Functional Jerky Movements

Yasmine E. M. Dreissen, Jeannette M. Gelauff, and Marina A. J. Tijssen

Clinical Vignette

Case 1

A 52-year old man with a history of ankylosing spondylitis presented after developing jerking movements of the abdomen and legs, which predominantly occurred in the supine position and started after a period of severe lower back pain (Video 9.1). Symptoms typically occurred in the evening before falling asleep, and could last for hours, with jerks occurring every few seconds. The symptoms waxed and waned over the years, but worsened again in the past year. Due to these symptoms, the patient was significantly impaired, and declared unable to work as a journalist. Symptoms also contributed to social isolation, which took its toll on his family as well. An MRI scan of the spine was normal. On polymyographic electromyography (pEMG) the jerks were preceded by a readiness potential (RP). The patient was treated with botulinum toxin (BoNT) injections with complete symptom resolution 1 week after treatment. The treating physicians and the patient made the joint decision to not pursue additional treatment. At 1 year follow-up visit, the patient’s symptoms had returned to their prior severity.

Case 2

A 56-year old woman with hypertension presented complaining of jerky movements of the left leg accompanied by dystonic posturing with inward positioning of the ankle and foot; this movement was especially prominent when she was seated (Video 9.2). Her symptoms started after arthroscopic surgery of the left knee; the patient perceived that local anesthetic of the femoral nerve during this procedure “went wrong”. Touching the left leg and assessing reflexes elicited the movements. The patient could resolve the dystonic posturing by tapping the upper leg at a certain spot. EMG/nerve conduction study (NCS) of the femoral nerve was normal. One year after these complaints started, the patient experienced an exaggerated startle reaction after a client at work made a loud noise by slapping his hand on a table. This evolved into a generalized attack with uncontrollable non-synchronous

Supplementary Information

The online version contains supplementary material available at [https://doi.org/10.1007/978-3-030-86495-8_9].

Y. E. M. Dreissen · J. M. Gelauff
Department of Neurology, Amsterdam University Medical Center, Amsterdam, the Netherlands

M. A. J. Tijssen (*)
Expertise Center Movement Disorders, Department of Neurology, University Medical Center Groningen, Groningen, the Netherlands

e-mail: m.a.j.de.koning-tijssen@umcg.nl
movement of the arms and legs, vocalizations, preservation of consciousness and hyperventilation. The patient was admitted to a neurology ward for several days, where an electroencephalogram (EEG) performed during a similar episode was negative for epileptiform activity. She was discharged from the hospital, but remained extremely sensitive to startling noises. The auditory startle reflex was assessed, which revealed an exaggerated startle reaction with enlarged and prolonged (minutes) muscle activity mainly in stereotypical movements, vocal utterings and predominantly prolonged onset latencies (>100 ms). BoNT treatment of the left leg resulted in considerable improvement in symptoms and the startle reflex normalized 1 year after treatment.

Functional Jerky Movements

Functional jerky movements (also known as functional myoclonus) are a heterogeneous group of involuntary abrupt movements that are mostly paroxysmal and can involve the limbs, trunk and/or neck. While the prevalence of functional jerky movements within general neurology is unknown, they represent one of the more common functional movement disorder (FMD) subtypes [1–4]. In a tertiary movement disorder clinic specialized in myoclonus, more than one third of patients seen suffered from a functional jerky movement disorder [5]. They can be challenging to diagnose, mainly due to their paroxysmal nature and their heterogeneous presentation, as highlighted by the above cases. Indeed, the diagnostic accuracy for functional jerky movements based on review of video recordings by movement disorder specialists showed moderate inter-rater agreement (kappa = 0.56 ± 0.1) [6]. The diagnosis can be made with the help of clinical criteria [7–9] and supported by neurophysiological tests. In this chapter we will discuss the clinical presentation, positive signs in history and physical examination for the diagnosis, additional testing and the most important differential diagnoses.

