Overwiev of Dystonia: Definition, Classification and Phenomenology

Bülent Elibol MD, PhD

History of Clinical Definitions of Dystonia: Course over Time

Year	Authors	Clinical definitions emphasized
1893	Gowers[42]	Tetanoid chorea
1901	Destarac[43]	Spasmodic torticollis and functional spasms
1903	Leszynsky[44]	Hysterical gait
1908	Hunt[45]	Myoclonia of the trunk
1908	Schwalbe[46]	Maladie des tics; tonic cramps
1911	Ziehen[48]	Torsion neurosis
1911	Oppenheim[41]	Dystonia musculorum deformans: alternating muscle tone
1911	Flatau and Sterling[49]	Progressive torsion spasms
1912	Fraenkel[50]	Rapid, twisting, and sustained movements
1916	Hunt[51]	Slow, torsion, twisting spasms of trunk and extremities, more with active movement
1919	Mendel[52]	Twisting axial postures
1920	Taylor[53]	Spasmodic sustained opisthotonus, violent twisting of the body
1922	Wechsler and Brock[54]	Hyperkinetic and tonic forms
1944	Herz[55]	Slow, long-sustained turning movements
1962	Denny-Brown[56]	Fixed or relatively fixed attitude
1967	Zeman and Dycken[57]	Generalized dystonia; focal dystonias are formes fruste
1976	Fahn and Eldridge[58]	Sustained, involuntary, twisting movements; may be fast or slow
1976	Marsden[59]	Focal dystonias are part of dystonia spectrum
1982	Fahn[60]	Abnormal involuntary movements that are usually twisting; the peak of the movement is sustained for a second or longer
1982	Marsden[61]	Continuous contractions forcing limbs and trunk into sustained postures or intermittent to cause repetitive, sometimes rhythmic abnormal movements
1984	Fahn[62]	Abnormal movements, usually of a twisting nature, ranging in speed from rapid to slow and usually being sustained for a second or longer at the height of the contraction
1987	Fahn, Marsden, and Calne[12]	A syndrome dominated by sustained muscle contractions, frequently causing twisting and repetitive movements or abnormal postures

Fahn, 2011 Mov Disord

“afin qu’il ne fust torty colly”

(in order to prevent him from any twisting of his neck)

François Rabelais (1494–1553) Pantagruel

- *Torticollis is historically the first type of dystonia described*
- *The word "torticolis" of French origin was first used in the mid-16th century.*

Q. D. B. V.

CAPUT OBSTIPUM

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AUCTOR

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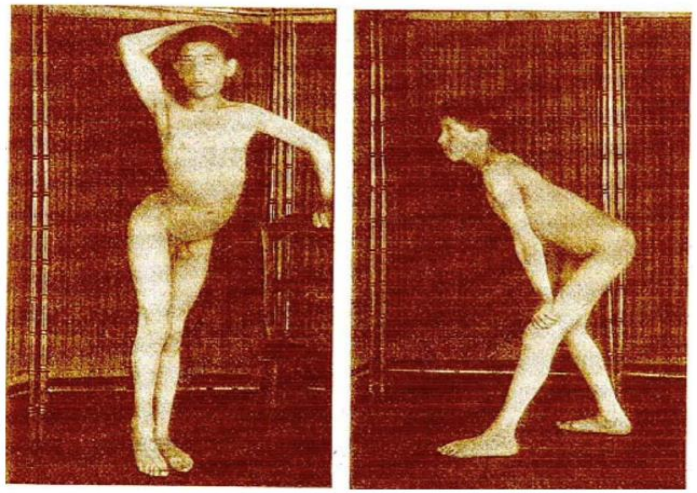
TUBINGÆ, Litteris ROEBELIANIS.

Bruosolle, 2015 Tremor and Other Hyperkinetic Movements

The definition of dystonia, as we understand it today, was first made in 1911…



HISTORICAL REVIEW

Translation of Oppenheim's 1911 Paper on Dystonia

Christine Klein, MD,[1*] and Stanley Fahn, MD[2*]

«dystonia musculorum deformans»

- *Muscle spasms affecting the extremities and trunk, particularly when standing, can cause distorted postures such as lordosis and scoliosis.*
- *A pronounced flexion of the spine, sometimes reminiscent of Oppenheim's "dromedary," worsens when walking.*
- *Movements can be rapid and rhythmic.*
- *Symptoms can lead to progressive and eventually permanent fixed postural deformities*
- *Sometimes accompanied by hypotonic and other times tonic (continuous) muscle spasms, usually occurring during voluntary movements.*
- *No additional neurological findings.*

Traditional Definition in modern era of MDs: During 1970-1990s…

"Dystonia is a sydrome consisting of *sustained* muscle contractions, frequently causing *twisting and repetetive* movements, or abnormal postures"

"Ad hoc coomittee of The Dystonia Reserach Foundation" report, 1984

"Dystonia is a movement disorder chracterized by *sustained or intermittent* muscle contractions causing abnormal, often *repetetive* movements, postures or both"

The modified versions in later guides

REVIEW

CME

Phenomenology and Classification of Dystonia: A Consensus Update

Alberto Albanese, MD,[1,2*] Kailash Bhatia, MD, FRCP,[3] Susan B. Bressman, MD,[4] Mahlon R. DeLong, MD,[5] Stanley Fahn, MD,[6] Victor S.C. Fung, PhD, FRACP,[7] Mark Hallett, MD,[8] Joseph Jankovic, MD,[9] Hyder A. Jinnah, PhD,[10] Christine Klein, MD,[11] Anthony E. Lang, MD,[12] Jonathan W. Mink, MD, PhD,[13] Jan K. Teller, PhD[14]

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Movement Disorders, 2013

REVIEW

Definition and Classification of Dystonia

Alberto Albanese, MD,[1,2*] Kailash P. Bhatia, MD, DM, FRCP,[3] Victor S.C. Fung, PhD, FRACP,[4] Mark Hallett, MD,[5] Joseph Jankovic, MD,[6] Christine Klein, MD,[7] Joachim K. Krauss, MD,[8] Anthony E. Lang, MD, FRCPC,[9,10] Jonathan W. Mink, MD, PhD,[11] Sanjay Pandey, DM,[12] Jan K. Teller, MA, PhD,[13] Marina A.J. Tijssen, MD,[14,15] Marie Vidailhet, MD,[16,17,18] and H.A. Jinnah, MD, PhD[19,20]

ABSTRACT: Dystonia is a movement disorder with varied clinical features and diverse etiologies. Here we present a revision of the 2013 consensus definition and classification of dystonia in light of subsequent publications and experience with its application during the last decade. A panel of movement disorder specialists with expertise in dystonia reviewed the original document and proposed some revision. There was broad consensus to retain the definition of dystonia with only minor clarifications to the wording. Dystonia is defined as a movement disorder characterized by sustained or intermittent abnormal movements, postures, or both. Dystonic movements and postures are typically patterned and repetitive and may be tremulous or jerky. They are often initiated or worsened by voluntary action and frequently associated with overflow movements. The two-axis structure for classification of the many different presentations of dystonia was also retained, with some revision. Axis I summarizes key clinical characteristics of dystonia, including age at onset, family history, body distribution, temporal dimensions, phenomenology, and whether dystonia is isolated or combined with other neurological or medical problems. Axis II organizes information regarding its etiological basis, including genetic, acquired, and anatomical, and common disease mechanisms. This consensus provides an update to the original definition and classification of dystonia with the aim of facilitating its clinical recognition and management. The revision retains the essence of the original proposal and aims particularly to provide a structure facilitating a uniform implementation.

Key Words: dystonia; classification; definition; consensus

Movement Disorders, 2025

Current Definitions of Dystonia

"*Dystonia is a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetetive, movements, postures or both.* Dystonia movements are typically *patterned, twisting*, and may be *tremulous*. Dystonia is *often initiated or worsened by voluntary action* and associated with *overflow muscle activation*"

Concensus Panel on Dystonia , 2013

Dystonia is a movement disorder characterized by sustained or intermittent abnormal movements, postures, or both. Dystonic movements and postures are typically patterned and repetitive and may be tremulous or jerky. They are often initiated or worsened by voluntary action, and frequently associated with overflow movements.

