Movement Disorders
CLINICAL PRACTICE
REVIEW

The Phenomenology of Functional (Psychogenic) Dystonia

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ABSTRACT: From the very first descriptions of dystonia, there has been a lack of agreement on the differentiation of organic from functional (psychogenic) dystonia. This lack of agreement has had a significant effect on patients over the years, most particularly in the lack of access to appropriate management, whether for those with organic dystonia diagnosed as having a functional cause or vice versa. However, clinico-genetic advances have led to greater certainty about the phenomenology of organic dystonia and therefore recognition of atypical forms. The diagnosis of functional dystonia rests on recognition of its phenomenology and should not be, as far as possible, a diagnosis of exclusion. Here, we present an overview of the phenomenology of functional dystonia, concentrating on the three main phenotypic presentations: functional cranial dystonia; functional fixed dystonia; and functional paroxysmal dystonia. We hope that this review of phenomenology will aid in the positive diagnosis of functional dystonia and, through this, will lead to more rapid access to appropriate management.

Keywords: functional (psychogenic) dystonia, functional movement disorders.

Functional (psychogenic) movement disorders (FMDs) are part of the spectrum of functional neurological disorders, which are commonly encountered in neurological practice.[1] There has been a clear shift in the clinical approach to these disorders in recent years. The presence of emotional distress or psychopathology is no longer considered of primary diagnostic importance or indeed as a universal etiological factor. Instead, emphasis has been placed on the use of positive clinical diagnostic criteria, supplemented, in some cases, by specific investigational procedures, in order to reach a diagnosis. A major aim of these changes has been to make the diagnosis of a functional neurological disorder a positive diagnosis and not simply a diagnosis of exclusion.

There are well-established clinical and electrophysiological methods to aid in the diagnosis of certain FMDs, in particular, tremor, myoclonus, and gait disturbance.[2] However, the diagnosis of functional dystonia (FD), the second-most common presentation of FMD,[3,4] remains controversial.

One important reason for the continued difficulty in the diagnosis of FD is that the clinical pattern of well-defined organic dystonia has emerged at a later stage than other movement disorders. However, recent advances in genetics have brought increasing certainty to the clinical characteristics of organic dystonia, and reports of large case series of patients with clear FD have helped to make the distinction clearer. Another area of difficulty has been paroxysmal dystonia. Again, here, great progress has taken place in genetics and phenotypic-genotypic correlations aided in defining organic and separating it from functional.

The aim of this review is to synthesize these advances and provide a definitive clinical guide to the phenomenology of FD to aid in the rapid positive diagnosis of this disorder.

History, Definition, and Diagnostic Criteria of functional dystonia

The 20th century witnessed a number of shifts in opinion regarding the nature of dystonic symptoms. Barraquer-Roviralta described, in early 1887, a 37-year-old patient suffering from what would now be recognized as sporadic generalized dystonia in early adulthood.[5] Subsequent reports, including the first familial case descriptions of generalized dystonia,[6] emphasized "hysterical" features.[6,7] Despite Oppenheim's seminal report on dystonia musculorum deformans in 1911, which clearly stated

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the organic nature of dystonia,[8] hysteria was still proposed as a reasonable explanation for dystonia, particularly in its focal forms: "torticollis mental"[9]; "torticollis hystericus"[10]; and writer's cramp[11] (for review, see an earlier work[12]). Reasons for this included the more common prevalence of focal dystonias in women, and the popularity of symbolic interpretations of the phenomenology; blepharospasm was thought to symbolize closing the eyes to the world or torticollis as looking away from a stressful situation.[13] However, as a result of the increasingly disappointing results of psychotherapy[14,15] contrasted to promising outcomes of stereotactic surgical procedures[16] and the development of the first animal models for dystonia,[17–19] the concept of nonorganic dystonia was progressively left behind. This was aided by David Marsden's treatise on adult-onset dystonia, which outlined reasons why focal dystonias, the "formes frustes" of generalized dystonia, had been mistakenly regarded as nonorganic (Table 1).[20] As an example of the shift away from considering any forms of dystonia as nonorganic, in Fahn and Eldridge's etiological classification of dystonic conditions published in 1976, "psychological dystonia" was placed at the end of their list of causes of dystonia "for the sake of completeness."[21] They considered it extremely rare, "if existent at all," and stressed the damage done to patients and families of the false attribution of dystonia to psychological causes.[22] It was during that period that the third version of the *Diagnostic and Statistical Manual of Mental Disorders* (DMS-III) replaced the diagnosis of *hysterical neurosis* with *dissociation* and *conversion*.[23,24]