Clinical Presentation

Although the clinical presentation is heterogeneous in terms of localization of the jerks, age at onset, associated symptoms and disease course, there are a number of factors that characterize functional jerky movements and can support the diagnosis.

History

There are a number of relevant features on clinical history that are typical for functional jerky movements. There is often an abrupt onset, triggered by either a physical or psychological trigger, after which the symptoms deteriorate [1, 10, 11]. Furthermore, like in other FMD subtypes, attention plays an important role [12], and patients either spontaneously report or confirm noticing that symptoms improve with distraction and worsen during relaxation (for example before falling asleep). Usually there is a discrepancy between the severity of the symptoms as observed by the examiner and their influence on daily functioning. Previous episodes of somatization and/or functional symptoms are relevant, but do not have high specificity, since FMD is comorbid with other non-functional movement disorders more often than expected by chance [13–15].

Although functional jerky movements can occur in any body part, their localization is helpful in distinguishing them from the differential diagnosis of tics and non-functional (“organic”) myoclonus. Generally, we find that axial jerks are likely to represent functional jerks. Facial and neck jerks point more often towards tics, especially when they start at a young age. Distal limb jerks are more likely to reflect non-functional myoclonus (see below). Functional jerks may be present continuously or episodically [10, 16, 17]. The episodic variant can be challenging to diagnose as it can be absent during the outpatient clinic visit.

In terms of non-motor symptoms in patients with functional jerky movements, similar to other FMD, high rates of pain and fatigue are found.
Interestingly, non-motor symptoms were equally common in patients with cortical myoclonus as in those with functional myoclonus, while pain was the only symptom that significantly occurred more commonly in the functional myoclonus group [5]. In general, hyperkinetic FMD are often accompanied by high proportions of psychiatric co-morbidity. Specifically, patients with functional dystonia, tremor and myoclonus exhibit high rates (50–80%) of DSM IV axis I disorders (mostly mood and anxiety disorders) and moderate rates (18–45%) of DSM IV personality disorders [1, 20–24].

**Neurological Examination**

Observation of the jerky movements may reveal variation in duration, localization, amplitude and/or direction of the movements within the same patient. High variability is suggestive of a functional etiology. The effect of attention and distraction can be tested during examination, but is not as reliable as in tremor as the jerks themselves are irregular and as previously mentioned can be paroxysmal. When distracted, the jerky movements can decrease or disappear [9]. Also, the examiner might induce adaptation of the patient’s jerks to a certain frequency, a phenomenon called “entrainment”, when asking the patient to perform a specific rhythmic task. Generally, if the character of the jerky movements can be influenced by the examiner, that argues in favor of a FMD.

Additional functional (motor) symptoms can be found, like functional weakness or dystonia, as patients commonly present with more than one functional neurological symptom [4, 19]. While this is supportive of the diagnosis of functional myoclonus, it does not rule out the possibility of a non-functional myoclonus being comorbid with a functional neurological disorder.

Abnormal stimulus-sensitivity can be observed in all FMD, and especially in functional jerky movements; this can present as exaggerated tendon reflexes or excessive startle reactions [25]. Further supportive clues are a marked response to placebo or suggestion, although, again, this is also observed in other movement disorders [10, 26].

**Additional Investigations**

Neurophysiological testing is a useful diagnostic aid in jerky movement disorders.

