

Chapter 128

Approach to a Patient with a Potential Sleep Disorder

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Man spends almost a third of his lifetime in sleep, but medical science has only woken-up to the importance of sleep disorders in the last two decades during which knowledge in this field has increased considerably. The development of tools like polysomnography (PSG) has provided us with means to research, diagnose, and treat these disorders scientifically. The International Classification of Sleep Disorders (ICSD) currently lists 85 disorders, each presented in detail with a descriptive text and diagnostic criteria¹. Conditions like obstructive sleep apnea (OSA) are common in the general population and frequently co-exist with other prevalent disorders in the general population like hypertension, ischemic heart disease, metabolic syndrome, neurological, gastroenterological, respiratory and endocrinal disorders. OSA can result from some of these, e.g. acromegaly, hypothyroidism on one hand while on the other hand it can compound morbidity in conditions like chronic obstructive pulmonary disease, ischemic heart disease, etc. Its etiological contribution to hypertension has also been sanctified². A two-way relationship may hence exist. A physician practicing any branch or medicine therefore must now acquire a working knowledge of sleep disorders. An abridged classification of sleep disorders based on symptoms is presented in Tables 1 and 2. The reader is advised to refer to a detailed classification for more complete information¹.

The present article discusses the symptoms and signs that point towards sleep disorders, the methods used to arrive at a differential diagnosis, and very briefly a few investigative modalities used to confirm a suspected disorder. In all these respects, sleep medicine follows the same practices as are being used by physicians in other branches of medicine.

CLINICAL HISTORY AND SYMPTOMS

Sleep history is best taken from the patient as well as the bed partner or a family member who is well placed to notice and report on symptoms and signs of which the patient may have no knowledge, e.g. snoring, abnormal movement or behavior during sleep. Sleep disorders present with three traditionally recognized symptoms, i.e. insomnia; daytime hypersomnolence or sleepiness (DS), and abnormal movements; behavior or sensations during, sleep or on arousal. Snoring and 'partner observed' apnea episodes have also been accepted as pointers to OSA.

INSOMNIA (Table 1): Insomnia is the perception of insufficient, inadequate, or no restorative sleep by the complainant. Its presence or absence cannot be

Table 1: Sleep disorders presenting with symptoms of dyssomnia (insomnia and daytime hypersomnolence or sleepiness)

<i>Intrinsic causes: cause within or inherent</i>	<i>Extrinsic: cause without or outside</i>
Obstructive sleep apnea syndrome	Environment dependant sleep disorders
Restless leg syndrome (RLS)	Insufficient sleep syndrome
Paroxysmal leg movement disorder (PLMD)	Adjustment sleep disorder
Narcolepsy, idiopathic insomnia	Alcohol or stimulant dependant disorders
Psychophysiological insomnia	Circadian rhythm disorders, e.g. Jet lag syndrome;
Sleep state misperception	Delayed or advanced sleep phase syndrome,
Central sleep apnea	Shift work sleep disorder
Idiopathic hypersomnia	

concluded on mere duration of sleep as the normal requirement is very variable. The key difference is the absence in normal individuals with short sleep requirement of symptoms which insomniacs complain of, e.g. irritability, restlessness, mood changes, memory defects, etc. Insomniacs also complain of difficulty in falling asleep, mid-sleep awakening which may be of short duration (days); intermediate duration (weeks) or chronic (months). At any given time, 10% of the general population may have chronic insomnia which peaks in the sixth decade, slightly favoring the female gender^{3,4}. Insomniacs also present with a greater proportion of mood and personality disorders when compared to the normal population.

The largest segment of insomniacs have psychiatric or psychological basis as its cause⁵. Medical diseases resulting in pain or discomfort, e.g. radiculopathy, asthma, heart failure, degenerative neurological conditions contribute to another significant number. Medications like amphetamines, steroids, β -2 agonists may stimulate the CNS. Withdrawal of hypnotic drugs may precipitate rebound insomnia. Circadian rhythm disorders resulting from asynchrony between the biological clock and requirement of a temporal clock may cause insomnia in disorders such as advanced or delayed sleep phase syndrome, jet lag or shift workers syndrome. Sleep state misperception is a situation where a sleeping patient is genuinely unable to realize that he has slept. Poor sleep hygiene and psycho-physiological conditioning to the surrounding of the bedroom may produce insomnia in a Pavlovian reflex. Restless leg syndrome (RLS) is a disorder where attempts to sleep evoke creepy, crawling, unpleasant sensations in the legs that disappear on leg movements, only to return on trying to sleep. Iron deficiency anemia, chronic renal failure, peripheral neuropathy, are some secondary causes in which it is described.

