# Clinical Diagnostic Evaluation of Dystonia

**A thorough patient history and examination are needed to establish the clinical diagnosis of dystonia; subsequent laboratory, imaging, and genetic testing can assist in identification of etiology and a course of treatment.**

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Dystonia is "*a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive movements, postures, or both*."[1] People often note abnormal pulling or posturing of a body part with impaired voluntary motor control. Dystonia can result in tremulous movements as well. Recognizing dystonia can be a challenge, but its identification is an important first step in discerning an etiology and suggesting a course of treatment. The Figure includes a suggested approach to the clinical evaluation of dystonia.

## History

Careful attention to patient history will highlight features that are consistent with dystonia or point to another diagnosis. Albanese et al[1] proposed a classification of dystonia by 2 axes, with clinical characteristics in axis I and etiologies in axis II. The clinical characteristics include age at onset, body regions affected, temporal pattern (overall disease course and situational variability), and associated features.

### Age at Onset

Age at onset is an important initial consideration that may have relevance in etiology. Generalized dystonia is more common in younger individuals with a genetic etiology compared with adults, who have a higher prevalence of idiopathic focal and acquired dystonias.

### Body Distribution

The body can be separated into craniofacial, oromandibular, cervical, truncal, limb, and laryngeal regions. In focal dystonia, 1 region is affected (eg, a foot turning in, twisting of the neck). Segmental dystonia affects 2 contiguous body regions, such as craniofacial and oromandibular regions. Multifocal dystonia involves >1 noncontiguous region. Generalized dystonia affects the trunk and 2 other regions. Hemidystonia affects >1 region, but only on 1 side of the body and is a red flag for potential lesional causes of dystonia, such as stroke, tumor, or demyelinating lesion.[1]

### Temporal Pattern and Variability

The temporal pattern of symptoms should be considered, including whether the movements started acutely or insidiously, and whether they have been static or progressive over time (either in severity of the initially affected body part or spreading to include other body parts). Most idiopathic and genetic dystonias have an insidious onset and slowly progressive course. Acute dystonic reactions typically occur suddenly, within hours to days after starting or increasing the dose of antidopaminergic medications, including antipsychotics or antiemetics.[2] Sudden onset followed by static course could suggest a vascular cause of dystonia. Rapid-onset dystonia-parkinsonism is a genetic disorder in which dystonia and other features can develop abruptly after a triggering event.[3] Time courses that suggest functional dystonia include history of rapid onset to peak severity, multifocal or generalized distribution at onset, and periods of complete spontaneous remission.[4]

Physicians also need to assess symptom variability (ie, whether the symptoms vary over the course of a day or only occur with specific activities). Dopa-responsive dystonia tends to be diurnal: classically less severe in the morning and becoming more disabling as the day goes on or with prolonged activity. Task-specific dystonia is only present with specific activity, such as with writing or when playing a musical instrument.[5] Runners may develop focal foot dystonia that affects their gait, and other athletes can similarly develop task-specific dystonias related to their sport. Paroxysmal dystonia may occur suddenly and not be associated with a specific

