

Cranial-Manual-Cervical Dystonia in Three Unrelated Women Associated with Acetylcholine Receptor Antibodies: ? A New Syndrome

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Dystonia is a phenomenon of presumptive central nervous system origin which, when idiopathic or primary, etiology is apt to be genetic. When focal or segmental, mean onset age is mid-adult, females outnumber males, loci by frequency of involvement: Cervical, lower cranial and/or manual, and increased rate of relatives with dystonia, tremor or scoliosis (Duane, Adv. Neur., 1998). The rate of elevated serum titers of antinuclear antibody (ANA) is increased in patients with cervical dystonia (Duane et al, Neurology, 1995). Botulinum toxin is an effective form of temporary ameliorative therapy for focal dystonia symptoms. The presence of clinical myasthenia gravis, an autoimmune disorder of neuromuscular transmission, is considered a contraindication to botulinum toxin therapy because of assumed supersensitivity to neuromuscular blockade in such patients.

We report here three adult women, all with onset of dystonic symptoms in late childhood, all who have clinically significant lower cranial dystonia, producing hyperkinetic dysarthria, dystonic hand manifestations, and mild cervical dystonia, two with elevated serum ANA titers, the third with elevated serum IgG levels, all three with elevated acetylcholine receptor antibody titers (AChR Ab) (Mayo Medical Labs). All three had EMG, one including single fiber studies which demonstrated increased jitter (J. Clarke Stevens, M.D.). Therapy elsewhere of one of these patients for myasthenia gravis (steroids and plasmapheresis) had no effect on the dystonic symptoms. That patient in our hands, is not unusually sensitive to botulinum toxin A. (Duane et al, Movement Disorders, in press). The other two are stable with oral anticholinergic medication. Two of these patients have first-degree relatives with possible cranial dystonia, the third an uncle with essential tremor. None possess the Dyt 1 gene marker. None are Sinemet sensitive.

The occurrence of AChR antibodies may represent: Chance, a co-morbid risk for antibodies other than ANA, or a genetic marker. Antibody screening of other dystonia patients and additional genetic testing in familial cases may clarify this relationship.

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