DAYTIME HYPERSOMNOLENCE OR SLEEPINESS (Table 2)

Sleep is normal when one desires to sleep as and when tired or at usual bedtime. DS may normally be also observed if night sleep was disturbed. When DS arrives unexpected, unwanted, and at an inappropriate time when the subject is occupied in activities like driving, eating, talking, it is definitely abnormal. Such episodes may be called sleep attacks and are potentially dangerous for the victim as well as others if they occur at a time when one is driving. Chronic DS is perceived by the patient as fatigue, weakness, lack of energy or drive. Repeated yawning, head nodding, drooping

Table 2: Sleep disorders presenting with abnormal movements behaviors sensations during sleep or on arousal

Primary sleep phenomenon	Secondary sleep phenomenon
REM sleep—RBD; Dreams, nightmares	CNS—Seizure; Headache
NREM sleep—confusional arousal	CVS—Angina, arrhythmias
Sleepwalking, sleep terror	Resp—Sleep hiccup, asthma
Either or Missln-Bruxism, enuresis, RLS, PLMD, sleep talking	EGE—Ge reflux Missln—Panic attacks, leg cramps

eyelids and impaired neuro-cognitive functions may be noticed. Mood and personality disorders are not uncommon. Epworth Sleepiness Scale summarized in Table 3 is a useful bedside tool to quantify DS⁶.

Inadequate sleep, as in insufficient sleep syndrome, may be the cause. Circadian rhythm disorders can also result in DS in a reversal of the manner by which they cause insomnia. OSA is a common cause of DS where disruptive snoring, observed apnea in an obese individual point to the diagnosis. History of episodic weakness after strong emotions, i.e. cataplexy in an individual with DS is a hallmark of narcolepsy. In a florid case history of sleep paralysis, visual, tactile, or auditory hallucinations at sleep onset or awakening may also be obtained.

ABNORMAL MOVEMENTS, SENSATIONS, AND BEHAVIOR

(Table 3 - Parasomnias) Conditions such as grinding teeth (Bruxism), enuresis, sleep walking, sleep talking, sleep terror, sleep drunkenness, nightmares—aptly

Table 3: Epworth Sleepiness Scale

Chances of falling asleep unplanned	Nil / Slight / Moderate / High in situations
Sitting and reading	
Inactive sitting – as in meetings	
As a passenger in a car for over an hour	
On lying down to rest during afternoon	
Sitting and talking	
Sitting after lunch without alcohol	
Watching TV	
In a car when stopped for a few minutes at traffic signal	
Score 0 for Nil, 1 for Slight, 2 for Moderate, 3 for High Maximum score possible is 24 Normal controls: usually up to 10	

describe the main clinical presentation. Rapid eye movement behavior disorder (RBD) is a dramatic condition in which unusual violent dreams experienced by the individual are enacted. Injuries to self and others may be caused, leading to medicolegal issues. Withdrawal from alcohol and sedatives can precipitate it. An association with narcolepsy and CNS pathologies has been established. PSG is confirmatory. Periodic leg movement disorder (PLMD) is a condition characterized by movements occurring every 20-40 seconds resulting in a brief arousal which can be only detected on a PSG even when resulting in significant distortion in sleep architecture. Most patients with RLS manifest PLMD though the vice versa is not true.

Sleep disorders presenting with parasomnias without systemic disease are called primary. These are further classified according to their time of occurrence on a PSG record as REM; NREM or no miscellaneous. Parasomnias arising as a manifestation of systemic disorder are called secondary⁷.

Snoring: Snore is a characteristic sound usually inspiratory, produced in the throat due to vibration of structures such as soft palate, uvula, faucial pillars, pharyngeal walls, and nearby structures during sleep. A wide range of prevalence between 3 and 86% has been described in different populations, reflecting the subjectiveness of the complaint and variance in methodologies in studies. Snorers are more commonly male, and the incidence peaks in the fifth-to-sixth decade^{8,9}.

The importance of snoring lies in its association with OSA and consequent co-morbidities¹⁰. It is almost always present in populations reported from sleep clinics. Such individuals are usually obese with definite daytime symptoms, whose bed partners have noticed that the patient "stops breathing" or "chokes" in sleep. This is usually followed by a loud snore when the patient resumes breathing sometimes called a "resuscitative snore". The presence or absence of snoring by itself cannot be used to exclude or include the diagnosis of OSA. Its presence in conjugation with other symptoms and signs considerably increases the sensitivity. Snoring that occurs without DS is sometimes referred to as "simple snoring". It may not prove to be as simple, since studies that have taken care to remove the confounding effect of OSA suggest linkages to adverse medical outcome of the type seen in OSA. This is an active area of research¹¹.

