

Combined Benign Essential Blepharospasm and Apraxia of Eyelid Opening - The New Missed Diagnosis

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In the 16th Century, Flemish artist, Brueghel painted "De Gaper" which depicted a subject with markedly distorted facial features and eyelid spasms. This is generally considered to be the first recorded case of benign essential blepharospasm and is, in fact, what we would now sub-categorize as Meige syndrome. From that time until the mid 1980's, benign essential blepharospasm (BEB) and Meige syndrome was a relatively unknown facial disease rarely diagnosed correctly by physicians and even less frequently treated appropriately.

In 1981, the beloved founder of Benign Essential Blepharospasm Research Foundation (BEBRF), Mattie Lou Koster, who had just been diagnosed with BEB, decided that she would bring the disease out into the open and make the diagnosis and its signs and symptoms as familiar to the medical community as the common cold. All neurologists, ophthalmologists, and neurosurgeons would certainly concur that she was immensely successful.

In 1990, Dr. Richard Anderson with the support of the BEBRF mailed 4,000 questionnaires to patients who registered prior to 1988 with the Benign Essential Blepharospasm Research Foundation, of which 1,653 were returned. One of the questions was, "How many physicians did you see prior to the establishment of a correct diagnosis?" At that time, the average response was four physicians with the range between 1 and 75. The study noted the cost of misdiagnosis, unnecessary studies, and unsuccessful therapy was staggering and in one percent of patients exceeded \$50,000. It is my personal experience that in the past few years virtually all patients referred to my office for treatment of benign essential blepharospasm and Meige syndrome have been correctly diagnosed by either the first or second doctor consulted for their symptoms. Furthermore, most patients are seen within six months of the onset of their symptoms as opposed to my experience in the late 1980's when most patients had had their disease for many years and were in the advanced stages of the syndrome. It is apparent that the past two decades have seen a marked improvement in the efficiency of diagnosing and managing BEB and Meige syndrome.

Conversely, blepharospasm combined with apraxia of lid opening is in my experience rarely initially diagnosed by the examining physician. Moreover there is a failure to diagnose by those physicians specialized in the management of this condition, including ophthalmologists and neurologists. Apraxia of lid opening was first described by Goldstein and Cogan in 1965 as "a non-paralytic motor abnormality characterized by the patient's difficulty in initiating the act of lid elevation." Apraxia is generally defined as the inability to carry out purposeful movements in the absence of motor paralysis,

sensory loss, or ataxia. To paraphrase the definition of apraxia of lid opening, it is a patient who reports, "I can't will my eyes to open."

Just as blepharospasm and Meige syndrome were only gradually recognized to be relatively common diseases, combined benign essential blepharospasm and apraxia of lid opening has also been statistically found to have a relatively common association with blepharospasm.

In 1990, Drs. David R. Jordan, Rachel Anderson, and Katherine B. Digre, evaluated patients who were referred to them with the diagnosis of benign essential blepharospasm or Meige syndrome. As they followed these patients from diagnosis, initial treatment, and in some cases surgical management, it became apparent that 7 patients (7%) of this population actually had apraxia of lid opening in addition to their referring diagnosis. Only 5 of the 7 patients were correctly diagnosed at the time of the initial visit despite these physicians' sophisticated attempts to diagnose the mixed condition. They also reported that other patients in the group of 100 were thought to have apraxia of lid opening, but later to be falsely diagnosed when they were fully controlled with botulinum toxin.

Between 1988 and 1992, Dr. P. Krack and M.H. Marion in the Department of Neurology, University of Giessen, Germany, evaluated 195 patients with benign essential blepharospasm or Meige syndrome and of that number, 20 were later diagnosed as also having apraxia of lid opening. These twenty patients represent a comparable 10% incidence of combined disease among their blepharospasm patients.

In the classical presentation, benign essential blepharospasm occurs in a female with an average age of 55 who initially notices increased blinking and sensitivity to light. This is followed by increasingly frequent intermittent forceful eyelid closure caused by spasm of the orbicularis muscles. The spasms of the corrugator superciliaris muscle and procerus muscles draw the brows medially and inferiorly from their normal position. In Meige syndrome, the patient would have also developed spasms of the mid and lower face. Both of these syndromes are frequently associated with varying degrees of dry eye syndrome.

Apraxia of lid opening can occur in association with blepharospasm, as well as other neurologic diseases. In the "pure" form, the patients experience sustained periods of time when they are unable to will their eyes open. During these episodes, the eyes are fully closed. In contrast to benign essential blepharospasm and Meige syndrome however, there is no evidence of orbicularis spasm and the brows are markedly elevated by the frontalis muscles, as would be expected in someone straining to open their eyes. In the fully developed clinical picture of combined blepharospasm and apraxia of lid opening, the patient presents with a combined pattern beginning with sustained spasms of the procerus and corrugator muscles lowering the brow as well as the orbicularis muscles squeezing the eyes shut. The patient eventually has relaxation of those muscles resulting in an elevation of the brows, but still lacks the ability to open

the eyes. This sequence of events is difficult to observe unless the physician has a very high index of suspicion.

The diagnosis of apraxia of lid opening is established by the use of the most effective treatment for benign essential blepharospasm - botulinum toxin. The patient should initially be treated utilizing the protocol each physician has found most successful for their BEB patients. Most patients return in three months for a reevaluation and adjustment of therapy. The typical patient with mixed disease however will return reporting that they have not appreciated improvement from the botulinum toxin treatment. In this situation, re-treatment is indicated followed by an evaluation two to four weeks later, at which time the maximal botulinum toxin effect should be apparent. Adequate paresis is confirmed when the patient demonstrates an inability to close the upper and lower lids against the resistance of the physician's fingers. In the patients with apraxia of lid opening, there are periods of persistent involuntary lid closure despite adequate botulinum toxin treatment of the orbicularis and brow depressor muscles. The patient will usually demonstrate brow elevation in a futile attempt to open the lids. Patients often report that gently elevating the lids open with their fingers or touching the skin on their temple can sometimes break the attacks. When the episode resolves, there is no evidence of ptosis or levator muscle weakness.

It has been uniformly observed by physicians with an interest in combined blepharospasm and apraxia that traditional blepharospasm treatments are ineffective in improving the apraxia of lid opening. Specifically, Botox, adjunct oral medications, and both limited and complete myectomies do not improve apraxia of lid opening. Apraxia of lid opening, however, can be successfully managed after control of the patient's blepharospasm with the surgical procedure of frontalis suspension. This is a commonly performed and successful eyelid elevating operation in appropriately chosen patients. In patients with mixed disease, it can be combined with a limited myectomy procedure that removes orbicularis muscle from the lids to provide improved control of both conditions.

In conclusion, mixed blepharospasm and apraxia of lid opening are a relatively common cause of the inability to maintain open and functional eyelids and is probably present in at least 7% of patients with blepharospasm. It is also likely the reason for a significant number of benign essential blepharospasm treatment failures. Observation of a patient's eyebrows throughout the complete cycle of eyelid closure, attempted reopening, and then full normal opening, are the key to early diagnosis and appropriate treatment.

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