Concensus Panel on Dystonia , 2025

Definition of dystonia is based on basics of phenomenology…

*Dystonia is a movement disorder characterized **by sustained or intermittent abnormal movements, postures, or both.** Dystonic movements and postures are **typically patterned and repetitive** and may be tremulous or jerky. They are often initiated or worsened by voluntary action, and frequently associated with overflow movements.*

Concensus Panel on Dystonia , 2025

Isolated (idiopathic: genes are ?) dystonia

DYT1-TOR1A dystonia

Video

Video

Video

Phenomenology of Dystonia

Dystonia is a movement disorder characterized by sustained or intermittent abnormal movements, postures, or both. Dystonic movements and postures are typically patterned and repetitive and may be tremulous or jerky. ***They are often initiated or worsened by voluntary action,*** *and frequently associated with overflow movements.*

Relationship with voluntary movement

- Task-specific (occurs only with one specific voluntary motor task)
- Action-induced (occurs with a variety of voluntary actions)
- Occurs also at rest (unrelated to voluntary movements)
- Fixed (it is continuous and unalleviated)

Concensus Panel on Dystonia , 2025

❖ *Task-specific (occurs only with one specific voluntary motor task)*

Dystonia is task-specific when it occurs only during a particular motor activity or task, such as writing or playing a specific musical instrument.

Concensus Panel on Dystonia , 2025

Video

Video

❖ *Action-induced (occurs with a variety of voluntary actions)*

A more severe degree of dystonia is induced by a variety of voluntary movements involving the affected body part without specificity for a certain task or activity. Dystonia appearing only with voluntary movement is called "action dystonia".

Concensus Panel on Dystonia , 2025

DRD –first visit

Video

DRD – after 2days of Rx

Video

Parkin mutation + EOPD

Video

❖ *Occurs also at rest (unrelated to voluntary movements)*

An even more severe degree is when dystonia also occurs at rest when the affected body part is not voluntarily activated.

Concensus Panel on Dystonia , 2025

Video

Video

DYT1-TOR1A dystonia

Journal of Neurology, Neurosurgery, and Psychiatry. Special Supplement 1989:96–100

Clinical variants of idiopathic torsion dystonia

STANLEY FAHN

.....classified as being part of other families of dyskinesias. In the former group are paradoxical dystonia, myoclonic dystonia, diurnal dystonia, and dopa-responsive dystonia…….....

However, voluntary movement can even suppress dystonic activity (paradoxical dystonia)

This paradoxical feature most often observed in tardive dystonia (+ dyskinesia) and may lead to confusion

Video

Video

❖ *Fixed (it is continuous and unalleviated)*

- *Finally, dystonia can be fixed or immobile. In this case, the abnormal posture is relatively constant and often cannot be significantly changed by passive movement.*

- *Fixed dystonia with acute or subacute onset in adulthood is often, although not invariably, a presentation of functional dystonia.*

- *Sustained or fixed dystonia can also be seen in rapid-onset dystonia-parkinsonism associated with pathogenic variants in ATP1A3, longstanding pediatric or adult dystonia cases (particularly if untreated or with contractures or osseous bridges), and in parkinsonian syndromes*

Video

Concensus Panel on Dystonia , 2025

Phenomenology of Dystonia

Dystonia is a movement disorder characterized by sustained or intermittent abnormal movements, postures, or both. Dystonic movements and postures are typically patterned and repetitive ***and may be tremulous or jerky.*** *They are often initiated or worsened by voluntary action, and frequently associated with overflow movements.*

Additional phenomenological characteristics

- *Alleviating maneuvers (sensory trick, geste antagoniste)*
- *Tremulous (tremor-like)*
- *Jerky (myoclonic-like)*

Concensus Panel on Dystonia , 2025

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Concensus Panel on Dystonia , 2025

❖ *Alleviating maneuvers (sensory trick, geste antagoniste)*

This term is synonymous with 'sensory trick' and 'geste antagoniste', indicating different types of voluntary maneuvers that temporarily improve dystonic postures

- *It is commonly observed in cervical dystonia, blepharospasm, oromandibulolingual dystonia, and writer's cramp. Most common in cervical dystonia (50%).*
- *It is known as "geste antagoniste." However, stimulation in both directions is effective. Cheek and chin contact are most effective. Attempting to normalize the antagonists by contracting them makes this maneuver more effective.*
- *Even approaching the arm and visualization can alleviate dystonic activity.*
- *As the severity of dystonia increases, a more forceful maneuver is required (a "forceful" trick).*

Müller et al., 2001, Schramm et al., 2004; 2007, Ochudlo et al., 2007

Concensus Panel on Dystonia , 2025

Video

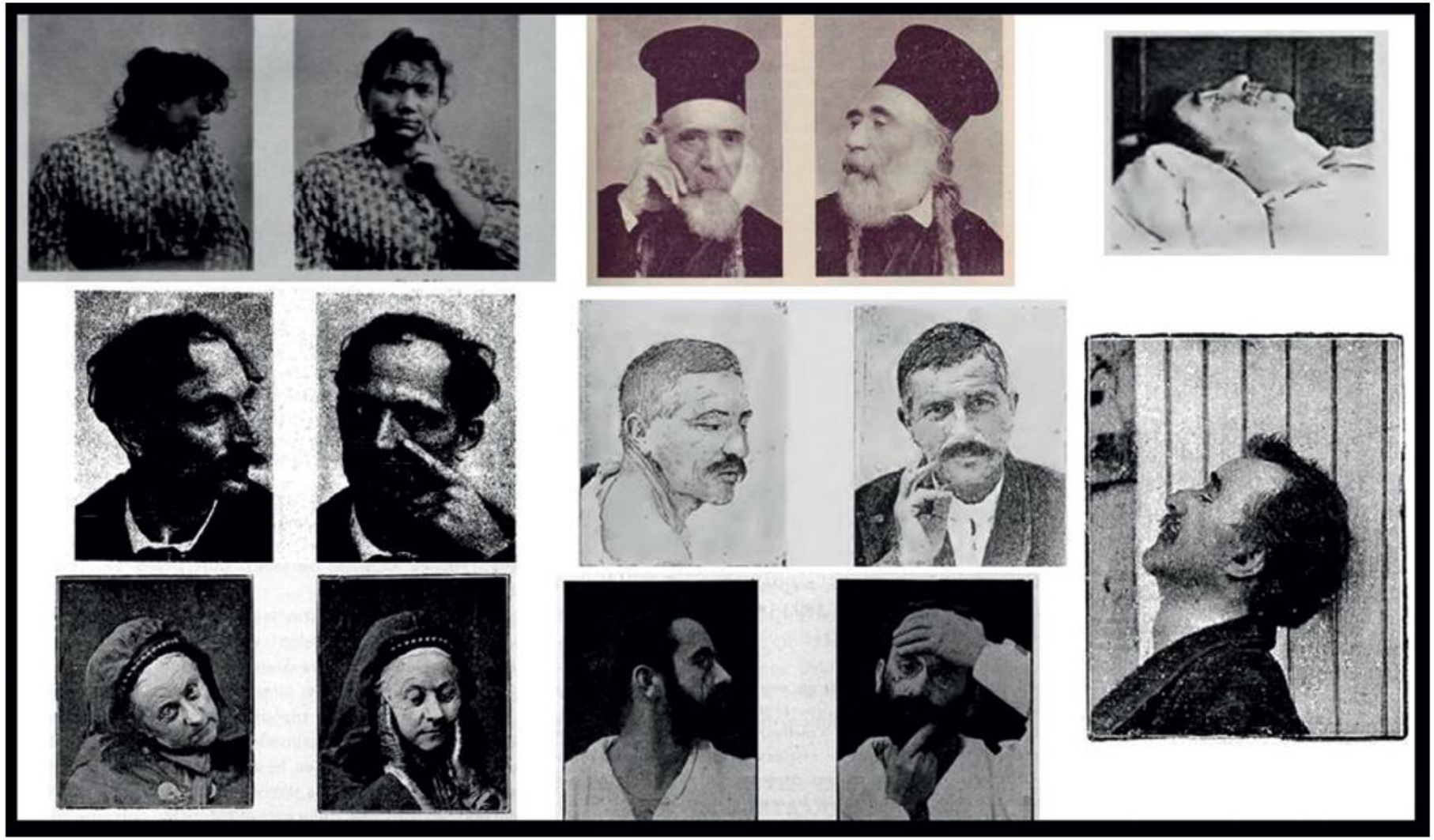

Figure 4. **Photographic Illustration of the Seven Cases Presented by Brissaud in His 1894 Lesson.** On the far right is a patient who tries to reduce

Broussole ve ark, 2015

REVIEW

Tricks in dystonia: ordering the complexity

Vesper Fe Marie Llaneza Ramos,[1] Barbara I Karp,[2] Mark Hallett[1]

J Neurol Neurosurg Psychiatry 2014;**85**:987–993.