Observed apnea: These are episodes described earlier when patient stops breathing. Closer observation in the episode described may reveal continued

respiratory effort to breath. This is a sign of obstructive apnea seen in OSA. Absence of effort would suggest central apnea seen in heart failure or metabolic disorders. PSG is required for accurate classification.

It must be stated that more than one symptom may be present in a sleep disorder. Patients with OSA may present with DS, insomnia, snoring, and observed apnea. Narcolepsy may present with cataplexy, DS, and abnormal sensations or behaviors. A detailed sleep history is recorded. The time when the patient lies down to sleep, the time taken to fall asleep (< 20 mts), any awakening during sleep, usual time of getting up from bed, whether refreshed on arising, are ascertained for weekdays as well as holidays. An assessment of the quality and quantity of sleep can be made. Several questionnaires have been devised to assess the quality and adequacy of sleep, personality and mood defects in patients with insomnia¹². Conditions like insufficient sleep syndrome, circadian rhythm disorders, and idiopathic hypersomnia come under consideration. A history of the waking schedule including daily routine, meals, exercise, and consumption of alcohol, coffee, and medications is also enquired. It is practical to also confirm these details from a close family member. The use of drug, alcohol and medications receive attention. Physical discomfort in the bedroom as a result of noise, adverse temperature, uncomfortable bedding is a feature seen in environmental sleep disorder. Episodes of unwanted DS may suggest a diagnosis of OSA or narcolepsy. Narcolepsy is associated also with history of cataplexy, sleep paralysis, and delusions. Bed partner may report on "choking" of OSA or abnormal movements like chewing, sleep walking, sleep talking or kicking. Sometimes violent and complex behaviors are reported as in RBD disorder and eating behavior disorder. An estimate of stress in the patients' life must also be made. Loss of near and dear ones can produce adjustment sleep disorders. Change of job, weddings in the family, an unexpected visit may also produce sufficient stress to affect sleep. A regular bed time habit suggests bad sleep hygiene, while good sleep in unfamiliar surroundings with an inability to sleep in ones own bedroom suggests psycho-physiological insomnia. Medical history should be thoroughly evaluated as some systemic disorders may manifest during sleep. Similarly, a psychiatric evaluation may be required in individual cases. Occupational history as a cause of sleep disorders is required, as in shift workers, and also for fitness in respect of jobs which require complete alertness like truck drivers and pilots. A good history therefore with a working knowledge of common

sleep disorders helps in obtaining a differential diagnosis.

A detailed physical examination is required since conditions arising from any major system of the body may present during sleep. In addition, special attention should be taken to look for signs supportive of a diagnosis suggested as a differential diagnosis. In case as when OSA is suggested one can look for evidence of obesity, cranio-mandibular abnormalities, obstruction in the upper airway. A systemic evaluation for neuro-cognitive, respiratory, cardiovascular, and metabolic consequences of OSA is carried out. Physical examination adds more information to narrow down the differential diagnosis.

Investigations: These are essentially of two types. The first category is used to confirm an associated causative condition like hypothyroidism, acromegaly, ischemic heart disease, asthma, epilepsy, estimation of drug levels, etc. This category also includes those investigations carried out to estimate consequences of sleep disorders, e.g. cor pulmonale in OSA. The second category comprises those used purely to diagnose and confirm a diagnosis wherever possible. Polysomnography is the prime example in this category. Several physiological parameters like EEG, EOG, ECG, EMG, respiratory movement, oxymetry, limb movements, and air flow, etc. are recorded in an overnight study carried out at a sleep laboratory. It is the current gold standard in the diagnosis of OSA. It also provides diagnostic support in RBD, PLMD, narcolepsy, sleep terror, and other conditions. Epilepsy that enters diagnostic considerations can be confirmed or negated from such a record. Multiple sleep latency test (MSLT) and maintenance of wakefulness (MWT) are tests carried out in a sleep lab where patients are given several opportunities over fixed periods to sleep or remain

awake in a conducive atmosphere while objectively recording sleep. These provide the most objective estimation of DS. Actigraph, a portable device worn on the wrist to record its movements, is also used to semi-quantify sleep duration.

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