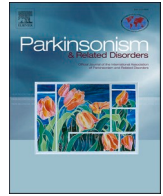


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# Parkinsonism and Related Disorders

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Short communication

## Functional ataxia in a specialized ataxia center

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### ABSTRACT

**Background:** Functional gait is a disorder of ambulation and balance internally inconsistent and incongruent with the phenotypic spectrum of neurological gait disorders. **Objectives:** This paper aims to clinically characterize patients with functional ataxia. **Methods:** Patients with functional ataxia were analyzed out of 1350 patients in Ataxia Unit of the Federal University of São Paulo circa 2008 to 2022. **Results:** Thirteen patients (1 %) presented with functional ataxia; all female, with a median age of 34.8 years. Six (46.2 %) had psychiatric comorbidities and 7 (53.8 %) endorsed a trigger. Diagnostic features included variable base and stride (100 %), “huffing and puffing” (30.7 %), knee-buckling (30.7 %), uneconomic posturing (38.5 %), tightrope walking (23 %), and trembling gait (15.4 %). Remarkably, no falls were reported in any case. 53.8 % recovered fully or partially, despite no treatment. **Conclusions:** Variability of base and stride are universal features of functional ataxia, yet falls are inconspicuous. Functional Ataxia is rare even in a specialized ataxia center.

### 1. Introduction

Normal gait and balance require precise control of limb movement, posture, and tone - a complex process that involves the entire nervous system [1,2]. Ataxia is defined as irregular and uncoordinated movements [1], caused by disruptions in a circuitry involving basal ganglia, cerebellum, cerebral cortex, proprioception, and peripheral sensory-motor pathways [3,4]. It may be a symptom of a multisystemic disorder or isolated phenomena [3,4].

Functional disorders are manifestations in which the primary pathophysiological processes are modifications in the brain function, not in its structure [5]. This paper aims to report a series of patients with a gait disorder characterized as functional ataxia based on a phenomenological assessment of inconsistency and incongruence with other neurological gait disorders.

### 2. Methods

From 2008 to 2023, a total of 1350 patients were evaluated in the Ataxia Unit of the Federal University of São Paulo. Different etiologies of

cerebellar ataxias, genetic and sporadic, were described. Thirteen patients presented with a functional gait pattern, characterized by the group as functional ataxia. Diagnosis was based on phenomenology and detailed examination, as well as exclusion of organic causes through ancillary investigation. Four movement disorders experts were invited to evaluate gait and characterize the main features of functional ataxia (JLP, PMF, OGB, AJE). Features that allow this categorization are detailed in the results and discussion. All 13 cases are comprised in Video 1 (Cases 1-7) and Video 2 (Cases 8-13) and summarized in Table 1. More detailed individual description follows.

**Case 1.** A 42-year-old woman presented with a buckling knee, broad-based, high stepping, cautious gait, with an inconsistent pattern when prompted to walk backwards, developing a trembling gait associated to uneconomic posturing. She had a negative Romberg test and a normal neurological examination, with anxiety and systemic lupus erythematosus as co-morbidities. Spontaneous recover happened in 10 days, and ancillary investigation was unremarkable (Video 1).

**Case 2.** A 25-year-old woman presented with two days history of gait abnormality characterized by a broad base and dragging gait, with knee

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**Table 1**

Description of 13 patients with functional ataxia.

	Gender	Age	Age of onset	Duration	Recovery	Phenomenology	Psychiatric changes	Additional features	Trigger
1	Female	42	42	5 days	yes (10 days)	buckling gait	Anxiety	Systemic Lupus Erythematosus	unknown
2	Female	25	25	2 days	yes (5 days)	dragging gait/inconsistency in walking pattern/moments of improvement	none	None	Unknown
3	Female	29	28	1 year	no	Waddling gait veering from side to side/Tightrope walking	none	None	viral infection + lumbar puncture
4	Female	43	42	1 year	no	changes in gait pattern when turn around	Depression	Depression (previous)	Unknown
5	Female	62	62	1 month	yes - a day after evaluation	Seeking for a far support	none	Hypertension	Unknown
6	Female	31	31	3 months	yes (a month)	Waddling gait	Anxiety	None	Family with SCA 3
7	Female	19	19	2 weeks	yes (one week)	exaggerated Truncal instability without falls/veering gait	None	involuntary movements	Recent hospitalization for sudden dyspnoea
8	Female	54	45	9 years	no	Slowing gait	Anxiety	Convergence spasm + urinary retention	Unknown
9	Female	29	28	1 year	no	Truncal instability without falls	No	La Belle Indifférence	Unknown
10	Female	45	45	6 months	no	slowness and huffing and puffing	No	Fatigue	Dengue infection 2 weeks before symptoms onset
11	Female	15	15	3 weeks	unknown	Tightrope walking/seek for a far support	None	None	IM metamazole
12	Female	36	36	months	Yes (still recovering)	Broad-base, instability without falls, unspecific trunk motion	anxiety	Involuntary movements	Medication overdose
13	Female	23	23		No	trembling gait and "huffing and puffing"	Skin lesions	None	Skin lesions.