The first step in this testing involves evaluating the jerky movements with *polymyographic surface EMG* (pEMG). This is helpful in establishing the burst duration and the muscle recruitment pattern. An EMG burst duration of <75 ms is very unlikely to represent a jerk of functional origin [27, 28]. In myoclonus-dystonia a longer burst duration can occur, so in these cases the duration is of less distinctive value. The pEMG muscle recruitment pattern can also help with the diagnosis, including findings of entrainment, distractibility, inconsistent recruitment pattern and stimulus-sensitivity supporting a functional origin [29], although this last feature can also be present in cortical myoclonus. In the case of axial jerks specifically, the recruitment pattern is of special interest. It is generally considered that patients with functional axial jerks reveal a more variable recruitment pattern. But even with a consistent pattern a functional origin cannot be ruled out and back-averaging of the electroencephalography (EEG) time-locked to the onset of the jerk (EEG-EMG co-registration) is essential [30, 31]. With this technique a Readiness Potential (RP) also known as the Bereitschaftspotential preceding the movements can be detected, which is a powerful diagnostic aid in functional jerky movements. The RP appears about 2 seconds prior to the movement as a slowly rising negative cortical potential (Fig. 9.1) and is especially useful in distinguishing functional jerks from other jerky movements. A drawback of this procedure is that it is a time-consuming procedure requiring at least 40 jerks in order to produce a reliable result. In a small study assessing the presence an RP preceding jerky
movements, a RP was found in 25 of 29 patients with functional jerks, with a sensitivity and specificity of 0.86 [31]. When the RP is combined with event-related desynchronization (ERD) in the beta-range (13–45 Hz) prior to the jerks, even higher diagnostic certainty can be reached [32]. However, the RP can also be found in a small proportion of patients with primary tics, albeit with shorter onset latencies [31].

Another important feature of functional jerks is their stimulus-sensitivity and the presence of exaggerated startle responses. Measuring the whole-body auditory startle reflex with surface EMG can be used to distinguish between functional and other types of jerks. The auditory startle reflex consists of two responses: a stereotyped first muscle response revealing a rostro-caudal recruitment pattern with short onset latencies (<100 ms) [33, 34] followed by a second response, which is more variable in pattern and associated with behavioral processing and autonomic changes [35]. Although normal first responses are seen in functional jerky movements, the rate of second responses (onset latency >100 ms) is higher and responses are enlarged compared to healthy controls [25].

**Differential Diagnosis**

The differential diagnosis of functional jerky movements includes tics, non-functional myoclonus, epilepsy, restless legs syndrome, and primary paroxysmal dyskinesias (see Table 9.1). Below we describe how these can be distinguished from functional jerky movements.

**Tics**

Tics are defined as sudden, repetitive, non-rhythmic, purposeless, irresistible muscle movements (motor tic) or vocalizations (vocal tic) which can be classified as simple or complex [36]. They are categorized into different tic disorders, including Tourette’s syndrome. The diagnosis of these tic disorders is based on history and clinical examination, using DSM-V/ICD-10 criteria [37].

Although tics co-occur with functional tic-like jerks in a considerable number of patients [38] and pathophysiological mechanisms overlap [39], there are a number of features that can be used to support the diagnosis of primary tic.

---

**Fig. 9.1** Readiness potential, or “pre-motor” potential. Average traces from simultaneous electroencephalography (EEG) (left) and electromyography (EMG) (right) recordings, as derived from 75 extension movements of the extensor carpi radialis muscle of the left hand. The readiness potential is visible 1500–2000 ms prior to movement onset.
Tics originate in most cases in childhood, with a mean age of onset of 5 years and male preponderance (male:female ratio 3:1) [36]. This is in contrast with functional jerks which usually start in adulthood [10, 16].

The most defining feature of tics is that the movements are intentional and, in line with this feature, suppressible, while functional jerky movements are perceived as involuntary [36, 40]. Most patients with tics experience an involuntary pre-motor urge, which is subsequently relieved by carrying out the tic. Patients are often able to suppress the tics for a certain period of time, in many cases resulting in a rebound of a temporary worsening of the tics. It is therefore not surprising that tics are more stereotyped and less variable compared to functional jerks. Recently it has been described that patients with functional paroxysmal motor symptoms, like jerky movements, may experience a pre-motor urge [16, 41], however they are considered to be much less common. Further, patients with tics may camouflage the movement by assimilating it into a purposeful movement whereas patients with functional jerks are not able to hide and/or suppress their movements [36, 42, 43]. Echophenomena like echolalia (repeating sounds) and echopraxia (repeating actions), and coprolalia (involuntary swearing) are present in up to 20% of patients with primary tics, while these are rare in patients with functional jerks [17].

