Other Movement Disorders

Movement disorders are conditions that cause **abnormal or unwanted movements**. Usually they are caused by damage or malfunction of areas of the brain that are responsible for movement. Movement disorders **can be the main condition or part of another condition**.

Movement disorders can affect actions you choose to do or those you cannot control. They include conditions characterized by having too little or too much movement, or movements arising from malfunctioning programs in the brain.

This information sheet focuses on the movement disorders that are common in individuals with 22q differences. [Note: Please see our sheets on Parkinsonism and Parkinson's Disease and Restless Legs Syndrome (RLS) & Periodic Limb Movement Disorder (PLMD).]

Dystonia

- Single or groups of muscles contract (continuously or intermittently) without full control.
- The patient experiences **twisting movements** and **abnormal postures** of a single or multiple body parts, which can be painful or painless.
- Dystonia has multiple possible causes. It can also be a side effect of antipsychotic medications that treat mental health conditions. Sometimes the exact cause of dystonia is not known.
- Medications, and in extreme cases, brain surgery, may provide relief from symptoms but do not cure or slow down the progression of dystonia.

Myoclonus (Muscle Twitch)

- One or more **muscles briefly contract or relax** .
- The patient experiences **sudden, startle-like movements**.
- Some types of myoclonus are normal (e.g. hiccups, jerking before sleeping, jumping when surprised), but some are abnormal.
- Myoclonus has multiple possible causes. It can be a manifestation of certain **seizure** types. It can also be a side effect of some medications that treat anxiety and depression. Sometimes the exact cause is not known.
- Management depends on the underlying cause.

Developmental Coordination Disorder (DCD; Dyspraxia)

- The cause is unknown, but the brain **struggles to convert the person's intention to move into physical action accurately**.
- The patient may be **clumsy** and **delayed in reaching milestones**. They may have **difficulty with fine motor skills** (e.g. writing, fastening buttons).
- DCD is common among children with 22q11.2DS.
- DCD has no cure, but therapies and adaptations (e.g. pencil grips) may help.

Stereotypies (Motor Stereotypies)

- The cause is unknown but may be due to **chemical imbalance in the brain**.
- The patient shows **patterned and semi-purposeful movements**, e.g. rock their body, nod their head, clench their teeth, flap their arms, open and close their hands, especially during states of heightened emotions.
- Behavioral therapy can be helpful, while medications may be ineffective.

Functional Movement Disorder (FMD)

- The brain has no structural abnormalities but has **problems sending and receiving signals for movements**.
- The patient experiences shaking, jerking, difficulty walking etc. that **feels out of control**, due to errors in the programs of the brain that are needed to produce swift and normal movements. Like a computer, the software becomes glitchy but the hardware is intact.
- Significant injuries, illnesses, stresses, etc. can play a role in FMD.
- Physical, occupational, and behavioral therapies may help the brain relearn movements.

Movement Disorders and 22q11.2DS

Most children with 22q11.2DS have some motor dysfunction.

A 2020 British study found that movement disorders were present in 18/19 (94.7%) of children with 22q11.2DS compared with 4/13 (30.8%) of the unaffected siblings.

Dystonia was the most common movement disorder found in the above study but is still rare in 22q11.2DS. It is recommended children with both 22q11.2DS and dystonia be checked for TANGO2-related disease.

A 2018 British study on 70 children with 22q11.2DS found that 57 (81.4%) had signs of **developmental coordination disorder (DCD)**. 8/9 (88.9%) of the children who were formally assessed indeed had it.

Movement Disorders and 22q11.2DupS

Dystonia, dyspraxia, stereotypies, along with additional unnamed movement disorders, have been reported in children with 22q11.2DupS

Brain and Nerves Series for Individuals with 22q11.2 Differences

Other Movement Disorders (continued)

Motor Tics

- Tics are **quick, repeated movements or sounds**, e.g. blinking that is too frequent, a shrug of the shoulders, a twitch that are often associated with an internal tension (or urge) to move. Performing the tic relieves the tension and can happen in any part of the face and the body.
- The cause is unknown but may be due to **changes in the brain network and chemical balance**.
- Tics can, in extreme cases, cause pain or harm, or are misunderstood as intentionally disruptive. They may increase the risk of the patient being bullied.
- Tics usually tend to disappear with time. If they don't, behavioral therapy and medications may help.

