ODD AND UNUSUAL MOVEMENT DISORDERS
Andrew J Lees

The hotch potch of miscellaneous and largely unclassified phenomena which comprise a significant and fascinating part of movement disorders are a challenge for neurologists working on the borders of psychiatry, sleep disorders, and epilepsy. Many of these conditions acquired exotic names like “Dubini’s electric chorea” and “the variable chorea of Brissaud” that are no longer acknowledged, and the historical conflict between neurological and psychiatric models continues to cause confusion. Where does a complex tic end and a compulsion begin? What about catalepsy and akinetic mutism? Are akathisia, habitual manipulations of the body (for example, trichotillomania), and stereotypies linked biologically?

Progress has been made, nonetheless, and many of these odd movements are now recognised to be symptoms of neurological and systemic disorders:
- oculomasticatory myorhythmia is virtually diagnostic of Whipple’s disease
- bizarre nocturnal movements including dystonia, stereotypies, chorea, and even vocalisations are increasingly recognised as a presentation of medial frontal seizures; autosomal dominant families with mutations of the neuronal nicotinic acetylcholine receptor gene on chromosomes 20q and 15 respectively have been reported.
- Ekbom’s syndrome has become de rigeur as a subject of consequence for neurologists following the demonstration that dopamine receptor agonists may be remarkably effective
- some classic neurological disorders can present with an uncharacteristic abnormal movement:
  - the Huntington mutation has been found in middle aged patients presenting with Parkinson’s syndrome and cases with generalised myoclonus have been described
  - chorea–acanthocytosis may present with parkinsonism
  - the spinocerebellar ataxias and ataxia–telangiectasia with dystonia or chorea
  - Wilson’s disease can present with stereotypies
  - Freidreich’s ataxia occasionally presents with myoclonus or chorea.

Few neurologists would now label spasmodic torticollis, musician’s tremors, propriospinal myoclonus or startle disease as psychogenic; even tics have a degree of acceptance in most neurological circles. However, one must not shy away from labelling an abnormal movement disorder as psychogenic if the balance of evidence points to this. As much harm can be done by inappropriately labelling a motor conversion disorder or abnormal illness behaviour as dystonia or tremor as vice versa.

WHAT IS IT?

The first step to diagnosis depends critically on an accurate description of the abnormal movement. Tremor, dystonia, chorea, and myoclonus are dyskinesias recognised to reflect neurological disease. These dyskinesias are not mutually exclusive and terms such as “dystonic tremor” and “mixed movement disorder”, while ungainly, are acceptable. Other terms are falling out of use—for example, athetosis, a writhing, sinuous distal limb movement frequently seen in cerebral palsy, is now often classified as dystonia and ballism linked with chorea.

Tics and stereotypies are distinct motor behaviours seen commonly in neuropsychiatric practice and not infrequently in movement disorder clinics. Chorea–acanthocytosis, post-streptococcal acquired autoimmune neurological disease, and Rett syndrome are disorders that may present with these complex movements.

Fixed muscle contractures, jerks, and spasms seen after peripheral trauma need to be distinguished from dystonia and myoclonus. Other odd motor behaviours seen in neurological practice include akathisia, compulsions and rituals, apraxias, cramps, geniospasm, mannerisms, hemifacial spasms, and myokymias.

The consulting room may not always be the best setting for evaluation of patients with movement disorders as many dyskinesias are strikingly situation specific and variable in severity. Emotion, bright light, specific physical tasks, startle, and sleep may all accentuate or unleash the abnormal movement. Discretely following the patient out into the street can be instructive. If the patient describes a specific situation or task that provokes the movement this should be reproduced...
Table 1  Some treatable disorders to consider when evaluating a movement disorder

<table>
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<th>Disease</th>
<th>Presentation</th>
<th>Confirmatory tests</th>
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<tr>
<td>Wilson’s disease</td>
<td>Tremor, dystonia, parkinsonism, myoclonus Kayser-Fleischer rings</td>
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<td>Segawas disease</td>
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<td>Oculomasticatory myorhythmia, myoclonus, supranuclear gaze palsy with PSP like picture</td>
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<td>Myoclonic hereditary dystonia</td>
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CSF, cerebrospinal fluid; PCR, polymerase chain reaction; PSP, progressive supranuclear palsy; SPECT, single photon emission computed tomography.

during the examination if possible. Camcorders and home videos provide another opportunity to supplement the examination, with archival family clips depicting the dyskinesia and its evolution.