extension, somewhat hinting a spastic quality, without a circumduction tendency, with periods of improvement. She had no weakness, spasticity, dysdiadochokinesis or dysmetria and could run normally, with no falls. She spontaneously improved in 5 days (Video 1).

**Case 3.** A 28-year-old woman presented with a 1-year history of waddling, veering from side to side, broad-based gait with hesitation, no falling history, and "huffing and puffing" at each step after a viral infection and lumbar puncture. Remarkably, she could perform tandem gait regardless of support and sometimes presented a tightrope motion. Neurological and complementary investigation was unremarkable (Video 1).

**Case 4.** A 43-year-old woman developed a gait pattern characterized by hesitation, inconsistent difficulty on bending her knees and a marked change of pattern while turning around, as well as "huffing and puffing". Neurological examination showed no abnormalities. Complementary investigation was normal (Video 1).

**Case 5.** A 62-year-old woman presented with 1 month of gait impairment markedly while turning. Neurological exam was unremarkable. She presented with difficulty on rising feet, reduced arm swaying, as well as broadening base, with constant far support seeking and need for aid while turning and, regardless, had no falls. Recovery was observed a day after medical evaluation (Video 1).

**Case 6.** A 31-year-old woman presented with a three-month history of a narrow based, waddling gait, with knee-buckling tendency and a stomping quality, recovering a month after medical evaluation, with no falls. Neurological and complementary examination was unremarkable. She had a family history of Spinocerebellar Ataxia type 3, which rendered a diagnosis of anxiety, before clinical manifestations (Video 1).

**Case 7.** A 19-year-old woman presented with a two-week history of impaired gait associated to abnormal trunk and limbs posturing and instability, without falls, which began after a recent hospitalization because of sudden-onset dyspnea. She had gradual and spontaneous improvement in a week after first medical examination, developing a sometimes broad, sometimes narrow based gait, with instability on tandem, posturing abnormalities or difficulty on turning. Neurological examination showed initially an intention tremor, worse on the left arm,

with seemingly bilateral dysmetria. Ancillary testing was normal (Video 1).

**Case 8.** A 54-year-old woman presented at age 45 with disequilibrium and urinary retention. At gaze examination, she showed convergence spasm, at all directions, particularly on up and downgaze. Gait was slowed, with variable base, with no difficulty on initiation, turning or abnormalities at arm swaying. She had anxiety disorder as a comorbidity, with no worsening or recovery until this date and no remarkable history of falls. Complementary investigation showed no abnormalities (Video 2).

**Case 9.** A 29-year-old woman presented with one year history of change on her walking, with no history of falls. She presented with a sometimes narrow, sometimes broad-based gait, uneconomic both trunk and hip swaying movements, as well as reduced arm sway velocity. Remarkably, she did not extend her knees while walking. When given the walker, her initiation and balance became poorer, being unable to benefit from the device. A marked feature was "La Belle Indifférence" (Video 2). Additionally, despite unbalanced, she had no falls.

**Case 10.** A 45-year-old woman presented with a 6-month balance complaint. At neurological examination, she had a narrow-based, unsteady gait, with no preferential side of deviation and need of bilateral aid for walking. Remarkably, she had huffing and puffing, fatigue, no falling, and a dengue virus infection 2 weeks prior to symptom onset (Video 2).

**Case 11.** A 15-year-old woman presented with a 3-month history of a gait abnormality characterized by excessive widening of step length and height – with tightrope walking quality, compensatory trunk, hip, and upper limbs motion to maintain optimal balance, always seeking for a far support. She had received intramuscular metamazole immediately prior to clinical onset and had no history of falls. Complementary investigation and additional neurological features were normal (Video 2).

