What is congenital myasthenia?

Congenital myasthenia is the term used for a group of uncommon hereditary disorders of the neuromuscular junction. Patients with congenital myasthenia tend to have lifelong or relatively stable symptoms of generalized fatigable weakness. These disorders are non-immunologic 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 myasthenia gravis (steroids, thymectomy, plasma exchange). Most patients with congenital myasthenia develop symptoms in infancy or childhood with variable degrees of fluctuating weakness.

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The MGFA mission is to facilitate the timely diagnosis and optimal care of individuals affected by myasthenia gravis and closely related disorders and to improve their lives through programs of patient services, public information, medical research, professional education, advocacy and patient care.

Approved by the MGFA Medical/Scientific and Nurses Advisory Boards

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CONGENITAL MYASTHENIA

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Common questions people ask about congenital myasthenia.

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Are there different types of congenital myasthenia?

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 myasthenia?

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, 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 myasthenia?

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 the condition. 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 myasthenia 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.

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