Cervical dystonia

Specific parts of the face, cheek, chin, occipital region, temple, forehead, nose, mastoid, occipital region, back of neck. Raising the arm and holding the finger near the target region without touching the face.

Visual fixation at a specific target, focusing on stationary objects while walking, looking at oneself in the mirror

Forcible tricks with counterpressure to the cheek, temple, chin, back of head, mastoid, forehead

Resting the back of the head or neck, bending the trunk forward, resting the back or shoulder, yawning, wearing a collar/a scarf, leaning the elbows on the armrest

Imagining or merely thinking about performing a sensory trick

Apraxia of eyelid opening, blepharospasm

Tight goggles or spectacles, Lundie Loops, Device inserted in glasses to mimic touch to lateral eyelid

Touching/pulling eyelids, tape on eyebrow; specific parts of face (forehead, nose, side of eyelids, chin)

Pushing back of the head, Massaging cheek bones, eyelid, forehead. Closing the jaw, chewing gum, Touching bitemporal skin beside eyes. Covering the eye Picking teeth Wearing a cap or turban.

Meige syndrome

Sleeping/relaxing, talking, singing/humming, pulling on upper eyelid, pinching back of the neck, yawning, belching, sucking in or blowing out cheeks, drinking cold and/or alcoholic beverage

Lower cranial dystonia, oromandibular dystonia

Toothpick in mouth, holding object clenched between the teeth, dental splint, touching lip or lower corner of face
'Mandibular sensory trick device'
Touching tongue to palate, biting lips, swallowing, pulling face up, bending neck forward
Chewing gum, sucking. Kissing, whistling. Pen/cigarette/tongue depressor in mouth
Biting food/plastic between left upper and lower molars→ dental prosthesis device 3 mm above molar
Smiling, singing, talking, thinking about talking,Biting piece of cotton or plastic, Dental splint
Playing with larger mouthpieces, 5 min ice massage of facial muscles

Laryngeal dystonia

Grimacing, laughing, loud background noise.

Writer's cramp

5 min immersion in cold water, Shifting pen holding, holding pen between index finger and thumb vertically, writing with a closed fist, Use pens of different sizes and calibres, using chalk and blackboard or painting
Touching specific part of the contralateral normal hand to a specific part of the dystonic hand

Runner's dystonia

Holding hands over head, run in a clockwise direction, mental imagery of running in a clockwise direction
Beach walking, applying pressure with hand at hip.

Camptocormia

Low-slung backpack, using wheeled walker, pressing back against hallway.

❖ ***Interestingly, sensory trick can also occur in psychogenic dystonia, but very rarely ...***

Video

Video

Phenomenology of Dystonia

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Additional phenomenological characteristics

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Concensus Panel on Dystonia , 2025

❖ *Tremulous (tremor-like) phenomenology - Dystonic tremor*

*There is a close relationship between **dystonia** and **tremor.** Some have attempted to characterize **tremulous dystonic movements** with the term **"dystonic tremor,"** separating this from a distinct postural and action tremor commonly seen in patients with dystonia **("dystonia with tremor").** This has resulted in the term "dystonic tremor" being used inconsistently over time to emphasize different aspects.*

*The revised consensus proposal also more explicitly recognizes dystonic movements with a **tremor-like (rhythmic or quasi-rhythmic)**. If the tremor is instead considered an independent movement disorder coincidental with dystonia, it is listed under the associated features. In the past, expressions such as "dystonic tremor" or "tremor associated with dystonia" were used to describe the varied phenomenological coincidence of dystonia and tremor. Usage of these terms is currently not recommended.*

Concensus Panel on Dystonia , 2025

'Dystonic tremor'

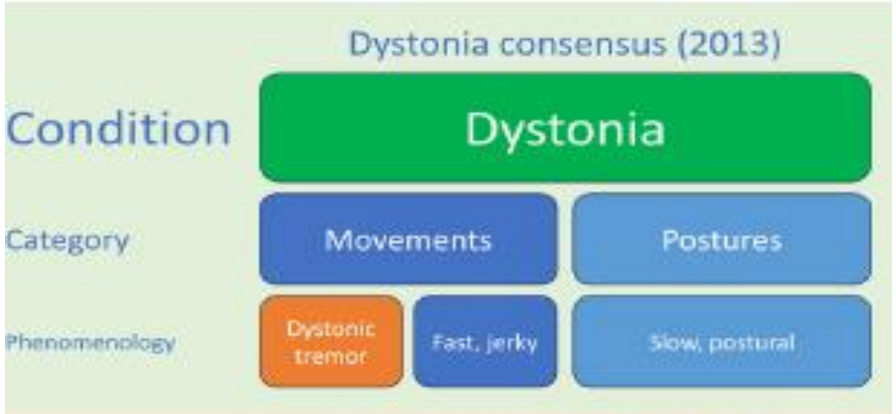
- Spontaneous, oscillatory, rhythmic, but not continuous movements are produced by the contraction of dystonic muscles, often exacerbated during the act of maintaining primary (normal) posture.

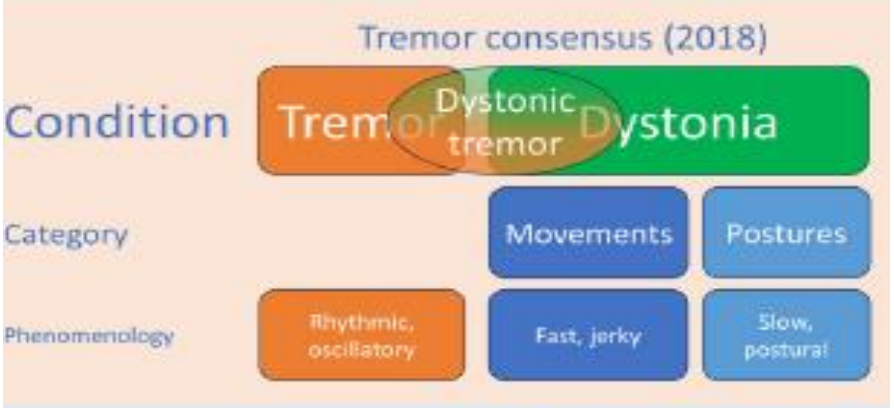
Video

- Dystonic tremor may resolve when the movement that caused the dystonic posture is allowed to fully develop (a "null point").
- Unlike the sinusoidal oscillations of true tremor, it exhibits directional dominance (rapid jerks in one direction alternate with slower jerks in the opposite direction).