A few rare but treatable movement disorders to always consider in the differential diagnosis are listed in table 1. A list of systemic disorders presenting not infrequently with dyskinesias is provided at the end of the article (see box).

THE “INVISIBLE” MOVEMENT DISORDER

Although “spot diagnosis” is an accepted part of movement disorder practice it is also commonplace for no movement disorder to be apparent during the taking of the clinical history. Tics should be the first thing to cross one’s mind, particularly if the movements are reported to occur in the face, head or neck. Eye winks, eye blinks, grimaces, and head tosses are some of the most common movements. They are rapid, stereotyped, wax and wane in severity, and are relatively easy to imitate. They are easier to suppress voluntarily than chorea or dystonia and are triggered by stress or boredom. There is a notable (3:1) male preponderance with a peak age at onset of 7 years. Tics may be accompanied by sensory impulses or urges, and voluntary suppression leads to a build up of internal tension and dysphoria.

When the tics are present for longer than a year and are accompanied by abnormal vocalisations the diagnosis of Gilles de la Tourette syndrome (GTS) can be made. There are important phenomenological overlaps and associations between GTS and obsessive compulsive disorder, and it is important to identify the principal diagnosis. A high incidence of tics occurs in Asperger’s syndrome and there are intriguing links with attention deficit disorder and developmental stammering. The natural history of tics is favourable with full resolution occurring in most instances by the end of adolescence. In contrast, severe cases of GTS with associated behavioural disturbances can bring serious social disadvantage and may need drug treatment.

The neurological examination needs to be tailored to the clinical history in many cases. Trombonist’s cramp, the golfer’s yips or writing tremor can be brought out by reproducing the specific task. It is helpful to ask the patient to bring the relevant tool or instrument to the consultation.

Paroxysmal movement disorders are another potential pitfall. These can be misdiagnosed as epilepsy despite the absence of any disturbance of awareness. The most common is paroxysmal kinesigenic choreathetosis/dyskinesia (PKC/PKD), which normally presents in childhood or adolescence with abnormal movements triggered by a sudden movement or change in movement velocity. It may run in families and there is a striking male preponderance. The best opportunity to see this is when the patient is called from the waiting to the consulting room. On rising after prolonged sitting, sudden, dystonic or choreic movements of the face and limbs which are frequently one sided occur, lasting no more than a few seconds. These attacks may occur many times a day and can also be triggered by over breathing, startle, continuous exercise or even a change in walking speed. They may cause great embarrassment but fortunately respond extremely well to low doses of phenytoin and carbamazepine. Paroxysmal non-kinesigenic dyskinesia (PNKD), although also of childhood onset and often familial, lasts much longer (10 minutes to six hours), is usually dystonic, and attacks are less frequent (three a day to two a year). These may be triggered by alcohol, caffeine, stress or fatigue and can respond to benzodiazepines and levodopa. A third intermediate type called paroxysmal exercise induced dyskinesia (PED) is triggered by prolonged exercise (at least 10 minutes) with the dystonic movements appearing in the part of the body which has been exercised. Recovery occurs over 5–30 minutes and attacks can also be triggered by vibration, passive movement or cold. The similarity of these familial conditions to the episodic ataxies and periodic paralyses, and occasional reports of therapeutic success with acetazolamide, has led to the suspicion that they also may to be caused by defects in genes regulating ion channels (channelopathies).

Familial PKD has been linked to chromosome 16 and PNKD to chromosome 2, both close to clusters of ion channel genes. Multiple sclerosis, cerebrovascular disease, hyperthyroidism, hypoparathyroidism, hypo- and hyperglycaemia, and head injury may all cause paroxysmal movement disorders and a paroxysmal presentation can occur in psychogenic dystonia.

