Dystonias

The dystonias are movement disorders in which sustained muscle contractions cause twisting and repetitive movements or abnormal postures. The movements, which are involuntary and sometimes painful, may affect a single muscle; a group of muscles such as those in the arms, legs, or neck; or the entire body. Early *symptoms* (problems) may include a deterioration in handwriting after writing several lines, foot cramps, and a tendency of one foot to pull up or drag after running or walking some distance. Other possible symptoms are tremor and voice or speech difficulties. Birth injury (particularly due to lack of oxygen), certain infections, reactions to certain drugs, heavy-metal or carbon monoxide poisoning, *trauma* (damage caused by an accident), or stroke can cause dystonic symptoms. About half the cases of dystonia have no connection to disease or injury and are called primary or idiopathic dystonia. Of the primary dystonias, many cases appear to be inherited in a dominant manner. Dystonias can also be symptoms of other diseases, some of which may be hereditary (passed down from parents). In some individuals, symptoms of a dystonia appear spontaneously in childhood between the ages of 5 and 16, usually in the foot or in the hand. For other individuals, the symptoms emerge in late adolescence or early adulthood.

TREATMENT

No one treatment has been found universally effective for dystonia. Instead, physicians use a variety of *therapies* (medications, surgery and other treatments such as physical therapy, splinting, stress management, and biofeedback), aimed at reducing or eliminating muscle spasms and pain. Since response to drugs varies among patients and even in the same person over time, the therapy must be individualized.

WHAT WILL HAPPEN

The initial symptoms can be very mild and may be noticeable only after prolonged exertion, stress, or fatigue. Over a period of time, the symptoms may become more noticeable and widespread and be unrelenting; sometimes, however, there is little or no progression.

RESEARCH BEING DONE

Investigators believe that the dystonias result from an abnormality in an area of the brain called the basal ganglia, where some of the messages that initiate muscle contractions are processed. Scientists suspect a defect in the body's ability to process a group of chemicals called neurotransmitters that help cells in the brain communicate with each other. Scientists at the NINDS laboratories have conducted detailed investigations of the pattern of muscle activity in persons with dystonias. Studies using EEG analysis and neuroimaging are probing brain activity. The search for the gene or genes responsible for some forms of dominantly inherited dystonias continues. In 1989, a team of researchers mapped a gene for early-onset torsion dystonia to chromosome 9; the gene was subsequently named DYT1. In 1997, the team sequenced the DYT1 gene and found that it codes for a previously unknown protein now called "torsin A."

Research is continually being done to learn more about this problem.

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