However, an unexpected consequence of the more precise definition of the typical (or organic) forms of dystonia was the increasing visibility of the atypical forms of dystonia and the possibility that these might reflect nonorganic dystonia. Thus, in 1978 and then in 1983, Fahn and colleagues reported on the first patients of the modern dystonia era with nonorganic dystonia.[25,26] Consequently, they published a systematic report on the characteristics of 21 patients with "psychogenic" dystonia based on diagnostic criteria that the authors proposed (Table 2).[27] These emphasized primarily neurological, rather than psychiatric, diagnostic parameters[27] and introduced a gradation of diagnostic certainty. Inconsistency over time and phenomenological incongruity from classical movement disorders constituted main diagnostic criteria.

Based on these criteria, Lang reported on 18 cases with FD.[28] This report underscored the diagnostic importance of clinical incongruities and inconsistencies, but also commented on the absence of obvious predisposing psychological factors.[28] In fact, associated psychosocial factors (including conflicts or stressors), necessary for the diagnosis of conversion disorder according to DSM-IV,[29] were not required in the original Fahn and Williams criteria, which were later applied to all FMDs.[30] This acknowledgment that psychological factors may not be as etiologically or diagnostically relevant as once thought[31,32] has recently led to their relegation from diagnostic criteria of *conversion disorder* (or *now functional neurological symptom disorder*) in DSM-IV[29] to "accompanying features" in DSM-5[33]

Although revisions of the Fahn and Williams diagnostic criteria have been proposed for all FMDs,[34,35] the former still constitute the gold standard in diagnosing FD. However, diagnostic agreement for FMD by applying these criteria is poor[36] and only one fifth of movement disorders clinicians rely solely on them in the diagnosis of FMD.[37] However, for the vast majority, clinical incongruities serve as useful diagnostic features.[37] Hence, it is the phenomenological characterization of FD and its clinical distinction from organic dystonia that bears the most significant implications for diagnosis.

Phenotypic Characteristics of FD

General Points

Certain general principles common to FMD also apply to FD. Usually, symptoms appear suddenly and are often precipitated by physical and/or emotional events.[26–28,38–41] There is marked

TABLE 1 Characteristics of focal dystonic conditions that have contributed in their mislabeling as psychogenic[20]

1.	The bizarre nature of the dyskinesias
2.	Their appearance frequently only on certain actions, other motor acts employing the same muscles being carried out normally
3.	Their relief by certain inexplicable trick actions
4.	Their exquisite sensitivity to social and mental stress
5.	The failure so far to find any anatomical, physiological, or biochemical abnormality in any of these conditions
6.	The belief that such patients show overt psychiatric disturbance
7.	A psychopathological interpretation of the significance of, for example, eye closure or neck turning

TABLE 2 Proposed diagnostic criteria by Fahn and Williams[27] for the diagnosis of functional dystonia

Documented[a]
Persistent relief by psychotherapy, psychological suggestion, including physiotherapy, or by administration of placebos or the patient was witnessed free of symptoms when believing being unobserved
Clinically established[a]
Dystonia is inconsistent over time or is incongruent with classical dystonia and at least one of the following:
1. Presence of other psychogenic signs
2. Multiple somatizations
3. Obvious psychiatric disturbance is present
Probable
Symptoms are inconsistent or incongruent with classical dystonia or psychogenic signs or somatizations are present.
Possible
Dystonic movements are consistent and congruent for organic dystonia, but there is an obvious emotional disturbance.