**Case 12.** A 36-year-old woman presented with difficulty walking after an overdose. In a year she recovered partially, being able to walk normally while singing sacred chants and while holding a water cup in one hand. Examination showed a wide length of step and overly extended

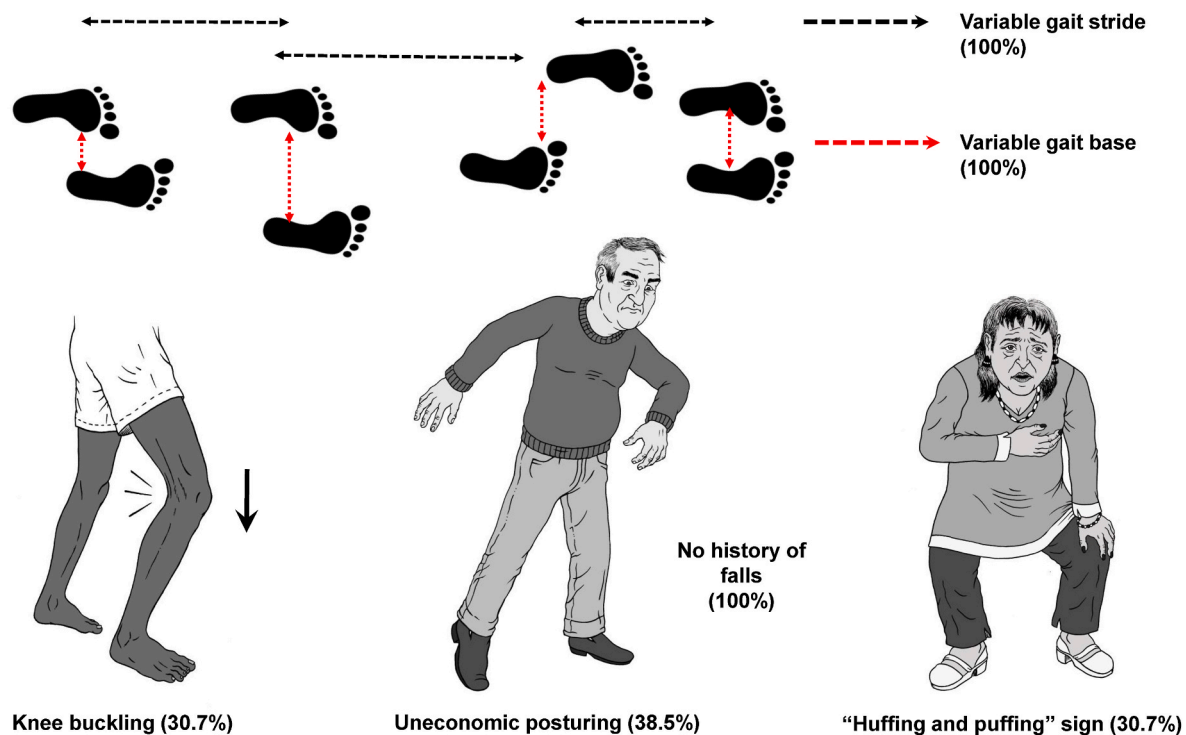


Fig. 1. Main Features of patients with functional ataxia.

knees in absence of spasticity or pyramidal signs, need of unilateral aid for walking and overstretched motion of the left arm, as well as an uneconomic trunk motion. No falls were reported (Video 2).

**Case 13.** A 23-year-old woman presented with onset of a gait disturbance after the appearance of skin lesions on lower limbs (Video 2). At examination, she had a trembling gait and “huffing and puffing”, with no falls reported. Ancillary exams, including somatosensory evoked potential were normal.

Supplementary video related to this article can be found at <https://doi.org/10.1016/j.parkreldis.2024.106006>

### 3. Results

Out of the 1350 patients evaluated, 13 (1 %) presented with features that supported the diagnosis of a functional ataxia, a smaller number than expected, but justifiable since many patients may be in other movement disorder centers. All patients were female, with a median age of 34.84 years (min 15, max 62) and of onset of 33.92 years (min 15, max 62). Six (46,2 %) patients had psychiatric comorbidities (5 anxiety and 1 depression) and 7 (53,8 %) had a recognizable trigger. 7 out of 13 patients had full or partial recovery (Table 1).

Most frequent features that support the characterization of functional ataxia (Fig. 1), were variable gait and stride - universally present - with additional features variably present: marked uneconomic posturing (38.5 %), knee buckling (30.7 %), and “huffing and puffing” (30.7 %). Other findings were dragging feet (15.4 %), tightrope walking (23 %), waxing and waning (15.4 %), trembling gait (15.4 %), overextension of knees despite no spasticity (23 %).

