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ORIGINAL ARTICLE

Expert recommendations for diagnosing cervical, oromandibular, and limb dystonia

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Abstract

Background Diagnosis of focal dystonia is based on clinical grounds and is therefore open to bias. To date, diagnostic guidelines have been only proposed for blepharospasm and laryngeal dystonia. To provide practical guidance for clinicians with less expertise in dystonia, a group of Italian Movement Disorder experts formulated clinical diagnostic recommendations for cervical, oromandibular, and limb dystonia.

Methods A panel of four neurologists generated a list of clinical items related to the motor phenomenology of the examined focal dystonias and a list of clinical features characterizing neurological/non-neurological conditions mimicking dystonia. Thereafter, ten additional expert neurologists assessed the diagnostic relevance of the selected features and the content validity ratio was calculated. The clinical features reaching a content validity ratio > 0.5 contributed to the final recommendations.

Results The recommendations retained patterned and repetitive movements/postures as the core feature of dystonia in different body parts. If present, a sensory trick confirmed diagnosis of dystonia. In the patients who did not manifest sensory trick, active exclusion of clinical features related to conditions mimicking dystonia (features that would be expected to be absent in dystonia) would be necessary for dystonia to be diagnosed.

Discussion Although reliability, sensitivity, and specificity of the recommendations are yet to be demonstrated, information from the present study would hopefully facilitate diagnostic approach to focal dystonias in the clinical practice and would be the basis for future validated diagnostic guidelines.

Keywords Focal dystonia · Pseudodystonia · Diagnosis

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Introduction

Adult-onset focal dystonia, the most frequent form of dystonia, usually manifests as blepharospasm (BSP), cervical dystonia (CD), oromandibular dystonia (OMD), laryngeal dystonia (LD), and upper limb dystonia (ULD) [1, 2]. In adulthood, the lower limb has rarely been observed as a site of dystonia presentation [3]. At present, the diagnosis of focal dystonias is mainly based on clinical grounds and is therefore open to bias. Dystonia may be diagnosed several years after the first symptoms manifest, and many patients may visit numerous physicians, delaying access to treatment [2, 4]. Family studies indicate that up to half of people with dystonia may be undiagnosed or misdiagnosed [5]. The phenomenological variability of dystonia and the existence of a number of neurological and non-neurological conditions (pseudodystonia) mimicking the abnormal movements or postures induced by dystonia are probably the main factors contributing to misdiagnosis [1]. Diagnostic accuracy varies considerably among physicians and experienced neurologists can diagnose focal dystonia with greater accuracy than general neurologists [6, 7]. Until validated diagnostic biomarkers are not available, clinical expert opinion will therefore be the gold standard diagnostic technique.

In 2013, a task force of the Movement Disorders Society provided a revised definition of dystonia as "a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive, movements, postures, or both. Dystonic movements are typically patterned, twisting, and may be tremulous. Dystonia is often initiated or worsened by voluntary action and associated with overflow muscle activation." [1]. The task force also highlighted several neurological and non-neurological conditions mimicking dystonia (pseudodystonia). Nevertheless, a currently unmet need is the formulation of diagnostic guidelines for each form of focal dystonia in order to systematize the diagnostic process, to make it reproducible across centers, and to make it applicable by clinicians with less expertise in dystonia [8].

To date, specific diagnostic guidelines have been proposed and validated only for BSP and laryngeal dystonia [9, 10]. The BSP diagnostic algorithm, yielding 93% sensitivity and 90% specificity, started with an item describing the phenomenology of eyelid spasms leading to eyelid narrowing/closure and followed by recognition of "sensory trick," a highly specific dystonic feature that however is not present in all patients. If sensory trick was absent, then features that could help to differentiate BSP from conditions mimicking BSP were analyzed.

In this paper, a group of Italian movement disorder experts developed diagnostic recommendations for cervical, oromandibular, and limb dystonia as part of the Italian Dystonia Registry initiative [5].