Video

- These partially rhythmic contractions, better visualized by electrophysiological recordings, can be observed over a wide range of frequences (3-12 Hz)





Movement Disorders
CLINICAL PRACTICE

Dystonic Tremor: Time to Change

Stefania Lalli, MD, PhD and Alberto Albanese, MD*

" dystonic tremor is spontaneous oscillatory, rhythmical, although often inconstant, patterned movement produced by contractions of dystonic muscles often exacerbated by an attempt to maintain primary (normal) posture'

2013 , Consensus Panel on Dystonia

*dystonic tremor is a syndrome "combining tremor and dystonia as the leading neurological signs"*According this definition any condition combining dystonia and tremor in the same patient configures a syndrome of dystonic tremor
= irregular tremolous dystonia + more rhytmic occilatory dystonic tremor and + 'tremor associated with dystonia'

2018 , Consensus Panel on Tremor

The suggested definition incorporates assigns the jerky component to dystonic movements and considers oscillating and regular tremor a separate feature of dystonia. This update also accommodates the observation that isolated tremor may herald overt dystonia with later appearance.

2024 , (Mov Disord Clin Prac).

Phenomenology of Dystonia

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Concensus Panel on Dystonia , 2025

❖ *Jerky (myoclonic-like) phenomenology*

This term is used when dystonic movements resemble myoclonic jerks. It should not be used as a synonym of irregularity of movements or postures. Occasionally electrophysiology may be necessary to distinguish myoclonus from jerky dystonia.

Video

Video

Jerky dystonia in the past was also called "myoclonic dystonia," an expression that is also currently discouraged, as it may cause confusion with myoclonus-dystonia.

Concensus Panel on Dystonia , 2025

Two other phenomenological characteristics of dystonia that may help to understand pathophysiology, but also are helpful in clinical practice

❖ *Overflow dystonia*

Dystonic movements and postures can "overflow" into apparently unaffected body segments that are contiguous with the affected body region, causing inappropriate and excessive movements unrelated to the intended voluntary movement.

Video

❖ *Mirror dystonia*

Mirror dystonia is defined as a dystonic posture or movement, usually similar to that produced during action, that can be elicited in a dystonic limb at rest, when contralateral movements or actions are performed with the unaffected limb.

When the dystonia is task-specific, the contralateral movement inducing the mirror dystonia is typically the task affected by the dystonia (eg, writing). Mirror dystonia is distinguished from mirror movements.

Video

But sometimes, dystonic activity can lead to interesting phenomenologies…

Video

The Classification of Dystonia

Traditional Classification of Dystonia

- *Based on age of onset*
  - Early onset: < 20/26 years *(for primary dystonia that begins in the legs or arms and progresses over time)*
  - Late onset: > 20/26 years (*for primary dystonia that begins cranio-cervical region or in the arms and generally does not spread)*
- *Based on topographic distribution*
  - *Focal (single body region)*
  - *Segmental (two or more related body regions)*
  - *Multifocal (two or more unrelated body regions)*
  - *Hemystonia (ipsilateral arm & leg)*
  - *Generalized (leg(s) + trunk + another body region)*
- *Based on etiology*
  - *Primary dystonias*
  - *Secondary dystonias*

REVIEW

CME

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Movement Disorders, 2013

Axis I. Clinical characteristics

Clinical characteristics of dystonia

Age at onset

- Infancy (birth to 2 years)
- Childhood (3–12 years)
- Adolescence (13–20 years)
- Early adulthood (21–40 years)
- Late adulthood (>40 years)

Body distribution

- Focal
- Segmental
- Multifocal
- Generalized (with or without leg involvement)
- Hemidystonia

- Temporal pattern
  - Disease course
  - Static
  - Progressive

Variability

- Persistent
- Action-specific
- Diurnal
- Paroxysmal

- Associated features

Isolated dystonia or combined with another movement disorder

- Isolated dystonia
- Combined dystonia
- Occurrence of other neurological or systemic manifestations

  List of co-occurring neurological manifestations

Axis II. Etiology

Nervous system pathology

- Evidence of degeneration
- Evidence of structural (often static) lesions
- No evidence of degeneration or structural lesion

Inherited or acquired

- Inherited
- Autosomal dominant
- Autosomal recessive
- X-linked recessive
- Mitochondrial

Acquired

- Perinatal brain injury
- Infection
- Drug
- Toxic
- Vascular
- Neoplastic
- Brain injury
- Psychogenic

Idiopathic

- Sporadic
- Familial

REVIEW

Definition and Classification of Dystonia

Alberto Albanese, MD,[1,2*] Kailash P. Bhatia, MD, DM, FRCP,[3] Victor S.C. Fung, PhD, FRACP,[4] Mark Hallett, MD,[5] Joseph Jankovic, MD,[6] Christine Klein, MD,[7] Joachim K. Krauss, MD,[8] Anthony E. Lang, MD, FRCPC,[9,10] Jonathan W. Mink, MD, PhD,[11] Sanjay Pandey, DM,[12] Jan K. Teller, MA, PhD,[13] Marina A.J. Tijssen, MD,[14,15] Marie Vidailhet, MD,[16,17,18] and H.A. Jinnah, MD, PhD[19,20]

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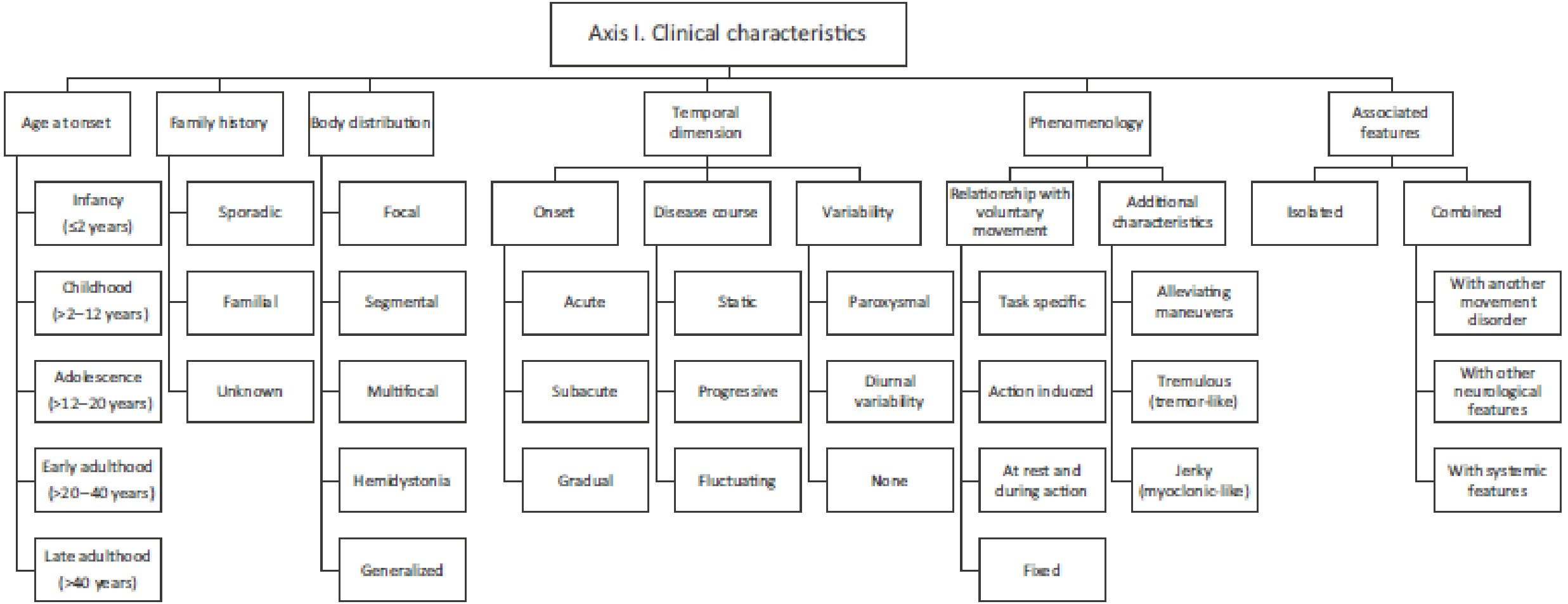
Key Words: dystonia; classification; definition; consensus