The entity of “functional tics” has been described in small groups of patients [44, 45]. However, it remains unclear how to distinguish them from functional jerky movements, as they do not possess the typical characteristics of a tic. Notably, premonitory urge, childhood onset, suppressibility and positive family history are lacking. Therefore, it seems more appropriate to use the broader term of functional jerky movements.

Table 9.1 Differential diagnosis of functional jerky movements

<table>
<thead>
<tr>
<th>Condition</th>
<th>History</th>
<th>Localization</th>
<th>Examination</th>
<th>Neurophysiology</th>
</tr>
</thead>
<tbody>
<tr>
<td>Functional jerky movements</td>
<td>Acute onset</td>
<td>Limbs/axial/face/neck</td>
<td>Stimulus-sensitive</td>
<td>pEMG, EEG-EMG back-averaging RP</td>
</tr>
<tr>
<td></td>
<td>Precipitating (physical event)</td>
<td></td>
<td>Entrainment</td>
<td>Enlarged startle reflex with predominant second responses</td>
</tr>
<tr>
<td>Tics</td>
<td>Premonitory urge</td>
<td>Face/neck</td>
<td>Temporarily</td>
<td>Burst duration &gt; 100 ms RP is rare, and shorter when present</td>
</tr>
<tr>
<td></td>
<td>Childhood onset</td>
<td></td>
<td>suppressible</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Vocalizations</td>
<td></td>
<td>Rebound effect</td>
<td></td>
</tr>
<tr>
<td>Cortical myoclonus</td>
<td>Variable</td>
<td>Face/distal limbs</td>
<td>Stimulus-sensitive</td>
<td>Burst duration &lt; 100 ms C-reflex</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Giant SSEP Corticospinal coherence</td>
</tr>
<tr>
<td>Myoclonus-dystonia</td>
<td>Childhood onset</td>
<td>Limbs/axial</td>
<td>Dystonic features</td>
<td>Burst duration &gt; 100 ms pEMG with brainstem recruitment pattern</td>
</tr>
<tr>
<td></td>
<td>Positive family history</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Alcohol-responsiveness</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hyperekplexia</td>
<td>Stiffness at birth</td>
<td>Generalized</td>
<td>Exaggerated startle</td>
<td>Exaggerated startle reflex with rostro-caudal recruitment pattern</td>
</tr>
<tr>
<td></td>
<td>Exaggerated startle reactions</td>
<td></td>
<td>with generalized</td>
<td></td>
</tr>
<tr>
<td></td>
<td>with generalized stiffness</td>
<td></td>
<td>stiffness</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Stimulus-sensitive</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Present in supine position</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Propriospinal myoclonus</td>
<td>Acute onset</td>
<td>Axial/limbs</td>
<td>Features of myelopathy</td>
<td>pEMG, EEG-EMG backaveraging RP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Stimulus-sensitive</td>
<td>MRI-spine, although most cases lack an underlying</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Present in supine</td>
<td>structural lesion</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>position</td>
<td></td>
</tr>
<tr>
<td>Restless legs</td>
<td>Presence when rested in</td>
<td>Lower limbs</td>
<td>Normal</td>
<td>Polysomnography</td>
</tr>
<tr>
<td></td>
<td>evening/night</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Paroxysmal dyskinesias</td>
<td>Childhood onset</td>
<td>Usually unilateral</td>
<td>Mixed movement</td>
<td>Not contributing</td>
</tr>
<tr>
<td></td>
<td>Positive family history</td>
<td></td>
<td>disorders</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Specific triggers</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

RP readiness potential (or Bereitschaftspotential), SSEP somatosensory evoked potential, MRI magnetic resonance imaging
instead of tic-like functional movements (for a more detailed description see Chap. 12).

