

Narcolepsy

Clinical Background

Narcolepsy is a sleep disorder characterized by excessive daytime somnolence, cataplexy and sleep paralysis.

Epidemiology

- Incidence – 1/1000 to 1/2000
- Age – first symptoms during teenage years
- Sex – M>F

Risk Factors

- Family history of narcolepsy in first degree relative increases risk by 10- to 40-fold
- Genetics – *HLA-DQB1*0602* haplotype found in many patients

Pathophysiology

- Abnormality in hypocretin (orexin) neurotransmitter found in the hypothalamus
 - Hypocretins function to stimulate appetite and arousal; increases muscle tone during awake periods

Clinical Presentation

- Excessive daytime sleepiness (EDS)
- Cataplexy – sudden episode of muscle weakness triggered by emotion
 - Knees buckle (typical)
 - Diaphragm and oculomotor muscles are spared
 - No loss of consciousness
- Sleep paralysis – occurs while falling asleep or waking up
- Hypnagogic hallucinations – intense auditory or visual hallucinations at onset of sleep

Treatment

- Lifestyle modifications
- Medications include modafinil for EDS, antidepressants and gamma hydroxybutyrate for cataplexy

Diagnosis

- Indications for testing – excessive daytime sleepiness without known etiology occurring almost daily for at least 3 months
- Laboratory testing
 - Genetic testing – *HLA* typing
 - Determines presence of *DRB1*15* or *DQB1*0602*; however, not a diagnostic test
 - Highly sensitive in cases with cataplexy
 - Polysomnography (PSG)
 - Overnight PSG followed by multiple sleep latency testing in the morning is diagnostic if positive
 - Positive test result – sleep latency ≤ 8 minutes plus the presence of REM sleep in at least 2 naps

Differential Diagnosis

- Adults
 - Depression
 - Drug abuse
 - Idiopathic hypersomnia

- Periodic limb movement disorder
- Sleep apnea
- Sleep deprivation state
- Seizures
- Children
 - Seizures
 - Sleep-related breathing syndrome
 - Sleep deprivation
 - Idiopathic hypersomnia
 - Schizophrenia
 - Depression
 - Brain tumor

Lab Tests

Indications for Laboratory Testing

Tests generally appear in the order most useful for common clinical situations. For test-specific information, refer to the test number in the ARUP Laboratory Test Directory on the ARUP Web site at www.aruplab.com.

| Test Name and Number | Recommended Use | Limitations | Follow Up |
|--|---|-------------|-----------|
| Narcolepsy - HLA DNA Panel 0095835 Method: Polymerase Chain Reaction | Identify <i>HLA</i> type associated with narcolepsy | | |

Guidelines

EFNS guidelines on management of narcolepsy. European Federation of Neurological Societies - Medical Specialty Society. 2006 October.

Practice parameters for clinical use of the multiple sleep latency test and the maintenance of wakefulness test. American Academy of Sleep Medicine - Professional Association. 2005 January 1.

General References

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