Movement Disorders, 2025


Axis I. Clinical characteristics
Age at onset
Infancy (≤2 years)
Childhood (>2–12 years)
Adolescence (>12–20 years)
Early adulthood (>20–40 years)
Late adulthood (>40 years)
Family history
Sporadic
Familial
Unknown
Body distribution
Focal
Segmental
Multifocal
Hemidystonia
Generalized
Temporal dimension
Onset
Acute
Subacute
Gradual
Disease course
Static
Progressive
Fluctuating
Variability
Paroxysmal
Diurnal variability
None
Phenomenology
Relationship with voluntary movement
Task specific
Action induced
At rest and during action
Fixed
Additional characteristics
Alleviating maneuvers
Tremulous (tremor-like)
Jerky (myoclonic-like)
Associated features
Isolated
Combined
With another movement disorder
With other neurological features
With systemic features

Axis I. Clinical characteristics

Clinical characteristics of dystonia

Age at onset
- Infancy (≤2 y)
- Childhood (>2–12 y)
- Adolescence (>12–20 y)
- Early adulthood (>20–40 y)
- Late adulthood (>40 y)

Family history
- Sporadic
- Familial
- Unknown

Body distribution
- Focal
- Segmental
- Multifocal
- Hemidystonia
- Generalized

Temporal dimensions

Onset
- Acute
- Subacute
- Gradual

Course
- Static
- Progressive
- Fluctuating

Variability
- Paroxysmal
- Diurnal variability
- None

Phenomenology

Relationship with voluntary movement
- Task-specific
- Action-induced
- Occurs also at rest
- Fixed

Additional characteristics
- Alleviating maneuvers (sensory trick, geste antagoniste)
- Tremulous (tremor-like)
- • Jerky (myoclonic-like)

Isolated or combined
- Isolated
- Combined
  - With another movement disorder
  - With other neurological features
  - With systemic features

Axis I: Clinical Features

a-Age of onset

According to previous classifications:

- *Early-onset: < 26 years*
- *Late-onset: > 26 years*

Or

- *0-12 years*
- *12-20 years*
- *>20 years*

According to the 2013 classification,

- *Infancy (birth - 2 years)*
- *Childhood (3-12 years)*
- *Adolescence (13-20 years)*
- *Young adulthood (21-40 y)*
- *Adulthood (>40 years)*

According to the 2025 classification,

- *Infancy (birth - 2 years)*
- *Childhood (3-12 years)*
- *Adolescence (13-20 years)*
- *Young adulthood (21-40 y)*
- *Adulthood (>40 years)*

Axis I: Clinical Features

b-Body distribution

According to previous classifications:

- *Focal*
- *Segmental*
- *Multifocal*
- *Hemi-dystonia*
- *Generalized*

(legs + trunk or another body part)

According to 2013 classification:

- *Focal*
- *Segmental*
- *Multifocal*
- *Hemidystonia*
- *Generalized*

(trunk + two body parts - with or without leg involvement)

According to 2025 classification:

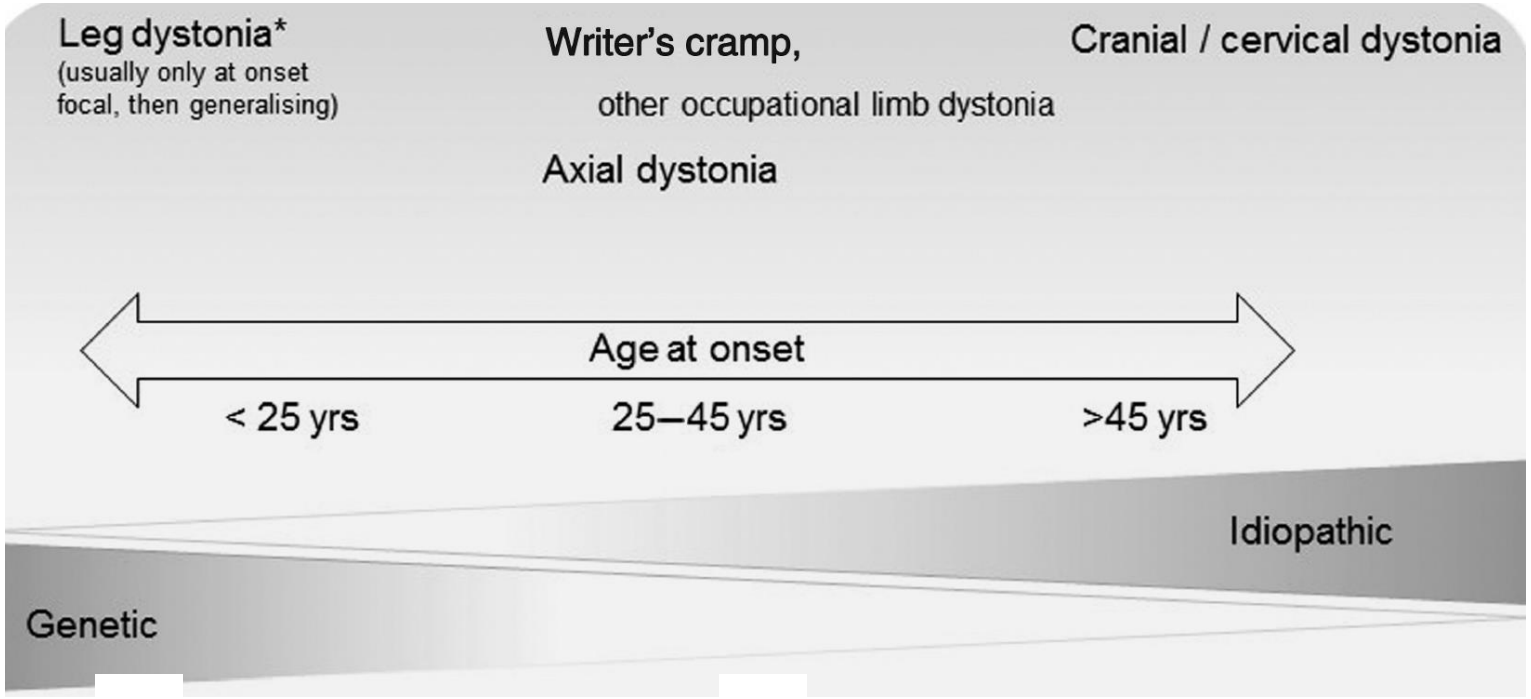
- *Focal*
- *Segmental*
- *Multifocal*
- *Hemidystonia*
- *Generalized*

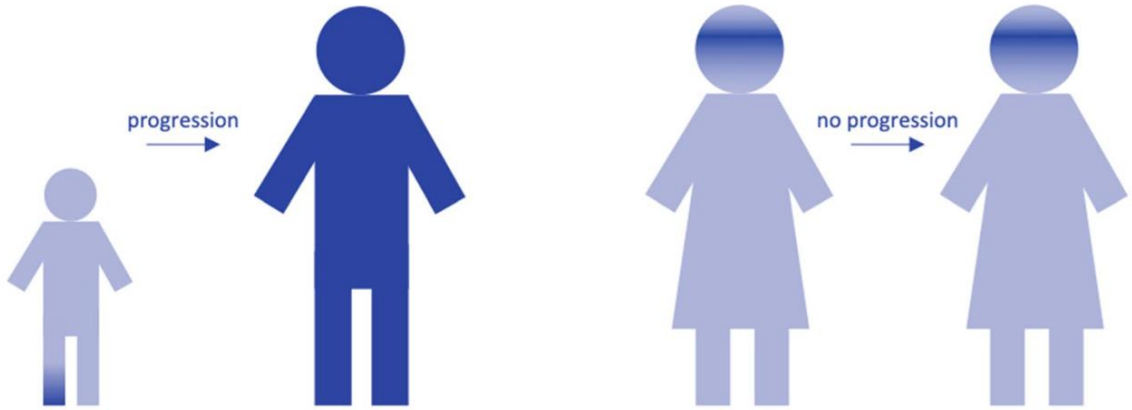
(Beyond multifocal dystonia: trunk + two additional regions, three or four body parts without trunk involvement)