Methods

Formulation of expert recommendations was first based on the careful assessment of the clinical features contributing to the spectrum of dystonia at each body site, and on the identification of neurological and non-neurological conditions mimicking focal dystonias. A panel of four movement disorder experts (GD, RP, ME, and CS) generated a list of clinical items related to the motor phenomenology (positive features) of CD, OMD, ULD, and lower limb dystonia (LLD). According to the validated diagnostic guidelines for BSP [9], sensory trick was not analyzed because there was wide consensus that it was a highly specific feature that is almost exclusively present in dystonia [11]. The expert panel (EP) also identified neurological/non-neurological conditions mimicking dystonia at different body sites. Then, a list of clinical features that are part of the clinical spectrum of these conditions but are usually absent in dystonia was developed (negative features).

In the next step, the complete list of positive and negative features was examined by a diagnostic recommendation committee (DRC) of ten experts with long-standing experience in dystonia who did not participate in the EP. Members of the DRC were asked to indicate what positive/negative features could aid in diagnosing focal dystonia and excluding conditions mimicking dystonia. To assess the diagnostic relevance of each item, the content validity ratio (CVR) was calculated according to the following formula [9]:

$$\mathrm{CVR} = (\mathrm{ne}–\mathrm{N}/2)/(\mathrm{N}/2)$$

where ne = number of raters indicating the item as "relevant" and N = total number of raters. The CVR ranged between − 1 (this means that all raters judged the item as non-relevant) and + 1 (all raters judged the item as relevant). For the diagnosis of focal dystonia, we arbitrarily considered potentially useful those positive and negative clinical features reaching a CVR $\geq$ 0.5 [4]. It is worth noting that there was sometimes reciprocity between positive and negative features. As an illustrative example, the "patterned and repetitive neck movements/postures" characterizing CD is opposite to the "non-patterned and non-repetitive head movements" characterizing choreic movements. When this occurred, the positive feature alone was included in the final recommendation.

Results

Content validity analysis

Table 1 lists the positive and negative clinical features identified by the EP and the results of CVR analysis. Positive clinical features included spontaneous or triggered movements/postures, change of abnormal posture with activity,

stereotyped/patterned movements/postures, tremor, and dystonia in other body parts.

The conditions mimicking CD and the correspondent negative features were orthopedic neck diseases (like atlanto-axial and shoulder subluxation, or fracture of the cervical vertebrae), rheumatologic neck diseases, and posterior fossa tumors, all leading to fixed involuntary posture; lower motor neuron disease/myopathy/myasthenia gravis inducing weakness of the neck muscles opposite to the abnormal posture; neck chorea producing non-repetitive head movements; neck tics associated with ability to mentally suppress the spasms; vestibular torticollis characterized by loss of balance induced by the voluntary correction of the abnormal posture; and ocular torticollis characterized by diplopia caused by the voluntary correction of the abnormal neck posture.

The conditions mimicking OMD and the correspondent negative features identified by the EP were hemimasticatory spasm producing unilateral masseter and temporal muscle hypertrophy, bruxism, a condition characterized by prolonged and forceful jaw-closing movements with marked dental attrition (causing lesions of teeth and periodontium) during sleeping time or emotional situations; geniospasm, a genetic condition characterized by autosomal dominant inheritance and high penetrance; lower motor neuron disease/myopathy/myasthenia gravis associated with masticatory muscle weakness and chewing difficulty; temporo-mandibular joint disorder resulting in jaw luxation, subluxation or ankylosis, and producing fixed postures; edentulous dyskinesia associated with edentulism; jaw chorea inducing non-repetitive movements; and jaw tics associated with ability to mentally suppress the spasms.