Abbreviations

CRPS: compound regional pain syndrome
EEG: electroencephalogram
EMG: electromyelogram
GTS: Gilles de la Tourette syndrome
PED: paroxysmal exercise–induced dyskinesia
PKC/PKD: paroxysmal kinesigenic choreathetosis/dyskinesias
PNKD: paroxysmal non-kinesigenic dyskinesia
SPECT: single photon emission computed tomography
PSYCHOGENTIC MOVEMENT DISORDERS
MASQUERADING AS NEUROLOGICAL DISEASE

An accurate diagnosis of a psychogenic movement disorder is one of the most difficult and important a neurologist has to make. Neurologists usually and appropriately recognise psychological causes but are sometimes reluctant to communicate their opinion to the patient because of lingering diagnostic uncertainty and a fear of litigation. Patients rarely accept this diagnosis and frequently request further opinions. This may lead to a catalogue of disappointments, substantial misuse of resources and, worst of all, inappropriate treatment. If a primary psychiatric disorder is suspected then a detailed psychiatric history needs to be taken in an attempt to unearth cryptic stresses and traumas. This takes time, compassion, and patience but can be achieved by a neurologist without necessarily involving a psychiatric colleague.

A motor somatiform disorder is one in which the physical symptoms are linked to psychological factors but is not under conscious control. The two main types are conversion disorder and somatisation disorder (hysteria or Briquet's syndrome). In conversion disorder a temporal relation to a stressful life event or unresolved conflict may be uncovered; it may be apparent that the movement disorder is freeing the patient from a perceived or real threat, or facilitating a repressed need. Briquet's syndrome involves recurrent and changing somatic complaints over years for which medical attention is sought. It may develop out of a chronic, untreated, recurrent conversion disorder.

Malingering is the deliberate production of a factitious movement disorder to achieve goals such as school avoidance, financial compensation or evasion of criminal prosecution. Finally, a factitious disorder can be produced intentionally, but as a result of a psychological need and, in contrast to malingering, is caused by an underlying psychological disorder—for example, Munchausen's syndrome. Factitious disorders are usually associated with masochistic, dependent or antisocial personalities. This classification depends on the degree of conscious intent which in practice is often impossible to determine with any degree of certainty. To be confident of a primary psychiatric cause or malingering the symptoms must be completely relieved by psychotherapy, psychological suggestion with supportive physical rehabilitation, or by placebo administration. Alternatively the patient must be witnessed to be symptom-free when left alone and supposedly unobserved.

Helpful clues to the diagnosis of psychogenic movement disorders are given in table 2. Shaking is probably the most common presentation, and the main differential diagnosis is an organic atypical tremor—for example, caused by early Wilson's disease. Helpful clues in the history include sudden onset, complete remissions, and onset in one limb with rapid spread to other body parts. Examination finds a bizarre combination of rest, and postural and action tremor, with considerable variabilities in frequency, amplitude, and direction. The tremor may increase with attention or loading and decrease with distraction, and there may be a total absence of finger tremor. Some cases may have additional unrelated and atypical neurological signs such as sensory loss or a bizarre gait. If one asks the patient to tap an unaffected (or affected) limb to a changing frequency, psychogenic tremor tends to follow the new speed as it is extremely difficult to maintain two different frequencies simultaneously; this is known as entrainment. Another useful test is based on the observation that tremor occurs only in the presence of increased muscle tone, and disappears when a body part is totally relaxed (the co-activation sign). Response to pharmacotherapy varies but complete abolition with any drug should raise suspicions.

Psychogenic parkinsonism is rare but single photon emission computed tomography (SPECT) using dopamine transporter and D2 receptor ligands (DAT and IBZM scans) can help to persuade a sceptical patient that their dopamine system is working normally. Dystonia musculorum deformans, because of the absence of a structural histopathological lesion and its paradoxical motor phenomenology, has since its beginnings been mistaken for a psychiatric disorder. Furthermore, it is only relatively recently that blepharospasm, spasmodic dysphonia, torticollis, and occupational hand cramps have been accepted as focal dystonic fragments. It is ironic, therefore, that neurologists are now faced with the difficult task of identifying rare cases of psychogenic dystonia. Some of the cases are paroxysmal and there is a pronounced female preponderance. Psychogenic dystonia should not be lumped with abnormal movements associated with a complex regional pain syndrome (CRPS), which is a more common problem.