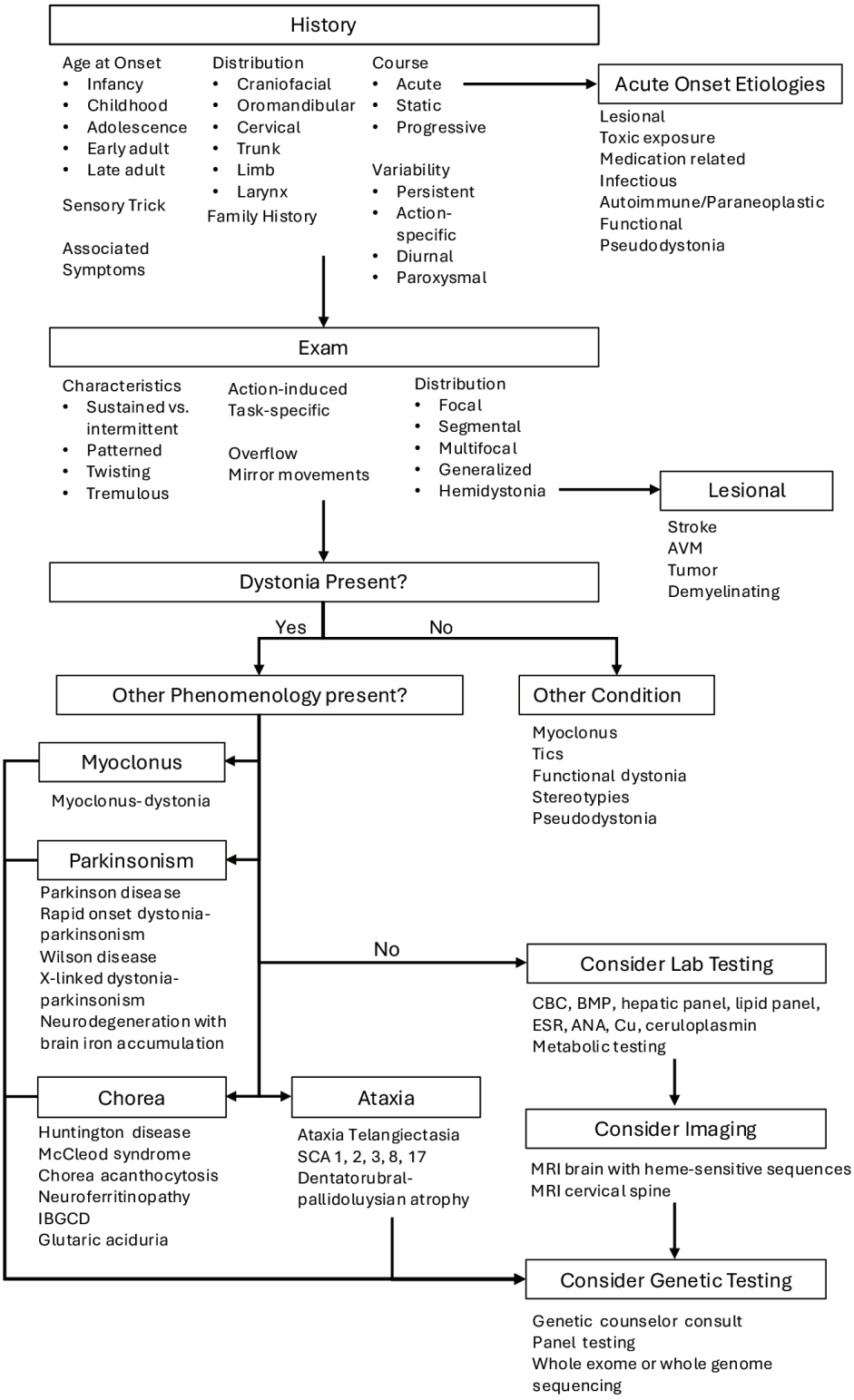


**Figure. A suggested approach to the clinical evaluation of dystonia.**
**Abbreviations: ANA, antinuclear antibody; AVM, arteriovenous malformation; BMP, basic metabolic panel CBC, complete blood count; ESR, erythrocyte sedimentation rate; IBGCD, Idiopathic basal ganglia calcification disease; SCA, spinocerebellar ataxia.**

task, but often has less specific triggers, such as arising from a chair, fasting, or caffeine intake in the cases of paroxysmal kinesigenic, nonkinesigenic, or exercise-related dyskinesia which may have dystonic-type pulling movements as part of the phenomenology.[1]

### Family History

When taking a family history, it is important to identify neurologic diagnoses other than dystonia (eg, Parkinson disease, cerebral palsy) and any family members with similar pulling, twisting movements, or with trouble walking. This may reveal cases of unfinished diagnostic workups or possible misdiagnoses of family members. People from certain ethnic backgrounds have a higher prevalence of specific types of dystonia (eg, Ashkenazi Jewish individuals with DYT-*TOR1A*,[6] male individuals from the island of Panay with X-linked dystonia-parkinsonism).[7] Common genetic dystonia syndromes are shown in Table 1.

