Recognize these symptoms? Ask your healthcare provider today about a rare disorder called Congenital Myasthenic Syndromes (CMS).
**What are Congenital Myasthenic Syndromes (CMS)?**

CMS is a group of rare inherited disorders that cause muscle weakness and other symptoms. It is caused by genes and is not an autoimmune condition. About 24 different genes cause CMS, and they are grouped into 3 distinct categories:

- Presynaptic: Affects certain nerve cells
- Postsynaptic: Affects cells in the muscles
- Synaptic: Affects the space between the nerve and muscle cells

All of these categories of CMS affect the neuromuscular junction, which is where the muscle cell meets the nerve cell. CMS interferes with the nerve impulses to the muscle by decreasing the amount of a substance called acetylcholine (ACh). The result is that the signal that tells the muscle to contract does not work the way it should.

**Who is affected by CMS?**

People who are affected are born with this genetic disorder, and they have it for a lifetime. CMS symptoms typically begin at birth or infancy, but sometimes appear later in life. The fact that CMS is causing these symptoms may not be so obvious. And CMS is very rare, only affecting 3 people out of a million, so the healthcare provider may not immediately identify the disorder.

“We are looking forward to future treatment advances for people with CMS.”

- Investigator
What are the symptoms of CMS?

In infants, symptoms can include weakness, tiredness, and droopy eyelids. Children and adults with CMS often have muscle weakness and/or muscles that are easily fatigued. They may have difficulty rising from a chair, climbing stairs, or walking.

Symptoms can depend on affected genes and the category of CMS.

**Symptoms may include:**
- Droopy eyelids
- Difficulty talking, chewing, swallowing, and holding up the head
- Feeding difficulties
- Respiratory problems
- Muscle weakness
- Easy fatigability
- Worsening weakness upon exertion

**In infants, symptoms may include:**
- Severe muscle weakness
- Breathing problems
- Delayed motor milestones (sitting, crawling, and walking)

Can CMS be difficult to diagnose?\(^{1-3,5}\)

Because of their similarities, CMS may be confused with other diseases like
- Various forms of myopathy
- Muscular dystrophy
- Myasthenia gravis

Symptoms: Weakness, loss of muscle tone, feeling tired, droopy eyelids\(^{1-3}\)
How is CMS diagnosed?

It is very important to tell your Healthcare Provider if you or your loved one has symptoms of CMS. Your Healthcare Provider will likely refer you to a neurologist who specializes in neuromuscular disorders.

What can I expect at the specialist’s visit?

- A review of medical history
- A physical examination to help check the degree of muscle weakness
- Genetic testing to identify the affected gene
- Electromyography (EMG) may be used to test the electrical activity of skeletal muscles
- Tests of the neuromuscular junction identify overactive versus underactive areas, helping identify the category of CMS

CMS is caused by defects in at least 24 genes

"When I couldn't keep up with my classmates, people thought it was laziness."

-CMS Patient

“We knew something was wrong with Jacob right after he was born. It is comforting to put a name to his condition—and to have hope.”

-Parent
How is CMS treated?

Treatment of CMS depends on the defective gene that is responsible for the disease. Identifying the gene is so important because therapy that can help a person with one category of CMS might be harmful for a person with another category.

While there is no FDA approved treatment for CMS, promising research is ongoing. The good news is that an investigational drug that helps improve the electric impulse between nerve and muscle cells in people with certain gene mutations may be available in the near future.

What can I expect over time?

What to expect depends on several things, including the category of CMS and the age at which the disorder was diagnosed. Other factors include how the respiratory system is working and how well a person can walk and swallow. CMS symptoms can change for a person at various times of life. What's severe in the newborn may improve in childhood or adolescence.

“Jenna won’t let CMS defeat her. She’s my inspiration.”
- Parent
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