Congenital Myopathy

National Institute of Neurological Disorders and Stroke (NINDS)

Source

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A myopathy is a disorder of the muscles that usually results in weakness. Congenital myopathy refers to a group of muscle disorders that appear at birth or in infancy. Typically, an infant with a congenital myopathy will be "floppy," have difficulty breathing or feeding, and will lag behind other babies in meeting normal developmental milestones such as turning over or sitting up.

Muscle weakness can occur for many reasons, including a problem with the muscle, a problem with the nerve that stimulates the muscle, or a problem with the brain. Therefore, to diagnose a congenital myopathy, a neurologist will perform a detailed physical exam as well as tests to determine the cause of weakness. If a myopathy is suspected, possible tests include a blood test for a muscle enzyme called creatine kinase, an electromyogram (EMG) to evaluate the electrical activity of the muscle, a muscle biopsy, and genetic testing.

There are currently seven distinct types of congenital myopathy, with some variation in symptoms, complications, treatment options, and outlook.

Nemaline myopathy is the most common congenital myopathy. Infants usually have problems with breathing and feeding. Later, some skeletal problems may arise, such as scoliosis (curvature of the spine). In general, the weakness does not worsen during life.

Myotubular myopathy is rare and only affects boys. Weakness and floppiness are so severe that a mother may notice reduced movements of the baby in her womb during pregnancy. There are usually significant breathing and swallowing difficulties; many children do not survive infancy. Osteopenia (weakening of the bones) is also associated with this disorder.

Centronuclear myopathy is rare and begins in infancy or early childhood with weakness of the arms and legs, droopy eyelids, and problems with eye movements. Weakness often gets worse with time.

Central core disease varies among children with regard to the severity of problems and the degree of worsening over time. Usually, there is mild floppiness in infancy, delayed milestones, and moderate limb weakness, which do not worsen much over time. Children with central core disease may have life-threatening reactions to general anesthesia. Treatment with the drug salbutamol has been shown to reduce weakness significantly, although it does not cure the disorder.

Multi-minicore disease has several different subtypes. Common to most is severe weakness of the limbs and scoliosis. Often breathing difficulties occur as well. Some children have weakened eye movements.

Congenital fiber-type disproportion myopathy is a rare disorder that begins with floppiness, limb and facial weakness, and breathing problems.

Hyaline body myopathy is a disorder characterized by the specific appearance under the microscope of a sample of muscle tissue. It probably includes several different causes. Because of this, the symptoms are quite variable.