[a]The revised version of their criteria merged the definition of both categories as "clinically definite."[30]

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variability in their phenomenology, progression, and duration. Spontaneous, iatrogenic, or life-event–related remissions and recurrences are common. In many cases, the onset age is unusual for the particular phenotype. Also, as with other FMDs, FD co-occurs with additional functional phenomena. Hence, the typical "company" that FD keeps is functional tremor, myoclonus, and effortful nonparkinsonian slowing of movements, but also additional nonorganic neurologic signs and multiple somatizations.[28,30,38,39,41–44] Suggestibility and placebo or atypical response to medication are commonly observed (see Video 1).[27,28,41] However, neither illness duration, symptom severity, nor previous attempted treatments help in discerning functional from organic.[28] Although a positive family history usually denotes organic disorders, FMD may also affect more than one family members.[45] Finally, FD may coexist with organic movement disorders or other neurological illness (functional overlay).[27,30,42] In these cases, they usually affect the same or adjacent body parts.[3,46]

However, in addition to these general characteristics, systematic clinical observations have led to the delineation of three distinct dystonic presentations likely to be functional. These include cranial, fixed, and paroxysmal dystonic phenotypes.

Functional Cranial Dystonia

FD predominantly involving the face occurs in approximately 16% of all FMDs.[43] Females are most commonly affected.[3,30,43,47,48] Symptoms usually appear between the fourth and sixth decades,[43] but onset in childhood is possible.[49] Symptoms emerge abruptly. The lower face is most commonly affected. Unilateral or asymmetric involvement is common. Symptoms are most often paroxysmal and consist of tonic spasms with brief periods of normal facial muscle activity in between.[30,43,47,48] However, sustained fixed posturing for several days is reported.[43] Unilateral downward lip pulling, which can be reported to be painful with adjacent platysma contractions, is very common (see Video 2, Segment A).[43] In many cases, speech is normal; however, difficulties may be present. In the vast majority of patients, swallowing remains unaffected.[43]

When the eyes are predominantly affected ("functional blepharospasm"), again abrupt and asymmetric symptom onset with constant–tonic–eye closure (see Video 2, Segments B–D) are common.[3,43,47] However, patients may also show continuous bilateral eye closure without prominent muscle activity ("psychogenic pseudoptosis"[50] and "hysterical blepharospasm"[51–54]; see Video 2, Segment E). Suggestibility and unusual cues hint toward a functional cause. In asymmetric presentations where intermittent spasms affect the entire side of the face mimicking hemifacial spasm, the lack of synchronicity between lower and upper facial muscles and occasional bilateral tonic contractions of the lower face with unilateral spasm of the upper face are useful diagnostic clues.[43] Typically, spasms are much more prolonged than the very brief electric shock-like spasms of typical hemifacial spasm. Also, the absence of the "other Babinski sign" (synchronous contraction of the orbicularis oculi and frontalis muscles leading to eye closure with ipsilateral eyebrow elevation)[55] aids in diagnosis (see Video 2, Segments B–D). As with other FMDs, *immediate* therapeutic response to botulinum toxin, if present, further contributes in recognizing a functional cause.[56]

Functional Fixed Dystonia

Fixed dystonia (i.e., reduced joint mobility resulting from dystonic posturing) is not a common phenotypic presentation of primary dystonia[57] (including idiopathic and/or inherited forms of dystonia, according to the new dystonia classification[58]). It may, however, be encountered in secondary dystonic conditions in advanced disease stages.[59] It also differs from mobile dystonia, in that it is painful and does not improve to sensory tricks. Patients with fixed dystonia do not exhibit overflow dystonia.[38,39] Fixed dystonia affects more females and is often elicited by minor traumas and commonly co-occurs with pain, which, in some cases, fulfills criteria for chronic regional pain syndrome type I (CRPS-I).[38,39,44] The latter denotes a syndrome consisting of autonomic (e.g., vasomotor, sudomotor, and trophic), sensory (e.g., pain or hyperalgesia), and motor (e.g., decreased range of motion, weakness, and posturing) symptoms.[60] Different from type II CRPS, type I develops in the absence of peripheral nerve injury.[61]