Clinical conditions mimicking ULD or LLD and the correspondent negative features were orthopedic and rheumatologic diseases (like arthrosis, De Quervain's disease, professional disease affecting musicians, typists, seamstresses, mechanics, shoemakers, Dupuytren's disease; tenosinovytis; trigger finger; kneecap or elbow bursitis; rotator cuff injury, epicondylitis, ankylosing spondylitis, gout, achillodynia, Morton disease, complex regional pain syndrome) leading to abnormal fixed postures of the upper/lower limb; lower motor neuron disease/miopathy inducing weakness in the upper limb or lower limb muscles (tarsal tunnel syndrome, lumbosacral stenosis, lesion of lumbosacral plexus, peroneal neuropathy) antagonizing the abnormal posture, upper motor neuron disease inducing spasticity and weakness of muscles causing the abnormal posture; chorea inducing non-patterned and non-repetitive involuntary movements; tics associated with ability to mentally suppress the spasms; and stiff person syndrome producing stiffness of the axial muscles leading to hyperlordosis and superimposed painful spasms triggered by tactile or auditory stimuli.

Diagnostic recommendations

The positive and negative features that reached a CVR $\geq$ 0.5 were included in the diagnostic recommendations. The selected items were combined in an algorithm (Figs. 1, 2, and 3) that started with recognition of those positive features considered to be the basic phenomenologic aspect of the various focal dystonias that is patterned and repetitive movements/postures in a body site that may occur either spontaneously or triggered by motor tasks. The patterned and repetitive quality helped to differentiate focal dystonia by choreic movements. The second step was identification of a sensory trick. In the absence of a sensory trick, negative features to be excluded to diagnose dystonia at different body sites were "fixed involuntary posture"; "weakness of muscles antagonizing the abnormal posture", "weakness of muscles causing the abnormal posture", "ability to mentally suppress the spasms," and, exclusively for CD, "diplopia induced by the voluntary correction of the abnormal head posture".

Discussion

Clinical studies have suggested that experienced neurologists can diagnose focal dystonia with greater accuracy than general neurologists [6, 7]. Until validated diagnostic biomarkers are available, clinical expert opinions will therefore be the gold standard diagnostic technique. The phenomenological variability of dystonia and the existence of a number of neurological and non-neurological conditions (pseudodystonias) mimicking the abnormal movements or postures induced by dystonia are probably the two main factors contributing to misdiagnosis [1]. The present recommendations were designed to minimize both sources of diagnostic errors and to provide practical guidance for clinicians with less expertise in dystonia.

The procedure we used started with formulation of a list including both phenomenological aspects of each focal dystonia and the most relevant clinical features characterizing the correspondent pseudodystonias. The DRC was then asked to select those aspects thought to have sufficient discriminatory power to identify focal dystonia and to exclude pseudodystonias. Even according to previous studies [9], we considered that four experts were enough to detail the phenomenological aspects of dystonia, and that including a larger number of raters in the second group would ensure an accurate weighing of the proposed diagnostic features.

According to the 2013 revised definition of dystonia [1], the present recommendations considered patterned and repetitive movements/postures in different body parts as the core features distinguishing dystonia from other involuntary movements like chorea. Clinical features contributing to the phenomenological spectrum of dystonia but possibly present in

Table 1 Positive and negative clinical features of focal dystonia at different body sites and results of content validity ratio analysis

