

Nemaline myopathy is a rare disorder that affects males and females. The incidence is unknown although two studies (one in Finland and one in an American Ashkenazi Jewish population) estimated the incidence to be 1 in 50,000 live births. An incidence of 1/500 has been reported in the Amish community. A diagnosis of nemaline myopathy is suspected based upon a thorough clinical evaluation, a detailed patient and family history and identification of characteristic findings. A diagnosis of nemaline myopathy is suspected based upon a thorough clinical evaluation, a detailed patient and family history and identification of characteristic findings. A diagnosis may be confirmed by the presence of thread- or rod-like structures (nemaline bodies) on muscle biopsy when stained with Gomori trichrome. A biopsy is the surgical removal and microscopic evaluation of affected tissue. Increasingly the diagnosis is made or confirmed by molecular genetic testing for mutations in the genes known to cause nemaline myopathy.