DYSTONIA – CASES AND TREATMENT

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Introduction

Dystonia is a movement disorder that causes the muscles to contract involuntarily. This can cause repetitive or twisting movements.[e]

The condition can affect one part of your body (focal dystonia), two or more adjacent parts (segmental dystonia), or all parts of your body (general dystonia). The muscle spasms can range from mild to severe. They may be painful, and they can interfere with your performance of daily tasks.[a]

There's no cure for dystonia, but medications and therapy can improve symptoms. Surgery is sometimes used to disable or regulate nerves or certain brain regions in people with severe dystonia.[b]

Types

Symptoms

Dystonia affects different people in different ways. Muscle spasms might:

- Begin in a single area, such as your leg, neck or arm. Focal dystonia that begins after age 21 usually starts in the neck, arm or face. It tends to remain focal or become segmental.
- Occur during a specific action, such as writing by hand.

- Worsen with stress, fatigue or anxiety.
- Become more noticeable over time.

Areas of the body that can be affected include:

- **Neck (cervical dystonia).** Contractions cause your head to twist and turn to one side, or pull forward or backward, sometimes causing pain.
- **Eyelids.** Rapid blinking or spasms cause your eyes to close (blepharospasms) and make it difficult for to see. Spasms usually aren't painful but might increase in bright light, reading, watching TV, under stress or interacting with people.
- **Jaw or tongue (oromandibular dystonia).** Slurred speech, drooling, and difficulty chewing or swallowing. Oromandibular dystonia can be painful and often occurs in combination with cervical dystonia or blepharospasm.
- **Voice box and vocal cords (laryngeal dystonia).** Have a tight or whispering voice.
- **Hand and forearm.** Some types of dystonia occur only while repetitive activity, such as writing (writer's dystonia) or playing a specific musical instrument (musician's dystonia). Symptoms usually don't happen when arm is at rest.

Causes

The exact cause of dystonia isn't known. But it might involve changes in communication between nerve cells in several regions of the brain. Some forms of dystonia are passed down in families.

Dystonia can also be a symptom of another disease or condition, including:

- Parkinson's disease
- Huntington's disease
- Wilson's disease
- Traumatic brain injury
- Birth injury
- Stroke
- Brain tumor or certain disorders that develop in some people with cancer (paraneoplastic syndromes)
- Oxygen deprivation or carbon monoxide poisoning
- Infections, such as tuberculosis or encephalitis
- Reactions to certain medications or heavy metal poisoning

Symptoms /Signs

1) Age – usually in second decade. Non-progressive in less than 28 years age of onset
2) Diurnal variation –Nocturnal presence signifies secondary causes.
3) Distribution –Focal segmental or diffuse

Findings

1. Cutaneous stigma
2. Organomegaly in secondary causes
3. Activation with specific tasks
4. Disappearance with specific tricks

5. Associated motor disorders –tremors etc.

Case summary

A 16-year-old young girl presented with cramps in hands during writing. Initially was during long time writing but later disabled from routine writing. No cramps were observed in other tasks involving hands. No family history and other secondary causes were ruled out. Routine investigations did not reveal any pathology and MRI could not be done due to claustrophobia. She did well with Baclofen and Clonazepam and refused Botox**.**

Complications

Depending on the type of dystonia, complications can include:

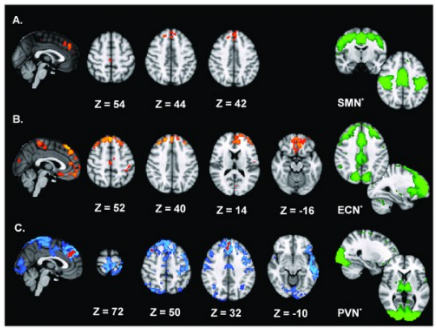
- Physical disabilities that affect your performance of daily activities or specific tasks
- Difficulty with vision that affects your eyelids
- Difficulty with jaw movement, swallowing or speech
- Pain and fatigue, due to constant contraction of your muscles
- Depression, anxiety and social withdrawal

Diagnosis

To determine if underlying conditions are causing symptoms the following are options:

- **Blood or urine tests.** These tests can reveal signs of toxins or of other conditions.

