Congenital Myasthenia

Congenital myasthenia is a group of disorders that affect the transmission of signals from the nerves to the muscles. These disorders are present at birth (*congenital*).

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CAUSES

There are two different kinds of causes:

- **Genetic.** The genetic type involves a defect in 1 of the more than 30 molecules associated with neuromuscular transmission. Depending on which molecules are involved, this form may or may not be treatable.
- **Autoimmune.** There are several different autoimmune forms. These forms may either be temporary or permanent.

SYMPTOMS

Symptoms are usually noticed in early childhood and include:

- Drooping eyelids.
- Facial weakness.
- Difficulty chewing.
- Limb weakness.

At the time of birth, an infant may have severe generalized weakness. The weakness may be so severe that the child cannot breathe. Parents of children with congenital myasthenia frequently show no symptoms of the disorder.

DIAGNOSIS

Diagnosis usually involves the following initial tests:

- Tests to study the electrical activity of muscles while they are working and while they are at rest (*electromyography*, *EMG*).
- Nerve conduction tests to study how well and how fast nerves work.

If the initial tests show that there may be a problem, further blood work will be done to determine if the patient has a **genetic** form or an **autoimmune** form.

TREATMENT

- Cholinesterase inhibitor medicines may improve the symptoms of congenital myasthenia by affecting nerves and muscles. Other medicines may improve symptoms by altering the immune system.
- Some forms of the autoimmune type may benefit from the removal of the thymus gland which produces the antibodies that are causing the weakness.

PROGNOSIS

Patients with the genetic form have the symptoms for life. Prognosis for the autoimmune forms are generally much better; many children have no symptoms as they get older.

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