For tics, clinical neurophysiological investigations do not add to the diagnosis. However, they are sporadically used to differentiate tics from other jerky movements, like functional jerky movements. The presence of a readiness potential before the movement is in favor of a functional neurological origin, while in tics the RP is seen less frequent and the configuration differs from FMD with shorter onset latencies (500–1000 ms) [31, 46, 47].

A correct diagnosis of either tics or functional jerky movements is important for choosing the right treatment strategy. The effect of treatment in tics is favorable, both for behavior therapy (either habit reversal or exposure to premonitory urges with response prevention) [48] with medium to large effect sizes [49] as well as for medication (dopamine D2-receptor-antagonists) with small to medium effect sizes [50].

Subcortical Myoclonus
This form of myoclonus is generated between the cortex and the spinal cord, mainly in the basal ganglia or brainstem. We will discuss the most important forms here, starting with myoclonus-dystonia (DYT11). This is a form of subcortical myoclonus generated in the basal ganglia. It is characterized by jerks of the upper limbs and trunk accompanied by mild dystonia (usually cervical dystonia and writer’s cramp) [59]. The key to the diagnosis is onset of symptoms in childhood, alcohol-responsiveness and a positive family history. High rates of psychiatric comorbidity including anxiety, depression and obsessive-compulsive disorders are also found [60]. In 50% of cases a mutation in the SGCE gene is detected [61]. Neurophysiological testing typically shows a longer (>100 ms) burst duration and is further aimed to rule out any signs of a cortical origin as mentioned above.

Brainstem Myoclonus and Startle Syndromes
Brainstem, or reticular myoclonus is characterized by a generalized, synchronous axial jerks [62]. Post-hypoxia is usually the cause [63], but other causes such as anatomical anomalies have also been described [64]. The jerks are usually stimulus-sensitive and can be elicited by startling stimuli. A syndrome which is closely related is hyperekplexia (HPX). This is a pathological startle syndrome, which is caused by different gene mutations (GLRA1, GLRB, Glyt2) involving the glycine neu-
Functional Jerky Movements

rotransmission pathway [65]. It is characterized by generalized stiffness at birth and non-habituating exaggerated startle reactions followed by short-lasting generalized stiffness in response to loud noises. Neurophysiological testing can be especially useful to distinguish the pathological startle reflex of hyperekplexia from a functional myoclonus. The startle reflex shows a consistent pEMG recruitment pattern, starting at the muscles innervated by the caudal brainstem (sternocleidomastoid), spreading in a rostro-caudal fashion with the intrinsic hand muscles showing a relatively long onset latency in the startle response [33, 65].

**Spinal Myoclonus**

This form of myoclonus can be differentiated into spinal segmental myoclonus and propriospinal myoclonus. Here, we will only discuss propriospinal myoclonus, since it is of particular interest in relation to functional jerky movements, and spinal segmental myoclonus is extremely rare. Propriospinal myoclonus is characterized by stereotyped rhythmic relatively slow axial jerks, which are often present in the supine position and elicited by tactile stimulation of the abdomen. Propriospinal myoclonus can be caused by pathology (e.g. tumor, infection) of the thoracic spine. However, most often an underlying structural lesion is lacking and it was shown that the typical movement pattern can easily be mimicked voluntarily [66]. The majority of cases of idiopathic propriospinal myoclonus have been shown to have a functional neurological origin [41, 67]. Neurophysiological testing including back-averaging in order to demonstrate the presence of a RP is especially useful.

**Epilepsy**

Repetitive jerky movements can also be seen in several forms of epilepsy. Functional jerky movement disorders show many overlapping features with functional seizures as well [68]. Functional seizures mimic epileptic seizures but lack an EEG-correlate. As in FMD, a substantial fraction (10–20%) of patients with functional seizures also have co-morbid epilepsy [69]. Despite well-formulated diagnostic criteria based on the semiology of functional seizures [70], the diagnosis remains challenging. Eyewitness history and especially video-EEG monitoring is very useful to distinguish epileptic attacks from functional seizures.

