

NEMALINE MYOPATHY

What is nemaline myopathy?

Nemaline myopathy (NM) is a group of rare, inherited conditions that affect muscle tone and strength. It is also known as rod body disease because at a microscopic level, abnormal rod-shaped bodies (nemalines) can be seen in affected muscle tissue. At various stages in life, the muscles of the shoulders, upper arms, pelvis and thighs may be affected. Symptoms usually start anywhere from birth to early childhood. In rare cases, it is diagnosed during adulthood. NM affects an estimated 1 in 50,000 people -- both males and females.

What are the symptoms?

Symptoms vary depending on the age of onset of the disease and the form of NM. Symptoms may include poor muscle tone and weakness especially in the face, neck, and upper arms and legs. Young children may have a delay in walking or are unable to walk. Many people experience breathing problems

due to chest muscle weakness, feeding and swallowing problems, and speech difficulties. Often, children with the condition have an elongated face and a high arched palate.

What causes nemaline myopathy?

The condition can be caused by a mutation in one of several different genes that are responsible for making muscle protein. Most cases of nemaline myopathy are inherited, although there are sometimes sporadic cases. People with a family history may choose to undergo genetic counseling to help understand the risks of passing the gene on to their children.

What are the types of nemaline myopathy?

There are two main groups of nemaline myopathy: 'typical' and 'severe.' Typical nemaline myopathy is the most common form, presenting usually in infants with muscle weakness and floppiness. It may be slowly progressive or non progressive, and most adults are able to walk. Severe nemaline myopathy is characterized by absence of spontaneous movement or respiration at birth, and often leads to death in the first months of life. Occasionally, late-childhood or adult-onset can occur. Attempts to classify nemaline myopathy into additional subtypes have been complicated by the overlap of features between the various types.

How is nemaline myopathy diagnosed?

NM is diagnosed by assessing physical symptoms plus a muscle biopsy. With a muscle biopsy, a needle is used to extract a small sample of muscle cells. The sample is examined under a microscope for the presence of rod-like bodies within the muscle cells. However, rod-bodies are present in certain other unrelated diseases, which is why taking physical symptoms into account is important too.

What treatments are available for nemaline myopathy?

There is no cure for nemaline myopathy. Treatments focus on managing the symptoms.

Breathing problems. Many people with NM have breathing problems due to weak breathing muscles, and should have respiratory function assessed frequently. For some people, use of a continuous positive air pressure (C-PAP) machine at night is an option. People using a C-PAP machine wear a small face mask at night which helps improve breathing. The device (which uses only room air) allows for carbon dioxide to be exhaled more effectively. This option can be discussed with a physician.

Chest infections. These are common in individuals with NM, especially in those with significant breathing problems. Setting up a plan for getting antibiotic therapy when chest infections are suspected should be discussed with a physician.

Weakness and lack of mobility. Physiotherapists teach people specific stretches and exercises which help maintain function and mobility, as well as breathing. Learning and doing appropriate exercises can also slow the onset of scoliosis (curvature of the spine). It is best to see a physiotherapist who has experience working with people who have neuromuscular disorders.

Difficulty swallowing and eating. Many people with NM have difficulty swallowing. A gastrostomy tube (G-Tube) bypasses the need for swallowing and can ensure good nutrition is attained. With a full assessment by a swallowing specialist, a person may be allowed to have both a G-Tube and be able to take some nutrients by mouth as well.

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