In contrast to primary (and mobile) dystonia, fixed dystonia commonly develops rapidly (overnight or in a few days) after a minor precipitating event.[27,28,38,39] It usually presents as resting dystonia at onset[28] and usually affects the lower limbs,[38,39] but can also manifest in the hands or neck/shoulders. It typically consists of foot inversion with plantar flexion and curling of toes (see Video 3, Segment A). In the hands, there is typically metacarpo- and/or interphalangeal flexion, mostly of the fourth and fifth fingers, with the thumb least or not functionally affected (see Video 3, Segments B–E).[39] In most cases, pain is a major complaint. For the neck, in the absence of severe trauma-induced musculoskeletal injuries, tonic dystonic posturing with ipsilateral shoulder elevation and prominent pain has been described.[62–65] A disappointing long-term response to local botulinum toxin injections is characteristic, and the term "post-traumatic painful torticollis" has been proposed.[62,63] Dystonic posturing can spread to both ipsi- and contralateral limbs or may co-occur with other functional motor symptoms. Giveway weakness, functional jerks, and tremor have been noted in affected and nonaffected body parts.[27,28,38,39,44,63]

In the largest study to date to assess the features of fixed dystonia in 103 patients (41 of which were prospectively examined), "psychogenic signs" had been documented for 33% of all patients, but were even more common in the prospectively examined group (46%).[39] In fact, 90% of patients of the latter group were found to fulfill different levels of the Fahn and Williams criteria for FD, with 37% (15 of 41 patients) receiving a diagnosis of either documented or clinically established FD.[39] Further, the co-occurrence of affective, somatization, and dissociative disorders was significantly higher for patients with fixed dystonia, when compared to primary dystonia controls.[39] Although pain was a main characteristic in nearly all patients,

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less than half of the prospectively examined subgroup exhibited cardinal features of CRPS and only 20% fulfilled diagnostic criteria.[39] The presence of CRPS has been shown to be the main predictor of poor outcome for fixed dystonia.[44]

A great controversy surrounds the etiological nature of fixed dystonia with or without CRPS-I.[66] However, for at least a subset of patients presenting with fixed dystonia in the absence of secondary causes, aforementioned syndromic characteristics should prompt the consideration of FD. Spontaneous symptom improvement upon conflict resolution in some cases, good therapeutic effects of multidisciplinary treatment approaches with a particular focus on cognitive-behavioral therapy, as well as immediate dramatic responses to placebo treatments further support this notion.[27,28,39,44,56]

The management of the severe pain reported by patients with fixed dystonia, which is exacerbated upon tactile stimulation or light movement, is challenging.[39,44] The severity of symptoms and uncertainty regarding the diagnosis often lead to invasive procedures, including limb amputation, however with poor outcome.[44] Of note, seeking of limb amputation is only encountered extremely rarely in other dystonic conditions,[67,68] but has been reported in CRPS-I.[69–71] Thus, the possibility has been raised that patients with fixed dystonia and CRPS-I who seek limb amputation might have deficits in their body schema perception as part of the body identity integrity disorder spectrum.[67]

Functional Paroxysmal Dystonia

Although paroxysmal FD has been well recognized and constitutes common clinical presentation, its characteristics had only been rarely highlighted in recent literature.[72–75] When it predominantly affects the extremities and/or trunk and on a background of unrevealing neurophysiologic, imaging, and laboratory examinations, its differentiation from other paroxysmal disorders, in particular, the primary paroxysmal dyskinesias, may be challenging.[41,72]

Fahn and Williams described 7 of 21 FD patients with paroxysmal symptoms, 4 of which had documented and 3 established FD.[27] They pointed out the high prevalence of FD in patients with paroxysmal dystonia and subsequently indicated that variable and inconsistent nonorganic startle responses often trigger paroxysmal episodes of FD (see Video 4, Segment A).[30] Increased suggestibility and the unusual combination of additional abnormal movements in addition to dystonic posturing, which are not observed in classic presentations of primary paroxysmal dyskinesias, typify these patients (see Video 4, Segment B).[28]

However, it was recent advances in genetic characterization of the three main primary forms of paroxysmal dyskinesias (paroxysmal kinesigenic dyskinesia [PKD], paroxysmal nonkinesigenic dyskinesia [PNKD], and paroxysmal exercised-induced dyskinesia [PED]) that have allowed their typical phenotypic characteristics to be recognized and distinguished from atypical forms.[76–88] We recently reported on the characteristics of a large case series of patients with functional paroxysmal movement disorders, including patients with functional paroxysmal dystonia, and compared them to proposed diagnostic criteria of PKD, PNKD, and PED.[41] In this regard, one of the most important pointers is that typically all three primary forms present very early in life, during the first or second decade. However, in functional paroxysmal dystonia, symptoms usually appear much later on.[27,28,30,41]