According to 2025 classification:

- *Focal: only one of the 7 body regions are affected*
  - *upper face, lower face (including jaw, mouth and tongue), neck (including shoulders), larynx, upper limbs (including upper and lower arms and hands but not shoulders), trunk lower limbs (including upper and lower legs and feet)*
- *Segmental: two or three contiguous body sites are affected (without qualifing for hemidytonia)*
- *Multifocal: two or three noncontiguous body sites are affected*
- *Hemidystonia: unilateral involvement of the upper and the lower limbs (with or without trunk, neck, or cranial regions)*
- *Generalized: Beyond affection of multifocal distribution; trunk + two additional regions or three or four body parts without trunk involvement*

The age of onset and body distribution are important for planning treatment strategies, but also help to predict the cause anf course in isolated dystonias…





Balint, Erro, Bhatia, 2017

Axis I: Clinical Features
c-Temporal pattern

According to previous classifications:

- *None*

According to 2013 classification:

- Disease course
  - *Static*
  - *Progressive*
- Variability
  - *Persistent*
  - *Action-specific*
  - *Diurnal*
  - *Paroxymal*

According to 2025 classification:

- Onset
  - Acute
  - Subacute
  - Gradual
- Disease course
  - *Static*
  - *Progressive*
  - *Fluctuating*
- Variability
  - *Paroxymal*
  - *Diurnal variability*
  - *None*

Axis I: Clinical Features

d- Phenomenology

According to previous classifications:

- *None*

According to 2013 classification:

- *None*

According to 2025 classification:

- Relationship with volantary movement
  - Task spesific
  - Action induced
  - At rest and during action
  - Fixed
- Additional characteristics
  - Alliviating maneuvers
  - Tremolous (tremor-like)
  - Jerky (myclonus-like)

Axis I: Clinical Features
e-Accompanying features

According to previous classifications:

Only tremor is included

(by definition)

Primary dystonias are defined based on accompanying features:

- *Primary pure dystonia*
- *Primary dystonia-plus syndromes*

According to the 2013 classification:

- *Isolated dystonia (tremor)*
- *Combined dystonia*

  With other Movement Disorders (parkinsonism, myoclonus, etc.)

- *With other neurological or systemic symptoms*

  'Complex dystonia'

  (Listing of accompanying symptoms)

According to the 2025 classification:

- *Isolated dystonia (tremor)*
- *Combined dystonia*
  - *With other Movement Disorders (tremor, parkinsonism, myoclonus, chorea, ataxia etc.)*
  - *With other neurological features (spasticity, epilepsy,develop.delay)*
  - *With systemic features*

Axis II. Etiology

Nervous system pathology
- Evidence of degeneration
- Evidence of structural (often static) lesions
- No evidence of degeneration or structural lesion

Inherited or acquired

Inherited
- Autosomal dominant
- Autosomal recessive
- X-linked recessive
- Mitochondrial

Acquired
- Perinatal brain injury
- Infection
- Drug
- Toxic
- Vascular
- Neoplastic
- Brain injury
- Psychogenic

Idiopathic
- Sporadic
- Familial

Concensus Panel on Dystonia , 2013

Axis II: etiology and pathogenesis

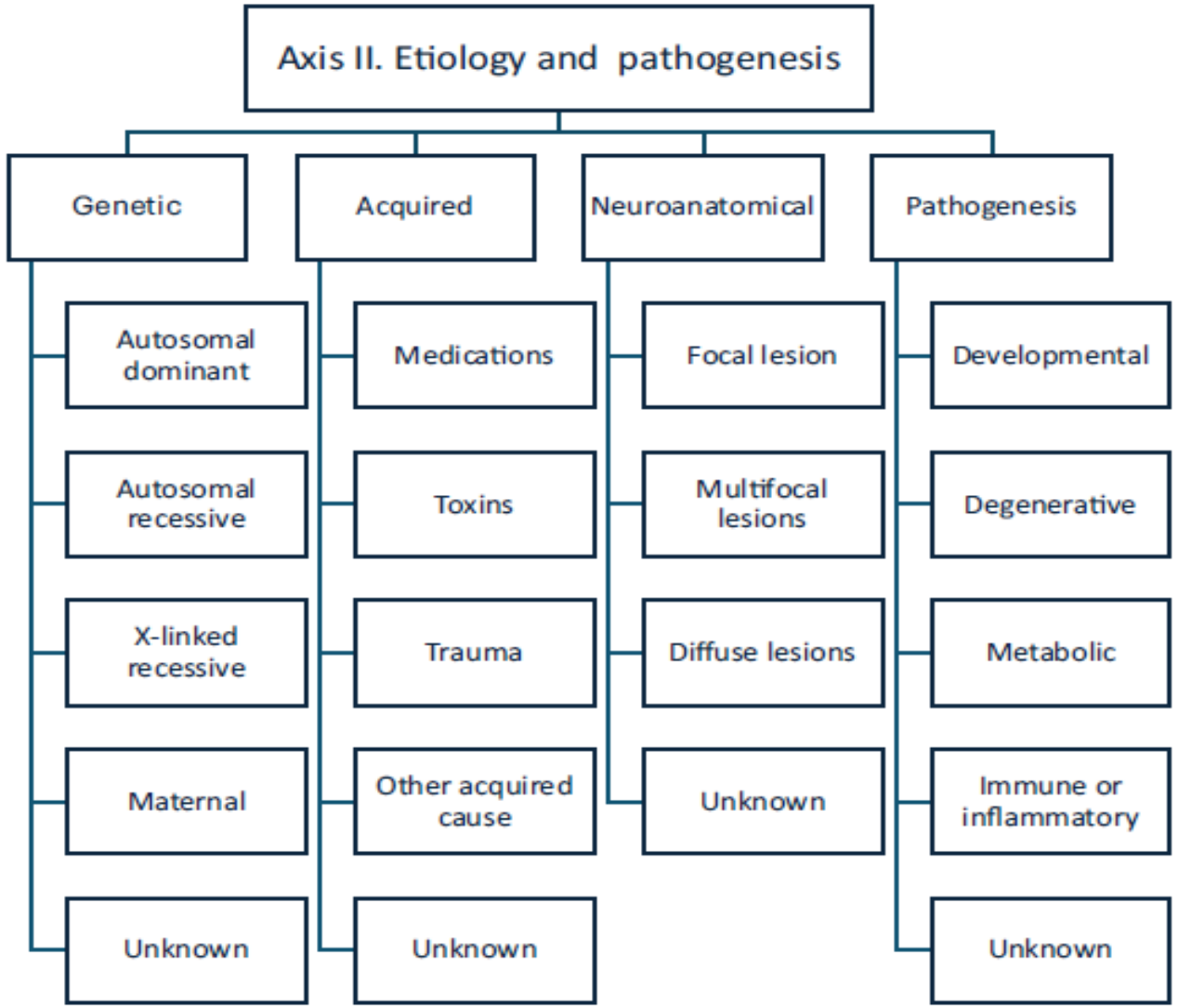
Genetic
- Autosomal dominant
- Autosomal recessive
- X-linked recessive
- Maternal
- Unknown

Acquired
- Medications
- Toxins
- Trauma
- Other acquired cause
- Unknown

Neuroanatomical
- Focal lesion
- Multifocal lesions
- Diffuse lesions
- Unknown

Pathogenesis
- Developmental
- Degenerative
- Metabolic
- Immune or inflammatory
- Unknown

Concensus Panel on Dystonia , 2025



Genetic

- This section corresponds to the "inherited" category of the 2013 classification with minör improvements.

