GENETICS OF BLEPHAROSPASM

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In order to truly understand blepharospasm and find the best treatment, it will be necessary to understand its cause. The cause of a disease is often not a single element but may be multiple elements. Moreover, the cause may differ in different people. Medicine is replete with examples of a single disorder resulting from several different etiologies.

It seems very likely now that one of the major etiological factors in producing blepharospasm is genetic. This means that a person's genetic makeup will be important in whether he or she develops blepharospasm or not. In some circumstances the genetic factor may be so powerful that it can produce blepharospasm by itself. In other circumstances it may provide a background on which another factor triggers and produces the blepharospasm.

What is the evidence that there is a genetic factor in blepharospasm? First of all, it has to be accepted that blepharospasm is a form of focal dystonia. Dystonia is a group of disorders characterized by involuntary muscle spasms. Common focal dystonias in addition to blepharospasm include: oral mandibular dystonia, spasmodic dysphonia, spasmodic torticollis, and hand cramps. We know that these focal dystonias are related to each other since many of them are seen in patients who have generalized dystonia or are early manifestations of generalized dystonia. Additionally, patients sometimes can have several different focal dystonias. Lastly, different family members can be affected with different types of focal dystonias. Already it is established that several types of generalized dystonia are produced genetically.

There is beginning to be strong evidence now that focal dystonias may also be caused genetically. The evidence comes from several fairly large epidemiological studies where families of patients with focal dystonia were examined. In these families, a number of other family members were found who had similar or different focal dystonias. For example, a patient with blepharospasm might well have another relative with blepharospasm or a relative with a different focal dystonia such as spasmodic torticollis. The frequency of other family members having focal dystonias is only about 5%, but this frequency is clearly more than what is seen in the general population and hence implies a genetic etiology.

The implication of this epidemiological information is that there is only a 1 in 20 chance of any relative of a patient with blepharospasm to also have a focal dystonia. This relates to children of patients with blepharospasm, for example. Hence, should you have blepharospasm, there is only a 1 in 20 chance for each of your children that they will have a focal dystonia like blepharospasm.

The possibility of finding a gene that causes blepharospasm is very exciting. Once a gene is found, this should lead to a much clearer understanding about blepharospasm. Such work is likely to take many years, but it should be a good start in the right direction. We are all hopeful that finding a gene relevant to blepharospasm will be a gateway to finding the final cure of this disorder.