Congenital Myopathies

Congenital myopathy is a term for any muscle disorder present at birth. Congenital myopathies could include hundreds of distinct syndromes and disorders. Congenital myopathies cause loss of muscle tone and muscle weakness in infancy. They can delay walking.

Three disorders are classified as congenital myopathies:

1. **Central core disease** is a mainly inherited genetic disease characterized by mild leg weakness. It appears in infancy. This weakness does not progress with age but leads to delay in walking.
2. **Nemaline rod myopathy** is a genetic disease characterized by:

- Infantile muscle weakness.
- Loss of muscle tone.

It may be accompanied by problems in:

- Suckling or feeding.
- Delay in walking.
- Breathing.

Other signs may surface with maturity, such as:

- Reduction of muscle bulk.
- Weakness of the trunk and limbs.
- Development of a long and abnormally shaped face with a protruding jaw.
- Muscle weakness and wasting may progress slowly throughout life.

3. **Centronuclear (myotubular) myopathy** is an inherited genetic disorder characterized by:

- Muscle weakness.
- Loss of muscle tone present at birth.

It also may slow development of muscle weakness later in infancy. Weakness gets gradually worse and can become severe. Infants may have paralysis of the eye muscles (*ophthalmoplegia*).

TREATMENT

Treatment involves helping patients cope with the problems of congenital myopathies.

WHAT WILL HAPPEN

Patients with central core disease generally have a normal life expectancy with non-progressive muscle weakness. Patients with nemaline rod myopathy may have a normal life expectancy with progressive weakness or may die early in infancy from respiratory distress depending upon the severity of the disorder. Patients with centronuclear (myotubular) myopathy generally have a poor prognosis and suffer an early death, usually in infancy.

FOR MORE INFORMATION:

Contact The National Institute of Neurological Disorders and Stroke

Document Released: 12/08/2003 Document Re-Released: 03/14/2011
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