TABLE 3 Clinical hints of functional paroxysmal dystonia adapted from Ganos et al.[41]

Adult age of onset
Presence of paroxysmal tremor
High phenomenological variability of episodes
Precipitation of attacks or increase in symptom severity during examination
Atypical and variable duration of attacks
Presence of multiple atypical triggers
Altered level of responsiveness
Presence of atypical precipitating factors
Presence of unusual relieving maneuvers
Additional psychogenic physical signs and/or medically unexplained somatic symptoms
Atypical response to medication

Another important aspect is the great variability between attacks in terms of duration and phenomenology. Although in primary paroxysmal dyskinesias between-subject variability exists in attack duration, great intraindividual variability is uncommon. For example, in PKD, attacks typically last less than 1 minute and only rarely do they vary by more than a few minutes in a single patient.[76,83,89] In contrast, attacks in functional paroxysmal dystonia may differ greatly in duration in individual patients (e.g., between seconds and hours/days).[41] Furthermore, in functional paroxysmal disorders, the predominant movement disorder may shift in nature between attacks. For example, paroxysmal tremor has been noted to occur at times in patients who have attacks of dystonia at other times, strongly hinting at nonorganicity.[41] A complete list of diagnostic red flags hinting toward a functional disorder is given in Table 3.

Neurophysiology as an Aid for Diagnosis of Functional Dystonia

A number of different electrophysiological techniques have been applied to patients with organic and presumed FD. These have shown some similarities, but also differences. From the clinical perspective, it would be of great use to have an electrophysiological test that reliably distinguished between organic and FD. However, despite the work that has been done in this area to date, there are no sufficiently robust tests available that could be used to provide what Gupta and Lang have proposed as a "laboratory-supported" level of diagnostic certainty for functional dystonia.[34]

The R2 component of the blink reflex recovery cycle has been found to be abnormally enhanced in patients with organic blepharospasm.[90] This differs from patients with atypical (or functional) blepharospasm, where it was found to be normal,[48] and separated patients with functional and organic blepharospasm fairly successfully on an individual patient basis. One study has provided limited evidence that the reduction in postexcitatory

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inhibition after transcranial magnetic stimulation in the affected facial side in patients with hemifacial spasm during spasms and the subsequent prolongation after spasms is not observed in patients with functional spasms and healthy controls,[91] but this awaits confirmation in a larger group of patients.

Other electrophysiological measures, including cortical and spinal cord inhibition (short intracortical inhibition [SICI] and reciprocal inhibition), assessments of associative plasticity, and sensory temporal discrimination, have been performed in patients with organic and fixed dystonia.[92–95] Similarities and differences have been found, but such data are hard to interpret from a pathophysiological point of view and do not as yet form a viable basis for a clinical diagnostic test. Measures such as SICI are vulnerable to interference from the effects of attention toward the limb as well as by the underlying personality disorder.[96,97] Temporal discrimination testing relies on self-report, and therefore it is difficult to be certain that the abnormalities reported in fixed dystonia are caused by the same underlying mechanism as the abnormalities reported in organic dystonia. In any event, the intraindividual variability of such measures, even in patients with organic dystonia, as well as the technical demands of some of the tests, makes it unlikely that they will be useful as diagnostic measures.

Management and Prognosis

Systematic data regarding the efficacy of available treatments in different forms of FD is limited. However, extrapolating knowledge from other functional neurological syndromes to FD might prove helpful.[42] In this regard, effective communication of the diagnosis seems of particular importance. Although this applies to any medical condition, there are reasons to suspect that the therapeutic benefit of effective communication is particularly high in this patient group.

