What is Dystonia?

Dystonia Educational Series

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Dystonia can begin at any age; in childhood, during early adulthood, or even in one’s senior years. Age of onset is an important feature in helping to determine the potential prognosis as well as the underlying cause of dystonia. Patients with childhood onset more commonly have a genetic cause of dystonia and it is more likely that their dystonia may gradually spread from one part of the body to other parts of the body and may become generalized and more severe. Patients with onset in middle to late years of life usually develop focal dystonia (such as affecting only the neck or the face) and although the dystonia may spread to some adjacent body parts such as from the neck to the face, spread to involve the limbs or entire body is very uncommon.
There have been a large number of genes that have been found to cause various forms of familial dystonia. In the majority of cases, the dystonia tends to have onset in childhood or early adulthood. Typically many members of these families develop generalized dystonia. More recently, two or three genes have been discovered to cause focal or segmental cervical or craniocervical segmental dystonia. Most of the dystonia genes exhibit what is called reduced penetrance. This means that even if one harbors the genetic mutation, one may never develop symptoms. Indeed, in general only about 30 to 40 percent of individuals who have mutations in the various dystonia genes ever develop symptoms of dystonia during their life.
A minority of patients have acquired dystonia as a result of non-genetic causes. Dystonia may occur as a result of brain injury occurring from head trauma or as a result of oxygen deprivation during birth. Other non-traumatic causes such as brain tumors, strokes, or brain infection may also cause dystonia. Dopamine receptor blocking drugs such as those used to treat nausea and vomiting and others used to treat schizophrenia or depression may cause what is termed tardive dystonia. Lastly, dystonia may occur as part of a more widespread brain disorder in certain brain degenerative or metabolic diseases.
Idiopathic means that we have not yet discovered the cause. It is thought that most cases of idiopathic dystonia are, in fact, due to a genetic cause, but we have not yet discovered the specific gene mutations responsible for the patient’s dystonia. At present, the vast majority of patients have focal dystonia and specific genes are just now beginning to be discovered associated with certain forms of cervical dystonia. It is also possible that a small contribution of many genes may also be responsible for the development of dystonia in some patients, so-called polygenetic inheritance.
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