### Sensory Tricks

Some maneuvers, collectively known as "*sensory tricks*", may ameliorate the dystonia. The presence of a sensory trick can support the diagnosis of dystonia. People with cervical dystonia may find relief from lightly touching their face or the back of their head. Some people with cervical dystonia may find that wearing scarves ameliorates symptoms and patients with dystonia of the foot or ankle may prefer wearing socks or shoes for similar reasons. Chewing on a toothpick may be helpful for people with oromandibular dystonia. People with writer's cramp may find that holding the pen a different way prevents cramping.

### Exposures

Patient history should reveal any exposures that may have caused or triggered a dystonic reaction, including the use of dopamine-blocking antipsychotics or antiemetics, toxins, physical trauma, or stress.

**TABLE 1. CHARACTERISTICS OF GENETIC DYSTONIAS**

| Dystonia | Inheritance | Onset | Distribution | Course | Associated features |
|---|---|---|---|---|---|
| **Isolated** | | | | | |
| DYT-*TOR1A* (DYT1)[6] | AD | Childhood; less commonly adolescence | Onset mostly lower limb, then generalizes | Progressive | More prevalent in Ashkenazi Jewish individuals |
| DYT-*THAP1* (DYT6)[6] | AD | Childhood; less commonly adolescence or adulthood | Onset upper limb, neck, craniofacial, laryngeal; generalized, multifocal/segmental, focal | Progressive | Phenotypic heterogeneity |
| DYT-*GNAL* (DYT25)[6] | AD | Adulthood | Onset mostly neck; focal, multifocal/segmental | Progressive | |
| DYT-*ANO3* (DYT24)[6] | AD | Infancy/childhood, adulthood | Onset mostly neck; multifocal/segmental | Progressive | Tremor |
| DYT-*KMT2B* (DYT28)[6] | AD | Infancy, childhood | Onset mostly lower limb; generalized | Progressive | Neurodevelopmental disorder |
| **Combined** | | | | | |
| DYT-*SGCE* (DYT11); myoclonus-dystonia[16] | AD | Childhood, adolescence | Upper limb, neck, lower limb; focal, segmental, generalized | Variable | Alcohol-responsive, maternal imprinting; also with myoclonus |
| DYT/*PARK-GCH1* (DYT5a); dopa-responsive dystonia[17] | AD<br>AR (rare) | Childhood, rarely adulthood | Onset in foot or leg; generalized | Progressive, diurnal | Levodopa responsive; worse in evening and with exertion |
| DYT/*PARK-TAF1* (DYT3); X-linked dystonia-parkinsonism[7] | X-linked recessive | Adulthood | Craniofacial, neck, limb, trunk; generalized | Progressive | Parkinsonism more prominent later in the course; predominantly Island of Panay in Philippines; Phasic knee-bending gait[18] |
| DYT/*PARK-ATP1A3*; rapid-onset dystonia-parkinsonism[3] | AD | Childhood, adolescence, adulthood | Prominent bulbar symptoms; rostro-caudal gradient; focal, segmental, generalized | Rapid onset, stepwise deterioration | Triggered by stress, alcohol, exercise, childbirth, illness, hypo- or hyperthermia |

Abbreviations: AD, autosomal dominant; AR, autosomal recessive.
Data from references 3, 6, 7, 16-18.