For fixed dystonia, reestablishment of movement as soon as possible is of paramount importance. Specialist physiotherapy input, perhaps in an in-patient rehabilitation setting for severely affected patients, therefore has a clear rationale. Given the presence of pain in many patients with fixed dystonia, hollistic pain management is important, focusing on minimizing medications and concentrating on both physical and cognitive behavioral pain management techniques. A multidisciplinary treatment approach may include both psychological interventions (e.g., cognitive-behavioral treatment and psychodynamic psychotherapy) and physical rehabilitation.[39,98,99] Any delay between symptom onset and treatment may lead to poor outcome, and in fixed dystonia, this might be complicated by intractable contractures.[39,100] Of note, the use of generalized anesthesia has been proven helpful in discerning whether such contractures are indeed present.[101] In one study of 35 patients with fixed dystonia and a mean follow-up period of 7.6 years, approximately half had no change in symptoms, less than one quarter experienced symptom improvement, and one third further deteriorated.[44]

Evidence suggests that paroxysmal presentations of FD might have a more favorable outcome.[41] Here, treatment techniques that are used successfully for nonepileptic attacks, including specific cognitive behavioral techniques that focus on methods of preventing or terminating seizures, seem a rational route for treatment.

TABLE 4 Summary of main clinical characteristics of common presentations of FD

	FD		
Type	Cranial dystonia	Fixed dystonia	Paroxysmal dystonia
Age at onset[a]	Fourth to sixth decade	Second to fourth decade	Third to sixth decade
Gender	F ≫ M	F ≫ M	F > M
Common phenotypic characteristics	Unilateral tonic downward lip pulling with ipsilateral platysma involvement	Lower limbs > upper limbs > neck/shoulder; fixed plantar flexion and inversion with toe curling; carpal flexion with prominent clawing of fourth and fifth fingers; tonic dystonic posturing of neck (latero/torticollis) with ipsilateral shoulder elevation	Attacks with variable phenomenology and duration; presence of paroxysmal episodes on a background of continual dystonic posturing; alterations of responsiveness during attacks possible; presence of atypical triggers and relieving maneuvers
Additional features	"Other Babinski sign"; asynchronous spasms of lower and upper facial muscles; bilateral tonic contractions of the lower face with unilateral spasm of the upper face	CRPS-I common; spread to other extremities possible; absence of sensory tricks or overflow dystonia	Frequency and severity increase during examination; presence of additional movement disorders during paroxysmal episodes; atypical response to medication
Presence of pain	Common	Prominent	Common
Neurophysiology	Blink reflex recovery cycle and postexcitatory inhibition normal	Normal sensorimotor plasticity	—
Reference	43, 48, 91	27, 28, 38, 39, 63, 95	27, 28, 41

[a]Cases with onset in childhood/adolescence or older than sixth decade possible.
CRPS-I, Chronic regional pain syndrome type I; F, female; M, male.

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Oral medications for the treatment of dystonic symptoms are not recommended, because responses are variable and inconsistent over time. Comorbidities, such as depression or anxiety, should be adequately addressed.[102] Invasive therapeutic procedures should be generally avoided because these appear likely to cause worsening of symptoms.[39,42,67,103]

In our experience, for some patients who are reluctant or unwilling to participate in multidisciplinary treatment, placebo interventions (e.g., small amounts of botulinum toxin to the affected sites; see Video 1, Segments C and D) can prove helpful.[41,56] However, lack of systematic data and ethical considerations complicate such approaches.[104]

Conclusions

We have based the distinction of FD from organic dystonia on the phenotypic patterns observed primarily in patients with primary genetically determined dystonia (main characteristics of main FD presentations summarized in Table 4). However, cranial dystonia, fixed limb dystonia, and paroxysmal dystonia can all also arise from secondary causes, and in such secondary forms, "atypical" presentations are possible. It is important therefore to consider whether the patient might be presenting with an unusual secondary form of dystonia, and therefore (limited) investigations are often appropriate. However, a careful, open-minded approach to diagnosis (which includes the consideration of functional overlay over an organic dystonia) should not be allowed to cause diagnostic paralysis where more and more tests are requested to rule out increasingly unlikely organic causes. This is where the search for positive clinical signs is of such diagnostic importance.

There will always be patients where diagnosis is difficult, and though the physician suspects the diagnosis to be that of FD, the level of certainty is not high. Perhaps because of the feeling that it is worse to diagnose a patient with an organic movement disorder as having a functional disorder than vice versa, it is our experience that such patients are often left to drift without a suitable management plan. However, in such cases, an honest discussion with the patient of the diagnostic possibilities, the presentation of the diagnosis of FD as a real diagnosis with a particular avenue of treatment associated with it, enrollment of the patient in a true rehabilitation approach to symptoms with both physical and suitable cognitive treatments, and a willingness to reconsider the diagnosis over time should new symptoms emerge can be successful.

