

Dystonia has only been recognised in recent years as a neurological disorder. The ability of neuroscientists to investigate brain activity non-invasively using techniques like Transcranial Magnetic Stimulation (TMS) has rapidly increased the understanding of the condition. However, there is still a general lack of knowledge in the medical and allied health professional domains, making diagnosis and management a difficult experience for many people. This problem is compounded by the varied clinical presentations of the disorder and the complex underlying mechanisms that may include genetic and environmental factors. Dystonia is most commonly described as a condition where people experience sustained or involuntary muscle contractions leading to twisted postures or abnormal movements. Most commonly it is classified according to the region affected. Therefore, focal Dystonia may affect just one hand or the neck or eyes, segmental Dystonia might affect a whole arm or leg or both legs, while generalized Dystonia can affect the entire body. Dystonia can also be classified according to its onset. Primary or idiopathic Dystonia usually presents in adults and may have no known underlying medical cause. The musicians or writer's cramp affecting people who perform hours of repetitive finger movements (task-specific focal Dystonia) are classic examples. Studies have shown that Dystonia has a large impact on peoples' lives, with many experiencing unremitting pain, difficulty walking or using their limbs in a normal way. In turn, these problems affect the ability to work, drive and function in everyday life. There are few treatment options and the current standard therapy for many consists of repeated injections of botulinum toxin into affected muscles.

While the causes and mechanisms contributing to Dystonia are still being understood, there are many neurophysiological studies showing it is a neurological disorder affecting the motor areas of the brain. Most research has been conducted in the area of focal hand dystonia and these studies have found the brain reacts abnormally to sensory inputs from the affected body part. The predominant ideas are there is a decrease in inhibitory mechanisms within the brain leading to a process that has been called maladaptive brain plasticity. Neuroplasticity is a term describing how the brain changes as a result of inputs such as exercise, repetitive practice, change in sensory inputs (for example, pain) and after neurological injury such as stroke. Neuroplasticity is how the brain 'repairs' its function by forming new connections between neurons to allow muscles to be used again in a functional manner. Neuroplasticity can be adaptive, i.e. the re-organization has functional benefits, or maladaptive where the reorganisation may not produce 'normal' intended movements. This is certainly the case in Dystonia, where movements or postures seem to take on 'a mind of their own' outside of voluntary control no matter how hard the person tries to overcome the dystonic movement patterns. Whether neuroplasticity occurs in an adaptive or maladaptive manner seems to depend to a large extent on the inputs to the brain. These can be considered 'drivers' of plasticity. For example, spasticity after stroke is thought to be a result of maladaptive neuroplasticity, resulting from the loss of movement and sensory feedback from the paretic limb. While it is still uncertain what the drivers might be in many cases, Dystonia is considered a condition of maladaptive plasticity within the motor areas of the brain. The prevailing thought is that deep areas of the brain called the basal ganglia begin to incorrectly modulate or control excitability of the more superficial motor areas (primary motor cortex). Recent evidence has shown us that the cerebellum is also involved in the pathophysiology of Dystonia, potentially contributing to the basal ganglia dysfunction. This is interesting as the cerebellum, a small but richly innervated region located at the bottom and back of the brain, is very important for learning new motor tasks (a process that requires neuroplasticity!) and for controlling excitability of the motor cortex. The cerebellum plays a role in sequencing and timing of movements, correcting movements when errors are made and generating motor 'programs' that control well learned automatic movements. Many of these cerebellar functions are affected in Dystonia. Studies using functional MRI have shown the cerebellum is underactive in Dystonia, which could limit the ability of these people to learn new (correct) movement patterns or replace aberrant motor programs that might be driving the dystonic movements. Scientists have recently discovered the cerebellum is important for 'gating' or controlling the level of the sensations coming in from the body. Therefore an underactive cerebellum could be responsible for the excessive response by the brain to feedback sensations from dystonic muscles, as seen in studies in focal hand dystonia.

These new and exciting discoveries may pave the way for scientists to develop new therapies to treat Dystonia. Behavioural techniques that target the cerebellum such as learning sequencing motor tasks may have potential as future treatments. Tasks that train 'implicit' or procedural learning may also be of benefit. There is also the real possibility that non-invasive brain stimulation to directly modulate excitability of the cerebellum may offer an alternative therapy for some people. While there is still a way to go to understanding the causes, mechanisms and most effective treatments for Dystonia, scientists around the

world are making steady progress towards an improved awareness. Recent discoveries that areas of the brain like the cerebellum are affected alongside the more traditional brain regions like the basal ganglia may pave the way for new therapies to treat this complex disorder, at least in some presentations of the disease.