**Restless Legs Syndrome**

Functional jerky movements are usually worse when patients are not distracted and therefore many patients report more symptoms when lying in bed. In these patients, the differential diagnosis of restless legs syndrome should be considered. Restless legs syndrome is a circadian disorder with an urge to move the legs, which is often, but not always, accompanied by uncomfortable and unpleasant sensations in the legs. This urge begins or worsens during periods of rest or inactivity such as lying down or sitting, and is relieved by deliberate movements [71]. As in tics, the presence of this urge distinguishes restless legs from FMD. In functional jerky movements, the movements are not deliberate. Neurological examination in restless legs syndrome is mostly normal [71], while functional jerks are often observed in clinic. The diagnosis of restless legs syndrome is made based on clinical criteria [72].

**Primary Paroxysmal Dyskinesias**

Primary paroxysmal dyskinesias (PxD) are a rare group of movement disorders (0.76% of all movement disorders) characterized by paroxysmal involuntary movements of brief duration [73, 74], that are often misdiagnosed as FMD given their paroxysmal nature. Inherited forms of primary paroxysmal dyskinesias include paroxysmal kinesigenic dyskinesia (PKD), paroxysmal non-kinesigenic dyskinesia (PNKD) and paroxysmal exercise-induced dyskinesia (PED) [74, 75], with the kinesigenic form by far the most common one. All three forms usually start in the first or second decade of life and are caused by different gene mutations. The most well-known are the PRRT-2, MR-1, and GLUT-1
genes, but recently more genes have been detected and there appears to be a large clinical and genetic overlap [76]. They are usually familial, in contrast to functional jerky movements. The main clinical feature discriminating functional jerky movements from PxD’s are that the attacks of dystonia, chorea or ballism or a mixture of those in PxD show a consistent pattern and have a consistent trigger, especially the kinesiogenic form.

However, coexistence of functional jerky movements with paroxysmal movement disorders is described in a substantial proportion of patients [13, 17], and should be kept in mind.

Premonitory sensations are reported in the majority of PxD patients and have been described as “butterflies in the stomach”, “electricity in the head” or numbness or a tingling sensation in the limbs [77]. Attacks tend to decrease with age. Video-recordings can be very helpful in making the diagnosis, as they can show the consistent pattern of attacks.

Management

An individualized treatment plan including physical, occupational and/or psychotherapeutic interventions can be helpful in treating patients with functional jerky movements. A thorough description of these different therapeutic modalities is found in Part III of this textbook.

Conclusion

Functional jerky movement disorders (or functional myoclonus) are a common manifestation of FMD. The diagnosis can be made based on clinical signs with the help of additional neurophysiological testing; EEG-EMG with back-averaging in order to demonstrate a RP is especially useful. Since functional jerks are heterogeneous in nature, the differential diagnosis is broad and dependent of the phenomenology of each specific patient.

Summary

- Functional jerky movements (functional myoclonus) are a heterogeneous group of involuntary abrupt movements that are mostly paroxysmal with preferential involvement of the trunk, although the movements can also occur in the limbs or neck.
- The diagnosis is based on clinical signs and symptoms that can be supported by clinical neurophysiological tests.
- Clinical signs that contribute to the diagnosis include acute onset, variability of the movements and of symptom severity, and presence of co-morbid functional neurological symptoms.
- Neurological examination often shows variability in duration, localization, amplitude and/or direction of the movements; influence of distraction on the severity of the movements and entrainment can also be appreciated.
- Functional jerky movements typically display a relatively long burst duration and variable muscle recruitment pattern in neurophysiological testing. Back-averaging of the electro-encephalography (EEG-EMG co-registration) can detect a Readiness Potential (RP).
- The differential diagnosis for functional jerky movements includes tics, myoclonus, epilepsy, paroxysmal dyskinesias and restless legs syndrome.

References

9 Functional Jerky Movements


