Giant axonal neuropathy is a rare neuropathy that severely affects the peripheral as well as the central nervous system. The first symptoms appear in early childhood. This disorder is characterized by abnormalities in the peripheral and central nervous systems including low muscle tone (hypotonia), muscle weakness, decreased reflexes, impaired muscle coordination (ataxia), seizures and intellectual disability. Pale, tightly curled hair is frequently seen in those affected. Giant axonal neuropathy follows autosomal recessive genetic inheritance. Symptoms of giant axonal neuropathy occur in early childhood before the age of seven years. Both the central and peripheral nervous systems are affected. The central nervous system includes the brain and spinal cord and the peripheral nervous system spreads out from the brain and spinal cord to all other areas of the body. Characteristics include low muscle tone (hypotonia), muscle weakness, decreased reflexes, impaired muscle coordination (ataxia), seizures and intellectual disability. In contrast to purely peripheral neuropathies, the reflex of the toes known as Babinski’s sign is often positive, indicating involvement of central motor pathways. Most affected children have pale, tightly curled hair unlike their parent’s hair. Cranial nerves may also be affected leading to facial weakness, abnormal eyes and poor vision. An unusual leg posture is present in some affected children. Skeletal abnormalities such as scoliosis and foot deformities have been described and are thought to be a result of the nervous system dysfunction. Mental development is in most cases initially normal, but later in childhood degenerative mental changes (dementia) may occur as the disorder progresses. Giant axonal neuropathy is rapidly progressive, usually leading to dependence on a wheel chair by the second decade of life and death in the second or third decade. Giant axonal neuropathy is a rare disorder that presents in early childhood. This disorder affects equal numbers of males and females. The prevalence of giant axonal neuropathy and the frequency of carriers of one defective copy of the GAN gene is not known, but it is known that giant axonal neuropathy is a very rare disease. Diagnosis of giant axonal neuropathy is made by clinical findings and specialized tests including nerve conduction velocity, brain MRI and peripheral nerve biopsy. The hallmark finding on a peripheral nerve biopsy is the appearance of "giant axons" which are caused by the accumulation of neurofilaments. Molecular genetic testing for abnormalities in the GAN gene is available to confirm the diagnosis. Negative mutation screening of the region of the GAN gene which encodes the protein does not exclude the diagnosis of giant axonal neuropathy.