	Cervical dystonia	Oromandibular dystonia	Upper limb dystonia	Lower limb dystonia
Positive features (clinical items related to the motor phenomenology of dystonia) [content validity ratio (CVR)]	Head/neck movements/postures inducing head deviation from neutral position [CVR, 1]	Oromandibular movements/postures—either spontaneous or triggered by motor tasks [CVR, 1]	Spontaneous or task-induced involuntary movements /postures of one or more segments of the upper limb [CVR, 0.8]	Spontaneous or action-induced involuntary movements /postures of one or more segments of the lower limb [CVR, 0.8]
	Change of abnormal posture with activity [CVR, 0.6]	Change of abnormal posture with activity [CVR, 0.6]	Change of abnormal posture with activity [CVR, 0.6]	Change of abnormal posture with activity [CVR, 0.6]
	Stereotyped or patterned movements [CVR, 0.6]	Stereotyped or patterned movements [CVR, 0.6]	Stereotyped/patterned movements [CVR, 0.6]	Stereotyped/patterned movements [CVR, 0.6]
	Head tremor [CVR, 0.4]	Jaw tremor [CVR, 0.4]	Upper limb tremor [CVR, 0.4]	Lower limb tremor [CVR, 0.4]
	Dystonia in other body parts [CVR, − 0.2]	Dystonia in other body parts [CVR, − 0.2]	Dystonia in other body parts [CVR, − 0.2]	Dystonia in other body parts [CVR, − 0.2]
Negative features (clinical items related to conditions mimicking dystonia) [content validity ratio (CVR)]	Fixed involuntary posture [CVR, 0.6]	Masseter and temporal muscles hemiatrophy [CVR, 0.2]	Fixed involuntary posture [CVR, 0.6]	Fixed involuntary posture [CVR, 0.6]
	Pain and other sensory symptoms [CVR, − 0.4]	Pain and other sensory symptoms [CVR, − 0.4]	Pain and other sensory symptoms [CVR, − 0.4]	Pain and other sensory symptoms [CVR, − 0.4]
	Weakness of neck muscles antagonizing the abnormal posture [CVR, 0.6]	Dental attrition causing lesions of teeth and periodontium [CVR, 0.2]	Weakness of muscles antagonizing the abnormal posture [CVR, 0.8]	Weakness of muscles antagonizing the abnormal posture [CVR, 0.8]
	Non stereotyped/patterned movements [CVR, 0.6]	Autosomal dominant inheritance [CVR, 0.2]	Weakness of muscles inducing the abnormal posture [CVR, 0.6]	Weakness of muscles inducing the abnormal posture [CVR, 0.6]
	Ability to voluntarily suppress spasms [CVR, 1]	Masticatory muscle weakness [CVR, 0.8]	Non stereotyped/patterned movements [CVR, 0.6]	Non stereotyped/patterned movements [CVR, 0.6]
	Loss of balance induced by the voluntary correction of the abnormal posture [CVR, 0.2]	Jaw luxation, subluxation, or ankylosis by temporo-mandibular joint disorders [CVR, 0.6]	Ability to voluntarily suppress spasms [CVR, 1]	Ability to voluntarily suppress spasms [CVR, 1]
	Diplopia induced by voluntary correction of the abnormal neck posture [CVR, 0.6]	Edentulism [CVR, 0.3]	Stiffness of the axial muscles leading to hyperlordosis and superimposed painful spasms [CVR, 0.1]	Stiffness of the axial muscles leading to hyperlordosis and superimposed painful spasms [CVR, 0.1]
		Non stereotyped or patterned movements [CVR, 0.6]		
		Ability to voluntarily suppress movements/postures [CVR, 0.9]		