ABNORMAL MOVEMENT DISORDERS SEEN IN PSYCHIATRIC DISEASE

Abnormalities of movement may accompany disorders of the mind and alert us to psychiatric illness. The slowness of movements of a patient with a major depressive disorder bears superficial similarity to Parkinson's syndrome, leading to considerable scope for diagnostic error. Tremor and habitual manipulations of the body, such as nail biting and motor restlessness, may point to chronic anxiety. Obsessional compulsive disorder may be accompanied by profound slowness, tics, and pathognomonic compulsions. Schizophrenia may be suspected by body language which can include an eccentric gait, gaze and eye movement disorders, and a constellation of bizarre mannerisms and stereotypies. Catalepsy, waxy flexibility, and extreme motor excitement with hyperthermia may all occur as part of the catatonic syndrome. Stereotypies are also rife in those with learning disability and are seen commonly in autism—for example, body rocking, head banging, and hand wringing.

Antipsychotic drug treatment can provoke a plethora of dyskinesias which may pose diagnostic conundrums. Many of these abnormal movements may rarely occur spontaneously in schizophrenia, but there is incontrovertible epidemiological evidence implicating neuroleptic drugs in their occurrence. Persistent generalised chorea with orofacial stereotypies in a

Table 2 Clues which may point to a psychogenic movement disorder

- Inconsistent or incongruous movements which “don’t look quite right”
- Additional volitional weakness
- Sensory signs which do not follow anatomical rules
- Presence of additional phenomena—bizarre gait, contrived slowness to command, shaking bouts or violent truncal spasms, babbbling, excessive startle response
- Severe accompanying fatigue
- Remissions
- Acute onset with fixed posturing
- Self inflicted injuries
- Previous or present multiple somatisations
- Employed in health profession or pursuing litigation
- Secondary gain such as continuing care by a devoted spouse
psychiatrically ill patient needs to be distinguished from Huntington’s disease and choreoacanthocytosis. Dystonic syndromes can occur acutely after starting antipsychotic drugs when they are painful and distressing, and may be mistaken in an accident and emergency department for tetanus, rabies or hysteria, or they may be delayed in onset and persistent even after drug withdrawal and include axial spasms, torticollis, bruxism, and limb dystonia. Persistent or tardive dyskinesias may improve for up to three years after neuroleptic withdrawal. Parkinson’s syndrome indistinguishable from Parkinson’s disease may be triggered by neuroleptic drugs. The response to levodopa is negligible because of dopamine receptor blockade, but anticholinergic drugs are beneficial. Some of these patients, particularly the elderly, do not recover following neuroleptic withdrawal, raising the possibility that they may have had presymptomatic Parkinson’s disease. Akinesthesia may occur after brief neuroleptic exposure, or on maintenance treatment, and may be persistent. It consists of pacing, marching on the spot or stereotyped leg crossing associated with dysphoria. Motor stereotypies may be associated. This needs to be distinguished from severe anxiety and Ekbom’s syndrome. Potentially fatal bulbar dyskinesias, and profound akinesthesia with hyperthermia (neuroleptic malignant syndrome) are less common.

ABNORMAL MOVEMENT DISORDERS ORIGINATING IN THE PERIPHERAL NERVOUS SYSTEM

Hemifacial spasm caused by facial nerve compression in the posterior fossa causes repetitive unilateral clonic and tonic contractions. Focal myoclonus can occur as a result of spinal roots or plexus lesions, and similar lesions less commonly cause spasms akin to dystonia. Tremor is a recognised manifestation of peripheral neuropathy (see article by Bain, p 13). Peripheral injuries may also unleash dystonia as occurs with whiplash injuries in carriers of the DYT1 dystonia gene. Neuromyotonia is a distinctive movement disorder characterised by continuous muscle fibre activity (complex repetitive discharges). These patients have myokymia with impaired muscle relaxation and stiffness. Abnormal postures of the feet and hands may occur resembling carpopedal spasm. Some cases caused by peripheral nerve, plexus or root lesions may respond to carbamazepine or phenytoin, and immunological mechanisms have been suggested. Cases of complex regional pain syndrome in which patients develop causalgic pain, hyperpathia, and allodynia with sudomotor, vasomotor, and trophic changes in skin, subcutaneous, and bone (reflex sympathetic dystrophy) may be occasionally associated with severe dystonic contractions in the affected limb. Some of these patients have more mobile jerky spasms and spread to other limbs is common. Many young women suffer from this syndrome and the prognosis is poor. It is speculated that often trivial trauma can initiate changes in sensory input and give rise to abnormal impulse transmission in sensory nerves, which leads to reorganisation of central processing of sensorimotor information. However, there is an opposing view that many if not all of these cases have primary psychiatric causes. Painful legs (arms) moving toes (fingers) is another disorder where peripheral root, plexus or peripheral nerve lesions have been implicated. Pain usually precedes the movements by at least a few days—and sometimes years—and may be unilateral or bilateral. The pain is burning or crushing (causalgic) and not linked to a peripheral nerve or dermatomal distribution. The toe (or less commonly finger) movements are distinctive complex sequences of abduction-adduction or flexion-extension movements (1–2 Hz). The movements are believed to be centrally generated.

