

Narcolepsy Need-to-Know Guide

How is narcolepsy diagnosed?

People with symptoms of narcolepsy often do not receive a definite diagnosis of the condition until several years after their symptoms first develop. This means that inappropriate treatment is relatively common. Some patients have to wait for over 10 years before they receive a positive diagnosis of narcolepsy. With increasing knowledge of the condition by doctors and other professionals it is hoped that more rapid diagnoses will occur in the future.

What to do first

Patients who suspect they may have narcolepsy should visit a GP in the first instance. It is useful to take a 'sleep diary' and/or a completed Epworth Sleepiness Questionnaire to the GP. The Epworth scale is an internationally accepted means of measuring daytime sleepiness, the major symptom of narcolepsy. If your score is 10 or below this indicates a level of daytime sleepiness found in the general population. A score of 18 or more indicates that you have very marked daytime sleepiness and that you should seek medical attention.

Referral by your GP

If a GP suspects that you may have narcolepsy he will most likely refer you to a specialised Sleep Centre. Once there, a careful history will be taken to determine the types of symptoms you are experiencing, the family history, the age when the first symptoms were noticed etc.

At the Sleep Centre, a number of diagnostic tests may be carried out:

Polysomnography

Unless diagnosis is clear from presenting symptoms such as cataplexy, you will probably undergo polysomnographic testing. This may involve an overnight stay in hospital or you may be sent home with a recording device and with fitted to your head. These tests involve a measurement of the electrical activity of the brain, eye and muscle movement and breathing as you fall asleep and while you are asleep.

Multiple Sleep Latency Test (MSLT)

MSLT is another very widely used diagnostic test for narcolepsy. This test measures how rapidly you fall asleep - this is called 'sleep latency'. Several such tests are usually carried out ("Multiple" Sleep Latency). People with narcolepsy usually fall asleep rapidly – they have low sleep latency. The type of sleep they enter into from consciousness will also be recorded. Evidence that they pass directly into REM sleep is regarded as a positive indication of narcolepsy. Such episodes are called 'Sleep Onset Rapid Eye Movement Periods'.

Sampling of cerebrospinal fluid

Because narcolepsy is believed to be caused by the failure of the brain to produce a molecule called hypocretin (or orexin), a reduced level of that molecule in the cerebrospinal

fluid is a strong indicator of narcolepsy. Unfortunately, however, the measurement involves a lumbar puncture, which is painful for the patient, and so this test is rarely carried out.

Genetic testing

Another relatively rare test involves taking a blood sample and carrying out a genetic test. This can show whether you have a particular type of genetic makeup that is common amongst people who develop narcolepsy. A positive result does not mean that you have narcolepsy, or that you will develop narcolepsy, but it may play a part in your doctor's assessment. Conversely, a negative result would indicate that your symptoms are probably not due to narcolepsy.

Cataplexy

The occurrence of cataplexy is almost always indicative of narcolepsy. If you suffer from cataplexy, that is very strong evidence that you have narcolepsy. However, further tests such as polysomnography and MSLT should be carried out to confirm the diagnosis. Not all patients with narcolepsy have cataplexy, however; if you do not experience cataplexy, you may still have narcolepsy.

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