Acquired

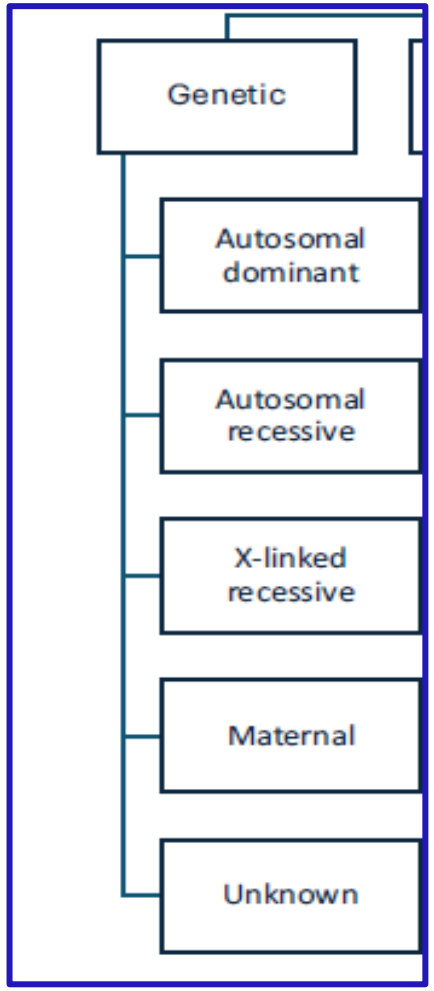
- This section improves and refines the acquired category of the 2013 classification.

Neuroanatomical

- This section now provides a strictly anatomical description of observed lesions; the "degenerative" descriptor has been moved to the next section.

Mechanisms/Pathogenesis

- This new section lists the most common pathogenic disease mechanisms.



Monogenic forms of dystonia

Isolated dystonia

- DYT-ANO3
- DYT-EIF2AK2
- DYT-GNAL
- DYT-HPCA
- DYT-KMT2B
- DYT-PRKRA
- DYT-THAP1
- DYT-TOR1A
- DYT-VPS16
- DYT-AOPEP

Combined dystonia

- DYT-COX20
- DYT-DNAJC12
- DYT-SLC39A14
- DYT/PARK-ATP1A3
- DYT/PARK-GCH1
- DYT/PARK-TAF1
- DYT/PARK-TH
- DYT/CHOR-GNAO1
- MYC/DYT-KCTD17
- MYC/DYT-SGCE

Complex dystonia

- DYT-ACTB
- DYT-ATP7B
- DYT-BCAP31
- DYT-DCAF17-(NBIA)
- DYT-DDC
- DYT-FITM2
- DYT-IRF2BPL
- DYT-MECR
- DYT-mt-ND6
- DYT-OPA1
- DYT-PANK2-(NBIA)
- DYT-SERAC1
- DYT-SLC19A3
- DYT-SUCLA2
- DYT-TIMM8A
- DYT-TUBB4A
- DYT-VAC14
- DYT/CHOR-ACAT1
- DYT/CHOR-ADAR1
- DYT/CHOR-FOXG1
- DYT/CHOR-GCDH
- DYT/CHOR-HPRT
- DYT/CHOR-MUT
- DYT/CHOR-PCCA/PCCB
- DYT/PARK-CP-(NBIA)
- DYT/PARK-GLB1
- DYT/PARK-PLA2G6-(NBIA)
- DYT/PARK-PTS
- DYT/PARK-QDPR
- DYT/PARK-SLC6A3
- DYT/PARK-SLC30A10
- DYT/PARK-SPR
- ATX/DYT-SQSTM1

Disorders that usually present with other phenotypes but can have predominant dystonia

- ATX-ATXN3
- HSP-C19orf12-(NBIA)
- HSP/ATX-FA2H-(NBIA)
- HSP/ATX-KIF1C
- CHOR-FTL-(NBIA)
- PARK-DNAJC6
- PARK-WDR45-(NBIA)

List of genes causing neurodevelopmental delay and dystonia

(Conditions where less prominent dystonia can be encountered in the setting of predominant developmental disorders or epileptic encephalopathy)

- *ARX*
- *CTNNB1*
- *GNB1*
- *SUCLG1*
- *VPS41*
- *YY1*

Thomsen et al., Annu. Rev. Pathol. Mech. Dis. 2024

Genotype–Phenotype Relations for Isolated Dystonia Genes: MDSGene Systematic Review

Lara M. Lange, MD,[1†] Johanna Junker, MD,[1,2†] Sebastian Loens, MD,[1,2†] Hauke Baumann, PhD,[1†] Luisa Olschewski,[1] Susen Schaake, BSc,[1] Harutyun Madoev, MSc,[1] Sonja Petkovic, PhD,[1] Neele Kuhnke, BSc,[1] Meike Kasten, MD,[1,3] Ana Westenberger, PhD,[1] Aloysius Domingo, MD, PhD,[4] Connie Marras, MD,[5] Inke R. König, PhD,[6] Sarah Camargos, MD,[7] Laurie J. Ozelius, PhD,[8] Christine Klein, MD,[1,2] and Katja Lohmann, PhD[1*]

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Lange ve ark., Mov. Dis. 2021

DYT-TO1A ……. isolated >>> isolated + tremor >>> combined with myoclonus

DYT-THAP1……. isolated >> isolated + tremor >>> combined with myoclonus

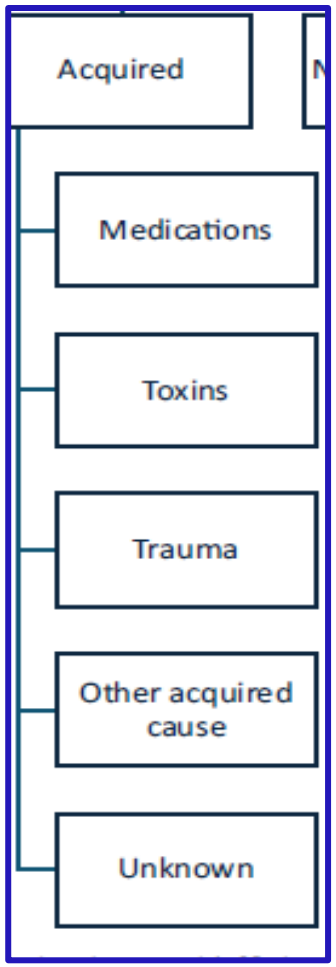
DYT-GNAL ……. isolated > isolatede+ tremor>>> combined with myoclonus

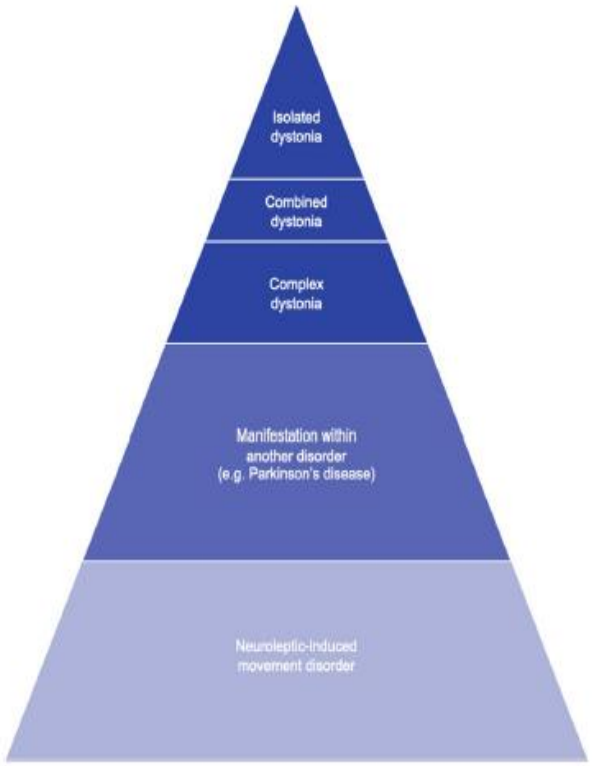
DYT-ANO3 ……. isolated ~ = isolated+ tremor> combined with myoclonus

DYT-PRKRA ……. isolated+ tremor ~ = myoclonus and/or Parkinsonism > isolated

DYT-KMT2B……. Complex > >> isolated/ + tremor >>> combined with myoclonus

Tardive dystonia: Phenomenology





Grütz & Klein, 2021

Phenomenologically usually similar to idiopathic dystonias, but typical perentations may help to Dx