Dyskinesia

- Parts of the body move involuntarily often.
- Dyskinesia are often faster and more random than dystonia.
- In adults with 22q11.2DS, dyskinesia may happen days or months after using certain medications, such as levodopa for Parkinson's disease. **Tardive dyskinesia** (delayed dyskinesia) can happen years after certain medications, such as antipsychotics.
- Adjusting the medication that caused dyskinesia may or may not help.

Tremors

- Parts of the body move rhythmically in a tremulous or trembling fashion. The trembling range from mild to severe.
- There are many causes and types of tremors. In 22q11.2DS, tremors may occur due to **issues in the brain or the nerves**.
- Managing the medical conditions that cause tremors may decrease the symptoms. Medications and physical therapy may also help
- It is recommended that patients with tremors and additional features (slowed movement, stiffness, changes in walking and balance) be **evaluated to see if they have other clinical features that may indicate parkinsonism or Parkinson's disease.**

Movement Disorders in Adults with 22q11.2DS

A 2025 Canadian study examined adults with 22q11.2DS who received care at a movement disorders centre. (This group does not include adults with 22q11.2DS who do not have movement disorders.) Here are the most common movement disorders in these 31 adults.

Non-parkinsonian tremor	68%
Parkinsonism	42%
Dystonia	36%
Myoclonus	29%
Dyskinesia	19%
Stereotypies	13%
Functional Movement Disorders	13%

77% of adults with 22q11.2DS had at least 2 movement disorders.

The complex movement disorders were still present even when drug-related movement disorders were excluded from analyses.

Movement disorders in individuals with 22q differences (especially 22q11.2DS) are complex. It is recommended that patients **receive neurologic exams** every few years. **Recording videos** of unusual movements and **keeping a log** (types of movements, preceding events, and durations) can help neurologists diagnose the condition(s).

Resources

- Movement disorders – Mayo Clinic
- Movement Disorders – Cleveland Clinic
- Dystonia – Cleveland Clinic
- TANGO2-related disease – Tango2 Research Foundation
- Myoclonus – Mayo Clinic
- Myoclonus – Cleveland Clinic
- Developmental co-ordination disorder (dyspraxia) in children – National Health Services, UK
- Dyspraxia – Cleveland Clinic
- Motor Stereotypies – Hopkins Medicine
- Stereotypic Movement Disorder – Cleveland Clinic
- Functional Movement Disorder – Cleveland Clinic
- Tics and Tic Disorders – Cleveland Clinic
- Tardive Dyskinesia – Cleveland Clinic
- Tremor – Cleveland Clinic
- Essential Tremor – Cleveland Clinic
- Essential Tremor – Mayo Clinic
- 22q11.2 duplications – Unique – Rare Chromosome Disorder Support Group
- Updated clinical practice recommendations for managing [children][adults] with 22q11.2 deletion syndrome – 2023

❖ Tremor graphic adapted from Freepik.com

- Dissecting the Phenotypic Spectrum and Complexity of Movement Disorders in 22q11.2 Deletion Syndrome – 2025
- Movement disorder phenotypes in children with 22q11.2 deletion syndrome – 2020
- Developmental coordination disorder, psychopathology and IQ in 22q11.2 deletion syndrome – 2018
- Severe Pediatric Dystonia Responding to Deep Brain Stimulation in 22q11.2 Microduplication Syndrome: Rare Clinical Presentation – 2024
- Atypical nested 22q11.2 duplications between LCR22B and LCR22D are associated with neurodevelopmental phenotypes including autism spectrum disorder with incomplete penetrance – 2019

International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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