**TABLE 2. FOCAL DYSTONIAS AND SUGGESTED APPROACHES TO CLINICAL EVALUATION**

| Dystonia | Cervical dystonia[19] | Writer's cramp[5] | Blepharospasm[20] | Adductor laryngeal dystonia[21] |
|---|---|---|---|---|
| Onset | Adulthood (40s) | Adulthood (30s) | Adulthood (50s) | Adulthood (40s) |
| Progression | • Gradually progressive over months to years, then stable<br>• Persistent, may worsen with certain activities<br>• With or without pain | • Focal or progressive; may spread proximally and contralaterally<br>• Task specific | • May progress and spread in 50% of cases | • Gradual, may have sudden onset<br>• Mostly focal, but about 20% spreads<br>• May improve with alcohol |
| Region | • Neck | • Hand | • Bilateral upper face | • Larynx |
| Examination | • Abnormal posturing of neck, with or without tremor<br>• Check active and passive range of motion with rotation, lateral tilt, and forward/backward<br>• Tremor tends to be multidirectional and jerky; decreased at null point | • Have the individual write a sentence multiple times, drawing loops across a page; look for overflow movements proximally<br>• Position the contralateral hand in a visible, relaxed position to look for mirror movements | • Forceful eyelid closure, associated with downward eyebrow movements | • Strained vocal quality, particularly with sounds requiring use of vocal cords<br>• Normal whispering, laughing, crying<br>• Direct laryngoscopy may show dysfunctional vocal cord movement with phonation |
| Sensory trick | • Touching cheek, neck, back of head | • Different grips, touching part of the hand | • Cold water, touching eyelid | • Touching throat, head, abdomen, humming before speaking |
| Mimics[9] | • Atlanto-axial subluxation<br>• Cervical tics<br>• Neck sprain<br>• Compensation for cranial nerve palsy<br>• Abscess<br>• Essential tremor<br>• In children: congenital muscular torticollis, benign paroxysmal torticollis, Sandifer syndrome | • Trigger finger<br>• Peripheral nerve entrapment<br>• Arthritis<br>• Tendinitis<br>• Dupuytren contracture<br>• Cramp | • Hemifacial spasm (unilateral upper and lower face, upward eyebrow movement)[22]<br>• Eyelid-opening apraxia (normal blink, but difficulty opening eyes after)<br>• Ptosis<br>• Myotonia<br>• Dermatochalasia | • Vocal tremor<br>• Muscle tension dysphonia<br>• Vocal cord injury/lesion |

Data from references 5, 9, 19-22.

## Examination

### Movement Characterization

Dystonic movements need to be characterized. Dystonia is characterized by sustained or stereotyped, often jerky, involuntary movements which may have dystonic type pulling movements as part of the phenomenology. As stated previously, these types of movements or postures may affect the face, neck, arms, trunk, legs, or combinations and may only be apparent in certain postures or with certain activities. Individuals' movements should be watched throughout the entire clinical encounter, looking for movement consistency, variability, and triggers. During the examination, it can be helpful to ask individuals to remove their socks and shoes and observe them at rest and with activity, looking for dystonic posturing to occur. Observation of individuals with their arms outstretched and in wing-beating position with eyes open and closed is a helpful way to characterize movements. Finger-tapping tasks may bring out dystonias that are more evident with action. With finger tapping and other unilateral motor movements, the contralateral side at rest may show similar mirror movements. Overflow—contiguous joints posturing when actions are performed (eg, wrist flexion, elbow flexion, shoulder elevation with writing)—also may be noted.

Gait examination is essential for evaluating lower limb dystonia, including assessment of normal walking and possibly stress gaits (eg, toe, heel, tandem, side of feet, running). Postures that occur with exertion may require a longer duration of walking to emerge. For individuals with leg and foot dystonia that affects gait, their gait may improve when walking backwards.

The task that most brings out the abnormal movement should be re-created when possible. For body parts that show posturing, active and passive range of motion and

strength should be checked. Joints affected by dystonia may have some reduced range of motion, but should not be contracted. Specific maneuvers for assessment of common focal dystonias are discussed in Table 2.

### Dystonia vs Other Movement Disorders

Dystonia causes abnormal postures that are stereotyped, may have a twisting or tremulous quality, and may worsen with voluntary action. Key differential diagnoses include other hyperkinetic movement disorders, functional dystonia, and pseudodystonias. Dystonia may be differentiated from chorea in that chorea is irregular and flowing, whereas dystonic movements are patterned and stereotyped. Tics may have a jerky quality to them that are stereotyped but can be suppressed for a time voluntarily and should have associated features of a premonitory urge to move and relief upon performing the movement. Myoclonus may also cause jerky movements, but these are more "shock-like" and much faster (usually <300 ms). Dystonic tremors tend to be jerky and multidirectional, and these characteristics can be demonstrated by having the patient draw spirals.[8] A key feature of dystonia is internal consistency, even if the movements appear unusual. In contrast, functional dystonia may show early, fixed posturing, inconsistent and variable distribution of movements, distractibility or suppressibility with other actions, and a lack of compensatory maneuvers (eg, athletic or effortful gait with no falls).[4] Pseudodystonias present like dystonia but can be attributed to a specific underlying problem, including musculoskeletal abnormalities, neuromuscular disease, sensory disorders, or mass lesions (Table 2).[9] In cases of pseudodystonia due to a muscular or sensory disorder, motor or sensory examinations often show abnormalities. People may have mixed phenomenology, which may point to a combined dystonia syndrome (eg, dystonia-parkinsonism, myoclonus-dystonia) or dystonia with functional overlay.