Remarkably, no patient had history of falls, which is extremely uncommon in patients with other neurological gait disorders - usually, patients and their families tend to seek medical assistance exactly due to excessive falling.

### 4. Discussion

Gait disorders are a major source of disability, morbidity, and

mortality, especially in the elderly, and might be neurological or non-neurological in origin [1,2].

Functional disorders, previously termed “psychogenic”, are diagnosed by features that fulfill criteria of inconsistency and incongruence [5–12]. Historical features, while suggestive (sudden onset, history of sexual abuse and/or major emotional stress, childhood adversity, etc) are neither necessary nor sufficient for the diagnosis.

Epidemiological studies suggest a female preponderance [6–9] (approximately 1:5), which was observed in our series (100 % women). There may be a genetic predisposition, with *TPH2* and *OXTR* implied, but further studies are required in this regard [6].

We have characterized these gait abnormalities as functional ataxia, because they fulfill such features of incongruence with other neurological disorders such as knee buckling, tightrope walking, trembling/shaking quality and excessive slowness [6–10]. Additional helpful signs were the excessive demonstration of effort during gait – namely the “huffing and puffing” phenomena (as seen in patients 3, 4, 10 and 13), a limited gait with incongruent dystonia [8,10] (e.g., fixed plantar flexion sign), as well as truncal jerks [6–10], observed in patients 1, 7, 9,11 and 12.

Most patients had no abnormalities in their neurological examination, and, when present, those were incongruent - dysmetria in patient 7 and convergence spasm in patient 8. Other findings that suggested a functional origin were dragging gait (patients 2 and 8), tightrope walking (patients 3, 6, 11), waxing and waning (patients 4, 9), trembling gait (patients 1 and 13) and overextension of knees despite no spasticity (patients 2, 4 and 12).

Outcome in functional movement disorders is variable [10–12]. In our series, patients that showed full recovery had a disease duration of less than six months. Treatment strategies include mainly psychotherapy, relaxation and stress management techniques, pharmacotherapy, and motor rehabilitation [11].

The main limitation of this study is the small sample size – it is possible that we may have missed additional functional ataxia phenotypes with a larger sample, using a multicenter approach. Another limitation is the absence of videos of patients after recovery, when happened.

Finally, functional disorders, initially mischaracterized as diagnoses of exclusion, are recognized as diagnoses of inclusion through detailed assessment of phenomenology. Hereby, we highlight examination features that may more commonly distinguishing functional ataxia from other gait disorders, including prominently variable base and stride, and absence of falls despite uneconomic posturing.

#### Declaration of competing interest

The authors declare the following financial interests/personal relationships which may be considered as potential competing interests:

Alberto J Espay reports a relationship with National Institutes of Health that includes: funding grants. Alberto J Espay reports a relationship with The Michael J Fox Foundation that includes: funding grants. Alberto J Espay reports a relationship with NeuroDerm Ltd that includes: consulting or advisory. Alberto J Espay reports a relationship with Amneal Pharmaceuticals LLC that includes: consulting or advisory and speaking and lecture fees. Alberto J Espay reports a relationship with ACADIA Pharmaceuticals Inc that includes: consulting or advisory. Alberto J Espay reports a relationship with Avion Pharmaceuticals that includes: consulting or advisory and speaking and lecture fees. Alberto J Espay reports a relationship with Acorda Therapeutics Inc that includes: consulting or advisory. Alberto J Espay reports a relationship with Kyowa Kirin Inc that includes: consulting or advisory. Alberto J Espay reports a relationship with Sunovion Pharmaceuticals Inc that includes: consulting or advisory. Alberto J Espay reports a relationship with Supernus Pharmaceuticals Inc that includes: consulting or advisory and speaking and lecture fees. Alberto J Espay reports a relationship with Herantis Pharma Oyj that includes: consulting or advisory and speaking and lecture fees. Alberto J Espay reports a relationship with Lippincott

Williams and Wilkins Inc that includes: consulting or advisory. Alberto J Espay reports a relationship with Cambridge University Press that includes: consulting or advisory. Alberto J Espay reports a relationship with Springer Healthcare Limited that includes: consulting or advisory. AJE cofounded REGAIN Therapeutics and is co-inventor of the patent “Compositions and methods for treatment and/or prophylaxis of proteinopathies.”

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