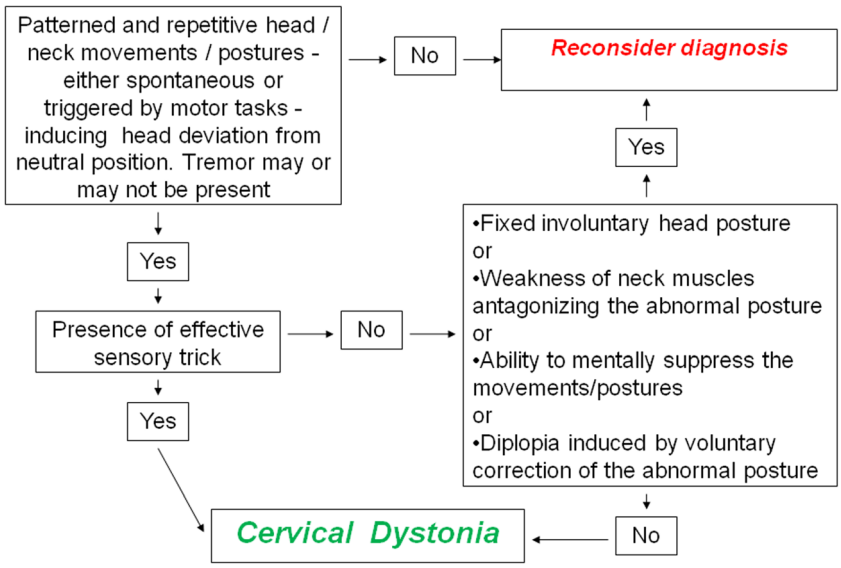

Fig. 1 Proposed diagnostic algorithm for cervical dystonia

other movement disorders (like tremor), signs that may merely reflect local disorders coexisting with dystonia (like pain and other sensory symptoms), and conditions due to involvement of distant muscles (like dystonia in other body sites) were excluded. Under recognition of patterned and repetitive abnormal movements/postures (that would be expected to be always present in dystonic patients but can also be caused by other pathologies), the presence of geste antagoniste (also called sensory trick), a highly specific maneuver that may induce transient amelioration of dystonia [11, 12], confirmed dystonia diagnosis. Sensory trick is particularly frequent in CD and OMD whereas it is probably negligible in LLD [11]. Being almost exclusively to dystonia, the presence of sensory trick adds specificity to diagnosis. However, a variable percentage of dystonia patients does not manifest sensory trick. In these patients, active exclusion of clinical features related to conditions mimicking dystonia (features that would be expected to be absent in dystonia) is therefore needed.

Negative features thought to be useful in the differential diagnosis of the various focal dystonias included "fixed involuntary posture"; a feature distinguishing dystonia from orthopedic or rheumatologic diseases inducing fixed postures; weakness of muscles antagonizing the abnormal posture, a feature that may prove useful to differentiate lower motor neuron diseases/myopathy from dystonia; weakness of muscles causing the abnormal posture, a feature that distinguished spasticity and dystonia; and ability to mentally suppress the spasms, a feature that is potentially useful to distinguish dystonia and tics. It is worth noting that suppressibility on demand needs to be carefully differentiated by compensatory movements that often counteract dystonic movements or postures and are also the result of voluntary action; moreover,