## Laboratory and Imaging Evaluations in Dystonia

If the predominant movement is diagnosed as dystonia, the next step is determining an etiology. The characteristics gathered through the history and examination should be used to direct laboratory and imaging evaluations if necessary to establish whether the etiology is inherited, acquired, or idiopathic.

### Laboratory Evaluations

The laboratory evaluation of dystonia should be directed based on suspicion built on the history and examination findings. If an acquired cause of dystonia is suspected, screening with a complete blood count, basic metabolic panel, hepatic panel, lipid panel, serum copper and ceruloplasmin, sedimentation rate, and antinuclear antibody may be considered. Dystonia has been described in the context of tuberculous meningitis, viral encephalitis, syphilis infection, HIV infection, and subacute sclerosing panencephalitis.[1] Toxic causes of dystonia include manganese, cobalt, carbon disulfide, cyanide, methanol, disulfuram, and 3-nitropropionic acid toxicity.[1] Dystonia has also been described as a late effect of carbon monoxide poisoning.[10] Autoimmune and paraneoplastic syndromes may have dystonia as part of the presentation; testing cerebrospinal fluid and blood for antibodies associated with these syndromes may be appropriate.[11] Some genetic syndromes, such as Wilson disease, have dystonia as a feature, and laboratory abnormalities may suggest specific underlying etiologies.

### Genetic Disorders

Genetic testing may be warranted even without a suggestive family history, particularly in children and young adults, due to genetic pleiotropy, variable penetrance, and overlapping phenotypes of the genetic disorders listed in Table 1. When genetic testing is unavailable or inconclusive, screening for metabolic abnormalities can provide supportive evidence. Lactate, pyruvate, creatine kinase, ammonia, plasma amino acids, plasma acylcarnitine profile, urine amino acids, urine organic acids, and cerebrospinal fluid amino acids and neurotransmitters may be assessed. Identification of a specific cause may allow for treatment of dystonia and prevent progression of other symptoms associated with the underlying abnormality.[12] The International Parkinson and Movement Disorder Society maintains a list of genes associated with dystonia as identified in the literature[13] (subset in Table 1).

### Neuroimaging

Most cases of idiopathic dystonia have normal imaging study results; however, neuroimaging is useful in cases where lesional or metabolic disorders are suspected. Neuroimaging with MRI brain is recommended if the individual has hemidystonia or other localizing signs. Stroke is the most common cause of hemidystonia, with onset acutely or more typically several months later.[14,15] Wilson disease or neurodegeneration with brain iron accumulation, which may have dystonia as a feature, will show characteristic T2 lesions in the basal ganglia. If Fahr disease is suspected, which may have dystonia as a feature along with parkinsonism, chorea, and neuropsychiatric changes, then CT of the head would best show intracranial calcifications. Imaging of the cervical spine and neck is suggested if pseudodystonia from cervical spine disease, tumor, or abscess is suspected, such as with rapid onset, fever, or pain.

## Conclusion

The evaluation of dystonia is challenging due to the pleomorphic presentations. Dystonia is a hyperkinetic movement disorder that may present with changes in posture; jerky, irregular tremors; and muscle pulling. Symptoms sometimes present consistently, but dystonia may only emerge with

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specific actions, and sometimes only with particular activities that generate specific movements. Dystonia may arise from genetic etiologies, but also may occur in the setting of other diseases (eg, Parkinson disease), or may be acquired with various toxic exposures or structural lesions. A thorough patient history and examination are needed to establish the clinical diagnosis, and subsequent imaging and genetic testing can be ordered to identify etiology and a course of treatment. ■


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