Dystonia

An update on the pathophysiology of isolated focal dystonia

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Dystonia has only been recognised in recent years as a neurological disorder. 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. There are few treatment options and the current standard therapy for many consists of repeated injections of botulinum toxin into affected muscles or physiotherapy with limited success [1]. The ability of neuroscientists to investigate brain activity non-invasively using techniques such as Transcranial Magnetic Stimulation (TMS) has rapidly increased the understanding of the condition. There are now several studies that show Dystonia is a neurological disorder affecting the motor areas of the brain [2]. Studies also show the brain reacts abnormally to sensory inputs from the affected body part. There is a decrease in inhibitory mechanisms within the brain leading to a process that has been called maladaptive brain plasticity[3]. 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. Dystonia is considered a condition of maladaptive plasticity within the motor areas of the brain as 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. The prevailing thought as to the cause of Dystonia 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[4], potentially alongside 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 [5]. 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[6] . 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. 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[6]. Neuroscientists around the world are making steady progress in understanding the causes, mechanisms and most effective treatments for Dystonia.

References

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