CONGENITAL MYASTHENIC SYNDROMES

MYASTHENIA GRAVIS FOUNDATION OF AMERICA

Common questions people ask about congenital myasthenic syndromes.

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What are congenital myasthenic syndromes?

Congenital myasthenic syndromes (CMS) is the term used for a group of uncommon hereditary disorders of the neuromuscular junction as distinguished from autoimmune myasthenia gravis (MG). There are several different subtypes of CMS, each the result of a specific genetic mutation. The major differences between these syndromes and the more common autoimmune MG include: 1) CMS, as the name implies, usually manifest early in life, often in infancy with variable degrees of fluctuating weakness. 2) CMS is not associated with antibodies against any one of the components of the neuromuscular junction. 3) All of the CMS disorders result from mutations that alter one of the components of the neuromuscular junction. Patients with a CMS disorder tend to have lifelong or relatively stable symptoms of generalized fatigable weakness. These disorders are nonimmunologic in nature and patients do not have acetylcholine receptor antibodies; therefore, patients do not typically respond to immune therapy often used in patients with autoimmune MG (steroids, thymectomy, plasma exchange). Most patients with CMS develop symptoms in infancy or childhood with variable degrees of fluctuating weakness.

Are there different types of congenital myasthenic syndromes?

Yes. Not all forms of congenital myasthenia are the same. A number of different types of congenital myasthenia have been identified with a variety of different structural and functional abnormalities of the neuromuscular junction. Patterns of inheritance, clinical symptoms, electrophysiology, and response to therapy vary depending on the type. Some of the subtypes that one may encounter include “familial infantile myasthenia,” a “congenital absence of acetylcholinesterase” presenting in infancy or childhood with generalized weakness and reduced muscle tone, “the slow channel syndrome,” which often follows an autosomal dominant pattern of inheritance with a variable age of onset and severity of symptoms, and a collection of disorders characterized by defective acetylcholine receptors.

Is there any reason to try to determine the exact type of congenital myasthenic syndrome?

A thorough diagnostic evaluation is worthwhile in patients with suspected congenital myasthenia because of the different types, and somewhat different treatment options. Patients with some subtypes may respond best to Mestinon® (pyridostigmine), while patients with other subtypes may respond best to other therapies (some types respond to ephedrine, some to 3, 4 DAP, quinidine or fluoxetine, as well as a variety of other drugs depending on the type of congenital myasthenia).

In general, what is the long-term prognosis for patients with congenital myasthenic syndromes?

Most patients remain fairly stable throughout their lifetime and tend not to have wide fluctuations of symptoms or function nor

myasthenic crises. Overall, patients tend to stay about the same on a long-term basis.

What is the difference between congenital myasthenia and transient neonatal myasthenia?

Transient neonatal myasthenia occurs in 10-15% of babies born to mothers with autoimmune myasthenia gravis. Within the first few days after delivery, the infant has a weak cry or suck, appears generally weak and, on occasion, requires mechanical ventilation. Maternal antibodies that cross the placenta late in pregnancy cause transient neonatal myasthenia. As these maternal antibodies are replaced by the infant's own antibodies, the symptoms gradually disappear, usually within a few weeks, and the baby is normal thereafter. Infants with severe weakness from transient neonatal myasthenia may be treated with oral pyridostigmine and whatever degree of general support (mechanical respiratory ventilation, for example) is necessary until the condition clears. Infants with transient neonatal myasthenia gravis do not have an increased risk for the long-term or future development of myasthenia gravis.

Should patients with congenital myasthenic syndromes avoid the same medications that may aggravate autoimmune myasthenia gravis?

Yes. It is advisable to be cautious when starting newly prescribed or even some over-the-counter medications. Patients should check with their myasthenia physician prior to taking any new medications.

Your Notes

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Our Vision: A World Without MG

Our Mission: Create Connections, Enhance Lives, Improve Care, Cure MG

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