Diagnosis of Restless Leg Syndrome (Willis-Ekbom Disease)

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KEYWORDS

• Restless leg syndrome (RLS) • Willis-Ekbom disease (WED) • Diagnostic criteria
• Mimics • Ferritin in RLS

KEY POINTS

- Diagnosis of restless leg syndrome, or Willis-Ekbom disease, (RLS/WED) is based on 5 diagnostic criteria: urge to move, rest worsens the urge, movement provides relief, evening or night worsening of the urge, and the best or sole explanation is RLS/WED.
- Serum ferritin should be tested in patients with moderate-to-severe RLS/WED, established patients with acute worsening of symptoms, or patients with illness producing blood loss.
- Polysomnography or nerve conduction–electromyographic study is not indicated unless a patient has additional clinical symptoms to warrant such testing (hypersomnia or poorly characterized neuropathy or radiculopathy).
- Diagnosis of young children or elderly with cognitive impairment may need evaluation of supportive or associated features and information from parents or caregivers to determine the presence of definite, possible, or probable RLS/WED.

INTRODUCTION

Restless leg syndrome, or Willis-Ekbom disease, (RLS/WED) is a neurosensorimotor disorder that is diagnosed through focused questioning rather than any specific test.[1] Although any limb may be affected, most patients have initial symptoms in the lower extremities and commonly report sleep onset and early maintenance insomnia. RLS/WED affects 2% to 4% of adults in North America and Europe at least twice weekly and is more common in women.[2,3] It is estimated to affect more than 10 million adults in the United States[3] and an estimated 1.5 million children and adolescents.[4] The clinical course of RLS/WED is commonly chronic and progressive in moderate-to-severe primary cases. Early onset of RLS/WED before age 30 is often intermittent and variably problematic until the patient reaches middle age. Presentation after age 50 is usually more rapid and clinically more troublesome.[5–7] On occasion, RLS/WED can be intermittent and enter remission.[8] Sleep onset and maintenance insomnia is often the primary reason for medical consultation.[5,9] Unless a patient has other symptoms of sleep disorder, particularly excessive daytime sleepiness, polysomnography is not needed for diagnosis of the sleep disturbance.[10] A patient with moderate-to-severe RLS/WED may average less than 5 hours of sleep per night. Interestingly, most RLS/WED patients with significant insomnia deny daytime sleepiness. For patients with mild

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RLS/WED, sleep disturbance may be only a minor inconvenience.[11]

Challenges to diagnosis arise when the patient has other medical conditions, such as metabolic disease, neurologic dysfunction, musculoskeletal abnormalities, autoimmune disease, and/or psychiatric disorders. Patients may have difficulty characterizing the sensory discomfort but may use terms such as crawling, creeping, bugs, worms, electricity, energy, burning, pain, ache, or nervous. Symptoms present sitting in a chair, automobile, airplane, or lying down attempting to sleep. The practitioner must systematically question a patient about their sensory and motor experience in relationship to lifestyle, time of day, and concomitant medications to properly characterize the diagnosis in more complex patients. There may also be differences in the presentation of RLS/WED in children or the cognitively impaired.

DIAGNOSTIC CRITERIA

RLS/WED has been variously characterized as primary, familial, idiopathic, early onset, late onset, or secondary (eg, related to iron deficiency, anemia, pregnancy, or end-stage renal disease). Because the cause and genetics of RLS/WED remain under investigation, appropriate diagnosis focuses on symptom presentation. Consensus criteria for the diagnosis of restless leg syndrome were established through a National Institutes of Health conference and published in 2003.[1] Four criteria were defined to arrive at a diagnosis. In 2012, a fifth criterion was added by the International Restless Legs Syndrome Study Group (IRLSSG) to address the potential mimics that could complicate diagnosis.[12] In 2004, the acronym URGE came into use to assist physicians in their diagnostic process. Subsequently, Lee and colleagues[13] added the fifth IRLSSG criteria that restless leg syndrome should be the sole or primary factor to account for the symptomatic complaint. The resulting acronym, URGES, stands for: (1) urge to move that is most commonly noted in the legs, (2) rest worsens the urge to move, (3) gyration or movement of the affected limb improves the urge, (4) evening and night intensification of the urge, and (5) the sole best explanation of the urge is RLS/WED. **Table 1** describes the 5 diagnostic criteria of URGES. The presenting symptom is the patient's urge to move because of a sensory disturbance that may prove difficult for the patient to describe. When the patient is at rest, the urge intensifies, while gyration or getting up to go (stretching or movement of the affected limbs) will at least temporarily provide relief of the urge to move. Unless the patient takes exacerbating medication or has the most severe presentation, the sensory component normally begins to intensify in the evening and first half of the night, lessening over the night and generally being of little problem in the morning hours. The diagnostic criteria have been incorporated in the *International Classification of Sleep Disorders*, 3rd edition, and in other diagnostic manuals.[14]

SPECIAL POPULATIONS: PEDIATRIC AND COGNITIVELY IMPAIRED

Pediatric diagnosis has been modified based on the ability of a child to describe the sensations.[15] The diagnosis is demarcated as definite, probable, or possible RLS/WED. The probable diagnosis of restless leg syndrome has been observed in

Table 1
Diagnostic criteria using the acronym URGES

*U*rge to move the limbs	Urge to move the legs (sometimes arms or other body parts) with or without dysesthesias
*R*est-induced	Onset or exacerbation with both decreased motor activity and mental activation Motor and sensory symptoms most often develop during periods of rest or inactivity, such as sitting or lying
*G*yration or movement improves urge	Relief with movement RLS symptoms are partially or totally relieved by movements such as walking or stretching High mental activation such as computer games may also reduce the urge
*E*vening or night worsening	Circadian pattern RLS symptoms usually occur or worsen in the evening or at bedtime Symptoms are generally minimal in the morning
*S*ole explanation for urge to move	Any other illness or disorder cannot explain the other 4 symptoms