# National Institutes of Health

## *Fact Sheet* — *Dystonia*

### Thirty Years Ago

- Dystonia was a seriously debilitating condition, affecting thousands of Americans with disabling involuntary muscle contractions that often produce twisting, abnormal postures, or repetitive movements.
- Although some clinicians had speculated that dystonia could be inherited, its potential genetic causes were unknown and the cellular biology underlying the development of disease was not understood at all.
- Treatment for dystonia was mainly limited to surgeries that destroyed small regions of brain tissue or severed peripheral nerves believed to be responsible for the abnormal movements. The use of oral medications to treat dystonia was just beginning, but none were very effective.

### Today

- Today, scientists and clinicians have a much greater understanding of the causes of dystonia. Researchers identified 15 different genetic mutations that can contribute, including mutations in the gene DYT1 which cause primary torsion dystonia, a whole-body or "generalized" form of the disorder.
- Genetic testing is now available to people with dystonia and their family members to determine whether or not they have a DYT1 mutation.
- Investigators are beginning to understand how abnormal changes in the cells of the nervous system contribute to the disease. These changes include the build-up of mutant proteins around the nucleus, or control center of the cell. Animals models of dystonia – based in some cases on the manipulation of the genes linked to the disorder in humans – are making a substantial contribution to these basic science research efforts.
- Today's patients have many more treatment options thanks to advances in dystonia research. Some patients who have a specific form of dystonia may experience considerable relief by taking levodopa, a drug that the body uses to synthesize the nerve chemical dopamine. Drugs that interfere with another neurochemical, acetylcholine (which stimulates muscle movements), may also be effective.
- The use of deep brain stimulation (DBS) – the electrical stimulation of very small regions of brain tissue – is effective for some people with dystonia, and has reduced the need for the older forms of surgery that created permanent damage to the nervous system.
- Other very effective treatment options are also available, including minute injections of botulinum toxin (sold under the trade names of Botox® and Myobloc™) into dystonic muscles. This therapy has revolutionized the treatment of focal dystonias, or dystonias that are limited to a single area of the body such as the neck, eyelid, hand, or voicebox (larynx). The toxin works by blocking the release of acetylcholine in the local area of injection, and it can improve symptoms for several months.
- Clinical researchers are exploring how to expand the uses of therapies like DBS and botulinum toxin to other types of dystonia patients.

### Tomorrow

- NIH-funded investigators are making tremendous strides in understanding the molecular and cellular mechanisms of dystonia.
- These mechanisms will serve as targets for future development of therapies that stop the development of abnormal movements at their cellular points of origin, rather than interfering with downstream events to simply quell the symptoms.

- Imaging techniques are revealing abnormal activation patterns in motor and sensory brain areas. This information is critical to understanding how current therapies interact with the nerve circuits that cause abnormal movements, and how to improve the effectiveness of these treatments.
- Advances in both the preclinical development of therapies and in our understanding of the brain circuits that cause dystonia will enable clinical researchers to design and test promising therapies and determine which are both safe and effective for use in people diagnosed with dystonia in the future.