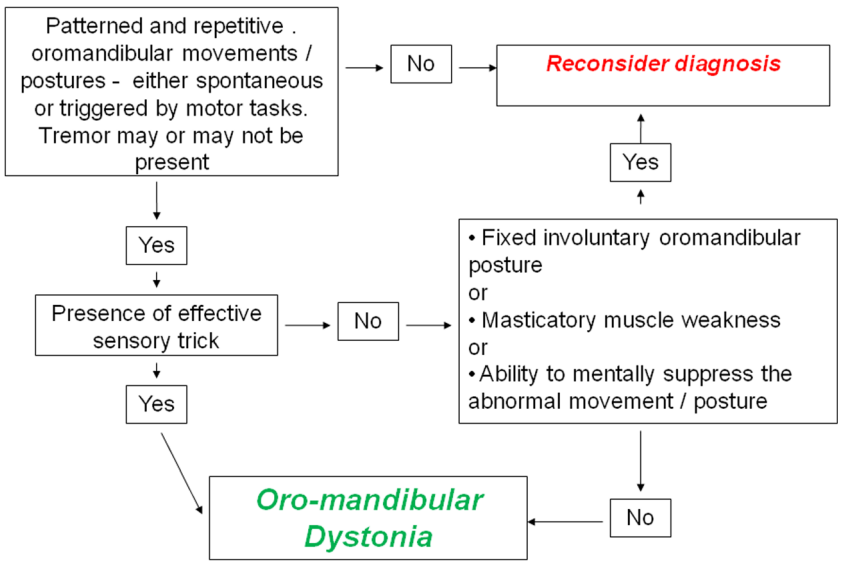

Fig. 2 Proposed diagnostic algorithm for oromandibular dystonia

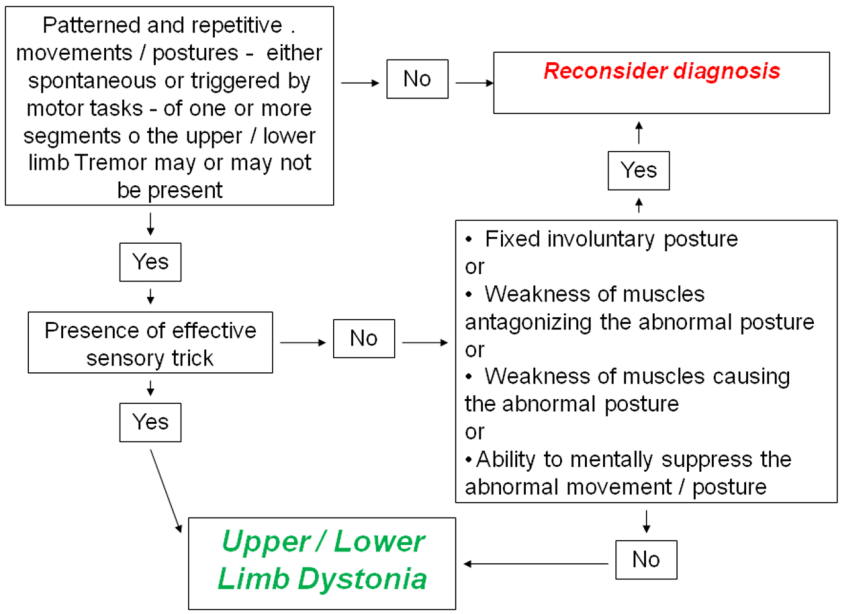

Fig. 3 Proposed diagnostic algorithm for limb dystonia

dystonic tics sometimes have intermediate features between dystonia and tics.

Negative features have to be all absent for focal dystonia to be diagnosed. If only one criterion is not satisfied, then specific alternative diagnoses need to be considered. We were however aware that signs suggesting an alternative diagnosis do not necessarily imply that a diagnosis of dystonia cannot be made. Rather, examiner judgment should be used to decide whether examination findings are entirely attributable to the alternative diagnosis or whether additional dystonia is present. For example, scoliosis, a potential cause of abnormal fixed posture of the neck and trunk, can nevertheless be associated with CD as a risk factor for the condition [13].

Recommendations for OMD did not aid in differentiating OMD from three rare conditions causing jaw spasms that may resemble jaw-closing OMD. Hemimasticatory spasm [14, 15], with or without facial hemiatrophy, is characterized by spasms of masticatory muscles. The disease course and, when present, facial hemiatrophy help to distinguish hemimasticatory spasm and OMD [4]. Bruxism is typically a nocturnal problem, even though awake bruxism does exist and pose diagnostic challenges with jaw-closing OMD [16]. Geniospasm is characterized by episodes of involuntary tremor of the chin and lower lip that typically start in early childhood [17]. Early onset and inheritability [18] can help to distinguish geniospasm and OMD, even though rare large families with adult-onset cranial dystonia have been reported [19].

Our recommendation could not be helpful to differentiate a few cases of ULD or LLD from limb rigidity due to stiff person syndrome. The progressive rigidity due to continuous motor unit activity in agonist and antagonist muscles that characterizes this rare disease may be sometimes difficult to distinguish from dystonia, and diagnosis should rely on the overall clinical picture, instrumental evaluation (presence of anti-GAD or anti-amphiphysin autoantibodies and needle-EMG pattern), and response to immunomodulation and muscle relaxants [20].

Finally, it should be emphasized that the proposed recommendations cannot aid in differentiating the various etiological categories of dystonia, including organic and functional dystonia [21]. As recently highlighted, the possible similarity between physical signs in functional disorders and those that occur in organic illness suggests that, whenever possible, selected investigations should help clinicians to reach a positive diagnosis [21].

To date, diagnostic guidelines have been only proposed for BSP and LD [9, 10]. The recently validated diagnostic guideline for BSP [9] (yielding 93% sensitivity and 95% specificity) was based on an algorithm that was similar to the algorithm characterizing our proposed diagnostic recommendations for CD, OMD, ULD, and LLD. The predicted impact of these diagnostic recommendations in terms of reliability, sensitivity, and specificity is yet to be demonstrated. Nevertheless, recommendations from Figs. 1, 2, and 3 would hopefully facilitate diagnostic approach to CD, OMD, and limb dystonia in the clinical practice of non-expert neurologists. The present data may be the basis to validate future diagnostic guidelines for CD, OMD, ULD, and LLD. Since recommendations were developed by Italian experts, validation should also be performed by international experts in movement disorders.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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