Dystonia is characterized by loss of voluntary control of movements due to excessive or exaggerated drive for movement. When these patients try to move, they suffer from excessive involuntary muscle contractions along with overflow contractions of nearby muscles not needed for the movement. The resulting abnormal movements depend on the strengths and patterns of muscles involved. In its mildest form, dystonia may appear merely as an exaggeration of an otherwise normal movement. In more serious cases, movements are twisting, stiff, and slow. In its most severe forms, dystonia is characterized by persistent involuntary abnormal posturing. Dystonia can occur alone or as a symptom of many other disorders including Parkinson disease, Huntington disease, cerebral palsy and a host of other inherited or acquired disorders of the nervous system. Although dystonia affects many people, we are just beginning to understand the neurobiological causes.

What brain regions are implicated?

Dystonia traditionally is associated with dysfunction of the basal ganglia, but recent studies have implicated abnormal cerebellar function too. These new studies have led to the concept that dystonia is a circuit disorder. Our work focuses on understanding how this circuit gets disrupted.


What neurotransmitters and cell signaling pathways are abnormal?

Some forms of dystonia are linked with dysfunction of dopaminergic pathways, while other studies have revealed dysregulation of cellular calcium homeostasis. Understanding common underlying mechanisms will provide insight into many forms of dystonia.


Interesting Dystonia Tidbits

- The most common dystonia is known as “torticollis,” which means “twisted neck” because affected individuals can’t hold their heads straight.
- Scott Adams, author of the cartoon Dilbert, had to temporarily stop his series because he developed dystonia of his drawing hand.
- Several musicians had their careers interrupted when they developed dystonia of their playing hands, for example the concert pianist Leon Fleisher.
- Academicians who write a lot are at high risk of developing writer’s cramp, a hand dystonia.
- Botox is not just for wrinkles! Its main medical use is treating overactive muscles in dystonia.

http://www.pharm.emory.edu/ehess/home.html


**What can animal models teach us about human dystonia?**

*Why are females more often affected than males? Why does stress make dystonia worse? What genes contribute to the expression of dystonia? We use mouse models to help us answer these questions, and have an interest in developing monkey models based on what we learn from mice.*


**Can we identify promising new treatments?**

Our anti-dystonia drug discovery program is aimed at identifying new treatments for dystonia, using a screening program based on animal models.


**How does dystonia affect people?**

We are conducting clinical and translational studies that include documenting natural history and co-morbidities in humans with dystonia, collecting samples for a biorepository for genetic and other biomarker studies, and developing the best designs for clinical trials.
