

Congenital hypomyelination neuropathy (CHN) is a neurological disorder present at birth. Major symptoms may include respiratory difficulty, muscle weakness and incoordination, poor muscle tone (neonatal hypotonia), absence of reflexes (areflexia), difficulty in walking (ataxia), and/or impaired abilities to feel or move part of the body. Symptoms of congenital hypomyelination neuropathy and the severity of these symptoms vary from patient to patient. Major symptoms can include delayed motor (muscle) development (ability to turn over, stand, crawl, walk, etc.), muscle weakness, poor muscle tone (hypotonia), impaired muscle coordination, absence of reflexes (areflexia), difficulty in walking or crawling, and/or impaired ability to feel or move part of the body (mild distal palsy). In some infants, respiratory problems or difficulty in swallowing may occur. Abnormal microscopic changes in certain nerves such as sural nerves (located in the calf of the leg) can occur. Congenital hypomyelination neuropathy is a rare disorder present at birth. It affects males and females in equal numbers. Testing for congenital hypomyelination neuropathy includes measuring the velocity of transmission of electrical impulses produced by the muscles (electromyogram), as well as nerve and/or muscle biopsies.