrestricted to a specific disease type.
Detailed neurophysiological analysis of the HJs in healthy subjects and
patients with parkinsonism, including the lack of any ordered propagation
of the spreading of muscular contraction suggested a subcortical origin of
the jerks. HJs presumably arise from sudden descending volleys that
originate in the brainstem reticular formation and are activated mostly at
the transition between sleep and wake. The variability in motor
recruitment and pattern during HJs may be related to variable spinal
motor neuron excitability in the spinal cord during sleep, indicating the
engagement of different and sometimes unsynchronized pools of spinal
motor neurons at different times for each HJs.
Usually sleep starts are common physiological phenomenon affecting up to
70% of the adult population and their course is benign, resolving without
any neurological sequel. Therefore reassurance and counselling are all
that is needed to reassure the patient. Some patients may require a small
dose of clonazepam (0.5-1 mg at bedtime) to ameliorate the symptoms on
a short-term basis.
Propriospinal myoclonus at the sleep wake transition

Propriospinal myoclonus (PSM) is a special type of spinal myoclonus characterized by brief, repetitive, mainly arrhythmic jerks. The jerks usually arise in the muscles corresponding to a given thoracic myelomere and then progressively and synchronously spread in rostral and caudal directions to the adjacent myotome at low velocity along propriospinal pathways. The jerks usually provoke a flexion movement, but less frequently an extension pattern has been reported.

Most patients with PSM have no recognizable cause.

PSM at the wake-sleep transition recurs only during drowsiness preceding falling asleep and, rarely, during intrasleep relaxed wakefulness and upon awakening in the morning. The disorder can make it very hard to fall asleep with consequent severe insomnia.

Videopolysomnography confirms that the myoclonic activity is restricted to the wakefulness period preceding sleep onset or falling to sleep, and manifests as trunk flexion or, less frequently, extension. The EMG discharges last typically 100-300 ms but sometimes longer with both reciprocal and co-contracting agonist-antagonist activity. The major characteristic of the EMG discharges is a simultaneous bilateral rostral and caudal slow recruitment (2-16 m/s) from the area of spinal cord origin, sparing the cranial muscles, in agreement with a propriospinal pattern of propagation. Jerks may recur at quasi-periodic intervals (every 5-40 seconds). Mental activation with the patient comfortably lying down or sitting (asking the patient to think, speak, count, and perform simple or complex motor tasks such as waving a hand or writing) desynchronizes the EEG activity and makes the jerks disappear.

Absence of a cortical pre-movement potential, lack of involvement of cranial muscles, pattern of propagation, variable delay between different
muscles, and slow propagation are all features that distinguish PSM from cortical and reticular reflex myoclonus.

“Diaphragmatic flutter”, “moving umbilicus syndrome”, “belly dancer’s dyskinesia, painful legs and moving toes and excessive fragmentary hypnic myoclonus are conditions clearly different from PSM.

Periodic limb movements also may appear during relaxed wakefulness, but in such cases they continue during light sleep stages. Moreover, their motor pattern is different, as periodic movements in sleep (PLMS) consist of dorsiflexion of the big toe and foot and flexion of the knee and hip, even if they may coexist with PSM in some patients with RLS. In such cases, PLMS and PSM could represent unrelated motor phenomena, coexisting either by chance or only in a subset of RLS patients, but due to different pathophysiological mechanisms as suggested by the temporal relationship of these two motor phenomena: PSM appears only during relaxed wakefulness and disappears with sleep onset, when PLMS in turn arise.

In around 80% of cases, PSM appears to be idiopathic, the remainder comprising patients with a broad range of medical conditions, including spinal lesions, namely cervical hemangioblastoma or cervical herniations, syringomyelia and dural arteriovenous fistula, paraneoplastic condition, paraproteinemic neuropathy and demyelinating lesions. A psychogenic etiology should be also considered because the motor pattern of PSM can be mimicked voluntarily.

There is no known effective treatment for PSM. It responds only in part to clonazepam (up to 2 mg at night). Other treatment options includes carbamazepine (up to 400 mg/day), gabapentin (up to 800 mg/day), levetiracetam (up to 2000 mg), pramipexole (up to 0.7 mg at night) and tramadol (up to 100 mg at night).
Sleep related hypermotor epilepsy

Sleep-related hypermotor epilepsy (SHE), previously known as nocturnal frontal lobe epilepsy (NFLE), is a rare form of focal epilepsy characterized by hypermotor seizures occurring predominantly during non-REM sleep. The clinical features and diagnostic criteria of SHE were recently revised during an international consensus conference.

The most common clinical expression consists of “hypermotor” events characterized by vigorous hyperkinetic features (complex body movements with kicking or cycling of limbs and rocking body movements), usually with vegetative signs, vocalization, and emotional facial expression. Asymmetric tonic/dystonic seizures with or without head/eye deviation are also observed. Typically, patients with SHE present a series of sleep-related motor events becoming increasingly long and complex, even during the same night. Clinical features range from brusque stereotyped arousals (i.e., paroxysmal arousals), repeated throughout the night, to complex hypermotor seizures and, more seldom, protracted ambulatory behavior known as epileptic nocturnal wandering.

Seizure onset may be at any age with a peak during childhood and adolescence. Seizure frequency may be very high, with occurrence either every night or almost every night, usually many times per night. Seizures are abrupt in onset and offset, typically brief and have a highly stereotyped motor pattern within individuals.
References