- **MRI or CT scan.** Brains with Dystonia disease appear normal under scan; however, the scan may reveal other conditions. These imaging tests can identify problems in your brain, such as tumours, lesions or evidence of a stroke. (d)



Task-Free Functional MRI in Cervical Dystonia Reveals Multi-Network Changes That Partially Normalize with Botulinum Toxin

Electromyography (EMG). This test measures the electrical activity within muscles. All dystonic muscle involvement detected by EMG evaluation represented genuine dystonic muscle coactivation rather than compensatory muscle activity. The EMG evaluation presented allows quantitative and qualitative identification of dystonic muscle involvement which cannot be achieved by clinical examination.

- **Can an EEG detect dystonia?**

  It will use electroencephalography (EEG) and electromyography (EMG) to compare brain function in normal subjects and in patients with focal hand dystonia. In dystonia, involuntary muscle movements, or spasms, cause uncontrolled twisting and repetitive movement or abnormal postures

- **Genetic testing.** Some forms of dystonia are associated with certain genes. Knowing if you have these genes can help guide treatment. (d)

Treatment

Drugs

1. Clonazepam

2. Anticholinergics

3. Baclofen - Oral /Intrathecal

Advanced

a. Botulinum toxin injections –Restoration of distorted brain motor maps. (b)

b. Deep brain stimulation - Currently, pallidal DBS is an established treatment option for medically refractive dystonia(b)

c. Physical rehabilitation

c.e.1. Concentration exercises

c.e.2. Socially accepted posture training – restoring tricks

c.e.3. Reducing anxiety background

d. Distortion of body image

e) Correction of Distortion of BODY IMAGE

Discussion

Many studies have demonstrated that training utilizing action observation and/or motor imagery improves motor performance. These two techniques are widely used in sports and in the rehabilitation of movement-related disorders. Motor imagery has also been used for brain-machine/computer interfaces (BMI/BCI). During both action observation and motor imagery, motor-related regions such as the premotor cortex and inferior parietal lobule are activated. This is common to actual execution and are involved with the underlying mechanisms of motor learning without execution. Since it is easier to record brain activity during action observation and motor imagery than that during actual sport movements, action observation, and motor imagery of sports skills or complex whole body movements have been utilized to

investigate how neural mechanisms differ across the performance spectrum ranging from beginner to expert. However, brain activity during action observation and motor imagery is influenced by task complexity (i.e., simple vs complex movements). Furthermore, temporal changes in brain activity during actual execution along the long-time course of motor learning are likely nonlinear and would be different from that during action observation or motor imagery. Activity in motor-related regions during action observation and motor imagery is typically greater in experts than in nonexperts, while the activity during actual execution is often smaller in experts than in nonexperts[(d).]

Conclusion

Dystonia, primary types may be varied. Drugs are conventional mechanisms of treatment. However, Botox, DBS, physical rehab and methods to restore altered brain moor maps are newer concepts and now in vogue.

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References

a. Balint B, Mencacci NE, Valente EM, Pisani A, Rothwell J, Jankovic J, Vidailhet M, Bhatia KP. Dystonia. Nat Rev Dis Primers. 2018 Sep 20;4(1):25. doi: 10.1038/s41572-018-0023-6. Erratum in: Nat Rev Dis Primers. 2018 Oct 19;4(1):37. PMID: 30237473.

b. Batla A. Dystonia: A review. Neurol India. 2018 Mar-Apr;66(Supplement): S48-S58. doi: 10.4103/0028-3886.226439. PMID: 29503327.

c. Hu W, Stead M. Deep brain stimulation for dystonia. *Transl Neurodegener*. 2014;3(1):2. Published 2014 Jan 21. doi:10.1186/2047-9158-3-2

d. New AI platform provides diagnosis with high accuracy-By RYAN JASLOW | MASS EYE AND EAR September 28, 2020 ResearchCare Delivery

e) Dystonia –Patient care and staff, Mayo clinic.