- More often cranio-cervical onset (~70-80%)
- Retrocollis and trunk extensor involvement are common.
- Adduction of the arm, extension of the elbow, flexion of the wrist, and pronation of the forearm are quite typical.
- Pelvic thrusting is quite specific to tardive dystonia.
- Only 15% remain focal, segmental progression is common
- Generalization is less common (13%)
- Sensory trics and the effects of Btx-A are similar to those of idiopathic dystonia.
- They may decrease with voluntary movement (e.g.,like paradoxical dystonia)

Tardive dystonia: cervical

Video

Tardive dystonia: focal-upper limb

Video

Tardive dystonia: cranio-cervical

Video

Tardive dystonia: truncal

Video

Video

Tardive Oculogyric Crises

- *OC are often a complication of acute neuroleptic Rx, and in some combined forms of dystonia syndromes*

- *But, they can be tardive.*

- *In this case, it presents more frequently with seizures and is accompanied by anxiety, increased psychotic thought disorder, and hallucinations.*

Video

Pseudo-dystonia

- *Dystonic (tonic) tics*
- *Head tilt (vestibulopathy, trochlear nerve palsy)*
- *Spinal curvature, camptocormia, scoliosis*
- *Atlanto-axial subluxation, shoulder subluxation*
- *Arnold-Chiari malformation*
- *Neck soft tissue mass*
- *Congenital muscular torticollis*
- *Congenital Klippel-Feil syndrome*
- *Satoyoshi syndrome*
- *Duputyren contractures*
- *Trigger finger*
- *Neuromuscular causes (Isaacs syndrome)*
- *Spasms (hypocalcemia, hypomagnesemia, alkalosis)*
- *Orthopedic and rheumatological causes*
- *Sandifer syndrome*
- *Deafferentation (pseudoathetosis)*

Concensus Panel on Dystonia , 2013

Dystonia mimics

- *This term is preferred to pseudodystonia to indicate a variety of motor abnormalities that conform in part to the definition of dystonia.*
- *Functional (psychogenic) dystonia is considered a dystonia mimic.*

Concensus Panel on Dystonia , 2025

Proposed classification of pseudodystonia and list of pseudodystonias:

Pseudodystonia in non-neurological disorders of the musculoskeletal system

- Camptocormia, scoliosis
- Subluxation (atlanto-axial, shoulder, hip, other joints)
- Joint deformities (rheumatic diseases)
- Arthrogryposis
- Trigger finger
- Dupuytren's contracture
- Limb contractures
- Soft tissue mass (e.g. neck, retropharyngeal space)
- Sandifer syndrome
- Congenital muscular torticollis
- Klippel-Feil syndrome

Pseudodystonia in disorders of sensory pathways

- Parietal lobe damage (stroke, mass lesion)
- Syringomyelia
- Subacute combined degeneration
- Mono- and polyneuropathy
- Myelopathy

Pseudodystonia in disorders of motor pathways

- Stiff-person syndrome
- Neuromyotonia
- Myotonic disorders
- Spasms due to electrolyte disturbances (e.g. carpopedal spasms)
- Tetanus

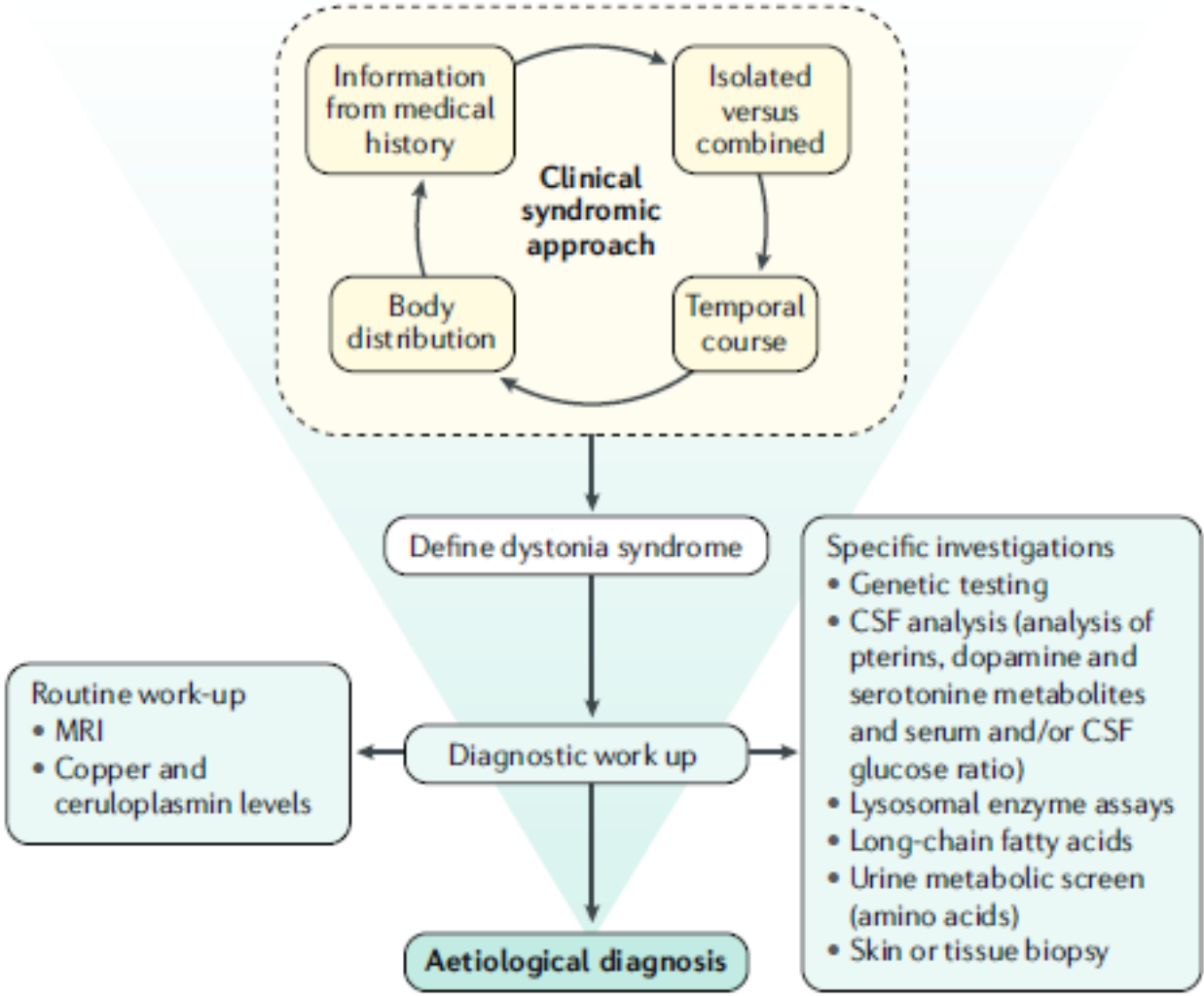
Multifocal motor neuropathy and other causes of peripheral nerve hyperexcitability

- Compensatory postures in other
- neurological disorders
- Head tilt due to vestibulopathy,
- trochlear/abducens nerve palsy
- Mass lesion in the posterior fossa

Berlot, Bhatia, Kojovic, 2019

Syndromic approach to dystonia diagnosis

- *What is the nature of the movement disorder that dominates the picture?*
- *What other movement disorders or neurological findings are present?*
- *What is the temporal evolution of the disease?*
- *Age of onset, body distribution*
- *The order in which neurological symptoms develop*
- *Are there other systemic findings?*
- *What information does brain imaging provide?*
- *Are the results of simple biochemical analyses informative?*
- *Are more specialized investigations necessary?*
  (electrophysiology, CSF analysis, urine metabolic screening)
- *Classify the patient's dystonic syndrome based on the above data and establish an etiological differential diagnosis.*
- *Perform specific diagnostic tests (genetic testing, tissue biopsy)*



Balint et al., Nature Rev, 2019

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Thank you…

Thank you for your attention…