Author Roles

(1) Research Project: A. Conception, B. Organization, C. Execution; (2) Statistical Analysis: A. Design, B. Execution, C. Review and Critique; (3) Manuscript: A. Writing of the First Draft, B. Review and Critique.

C.G.: 1A, 1B, 1C, 3A, 3B

M.E.: 1A, 1B, 1C, 3A, 3B

K.B.: 1A, 1B, 1C, 3A, 3B

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Supporting Information

A video accompanying this article is available in the supporting information here:

Video 1. Placebo treatment responses. (A) Twenty-three-year-old female with a 3-year history of generalized spasms resembling myoclonus-dystonia, during which she is unable to stand and walk. (B) Immediate (<2 minutes; placebo) response to alcohol consumption with significant reduction of hyperkinetic movements. (C) Twenty-two-year-old female with a 4-year history of painful fixed retrocollis. This emerged 1 day after a chiropractic treatment for her neck, which had left her in severe pain. (D) Immediate (<5 minutes; placebo) response to botulinum toxin injected bilaterally to the semispinalis capitis muscles.

Video 2. Functional cranial dystonia. (A) Twenty-six-year-old female with a 7-year history of intermittent spasms of the lower face causing the left angle of the mouth to be depressed with ipsilateral platysma activity. (B) Forty-three-year-old male with a 10-year history of intermittent tonic spasms of predominantly the left side of the face lasting from a few minutes to 2 hours. The absence of the "other Babinski sign" can be noticed. (C) Bilateral involvement of the upper face and unilateral lower face involvement during another attack in the same patient. (D) Forty-nine-year-old female with a 4-year history of intermittent tonic spasms of the right side of the face with occasional bilateral involvement. Again, the absence of the "other Babinski sign" is notable. (E) Fifty-three-year-old female with an approximate 3-year history of difficulties in opening her eyes. Her first symptoms were increased eye blinking and soreness of the eyes. This has led to constant (flaccid) eye closure (pseudoptosis), as demonstrated here.

Video 3. Functional fixed dystonia. (A) Fifty-seven-year-old female with bilateral fixed foot dystonia with onset at the age of 56. This developed after two episodes of "collapse" and "unresponsiveness" of unclear etiology. She also experiences regular

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"whole body spasms" with associated eye closure that can last between 1 hour and 1 day. As a result of the dystonic posturing of her feet, she is functionally unable to walk and thus wheelchair bound. (B) Sixty-year-old female with a 12-year history of fixed dystonia of both hands affecting the medial three fingers. The thumb and index fingers are unaffected. Attempts to passively extend the affected fingers lead to an increase in resistance. (C) Fixed dystonia of all four fingers sparing the thumb in a 20-year-old female with inability to extend them actively or passively. (D) Fifty-eight-year-old female with a 3-year history of fixed dystonic posturing of the three medial finger of her left hand, which developed over a period of a few weeks. The thumb and index finger remain unaffected. Additional symptoms over this period included an 8-month period of aphonia, difficulties in catching her breath, and feeling a lump at the bottom of her throat. (E) Examination of the same patient under general anesthetic revealed damage to the skin of the hand, resulting from complete flexion of the three medial fingers. An approximate 50% contracture in flexion at the metacarpo- and interphalangeal joints of the medial three fingers was noted.

Video 4. Functional paroxysmal dystonia. (A) Twenty-one-year-old female who was admitted for investigation of paroxysmal attacks. The symptoms had appeared a few years ago in the context of severe chronic back pain. They consisted of short-lasting uncontrolled jerking of the right side of the body, also elicited by tapping of the reflexes of the same side. Occasional speech difficulties and drowsiness occurred as well. (B) Forty-five-year-old female with paroxysmal attacks triggered by physical activity (e.g., walking). They consist of generalized dystonic posturing, but also prominent grunting. Of note, the demonstrated episode subsides as soon as the patient is able to rest.

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