ODD TREMORS

Primary orthostatic and other tremors are dealt with elsewhere (see article by Bain, p 13). Palatal tremor can be divided into two subtypes. The essential form presents with rhythmical ear clicks caused by tremor of the tensor veli palatini which connects to the eustachian tube. The tremor frequency ranges between 1.5–3 Hz and local injection of botulinus toxin can be helpful. Symptomatic palatal tremor is caused by a structural lesion within the Guillain-Mollaret triangle and results from rhythmical oscillations of the levator veli palatini. Olivary hypertrophy can be seen on magnetic resonance imaging, and Whipple’s disease should be considered in the differential diagnosis.

ODD JERKS

Myoclonus is an electric shock-like, short, rapid involuntary muscle jerk which arises within the central nervous system and may be irregular or rhythmical. Asterixis is sometimes referred to as negative myoclonus. It may be difficult to distinguish from jerky dystonic movements or tics, the major difficulty usually being the distinction from psychogenic jerks. Neurophysiological techniques include back averaging electroencephalogram/electromyogram (EEG/EMG) recordings. Jerks occurring in response to a specific stimuli can also be studied by recording the latency of reflex activity to the stimulus. Cortical myoclonus is multifocal and worse on action. There may be an epileptic EEG, giant cortical somatosensory evoked potentials, and a cortical correlate preceding the jerk on back averaging. The most common causes are post-anoxic myoclonic encephalopathy (Lance-Adams syndrome) and syndromes of myoclonic epilepsy with ataxia. Myoclonus generated in the brainstem provokes generalised jerks, including the face, with characteristic reflex jerks precipitated by sound. Some of these cases are caused by hereditary hyperekplexia with mutations in the α subunit of the glycine receptor. Another cause is brainstem stroke. Propriospinal myoclonus causes axial jerks sparing the face, worst when resting supine; traumatic extrinsic and intrinsic spinal cord lesions should be searched for. Spinal myoclonus leads to focal rhythmical jerks with denervation on neurophysiological studies; as with propriospinal myoclonus, structural cord disease needs to be looked for.

EXOTIC ODD MOVEMENTS

- The dancing larynx: complex arrhythmical movements of the laryngeal cage sometimes associated with tongue and posterior pharyngeal movements and clicking but no uvula movements. Probably represents a palatal tremor variant
- The moving ear: slow semirhythmic movements of the ear under some voluntary control and often associated with discomfort have been reported as a focal dyskinesia. Ear movements may also be seen occasionally as tics, part of palatal tremor, and as a psychogenic disorder
- The Pisa syndrome: an axial dystonia characterised by truncal rotation and lateral flexion. It occurs during chronic neuroleptic treatment and has rarely been reported following dopamine agonist use in Parkinson’s disease, multiple system atrophy, and following the use of cholinesterase inhibitors in Alzheimer’s disease. Anticholinergic treatment may be beneficial
- Sandifer syndrome (dyspeptic dystonia): twisting torsion movements of head, neck, and trunk with abnormal postures
occurring during feeding and immediately afterwards. This is a childhood disorder caused by gastro-oesophageal reflux with or without hiatus hernia

- The rabbit syndrome: a distinctive perioral tremor most often seen as a complication of neuroleptic treatment but sometimes occurring spontaneously or in association with Parkinson’s disease. It responds well to anticholinergic drugs

- Tonic inverse masticatory activity: abnormal contraction of a mouth closing muscle during mouth opening and no activity during mouth closing—resembling hemimasticatory dystonia caused by pontine or cerebellopontine angle structural lesions

- The jumpy stump: jumping of an amputation stump accompanied by lancinating pain, not uncommonly occurring transiently in the postoperative period, but there are other cases of tremor, spasms, and jerk with delayed onset and persistence over many years. The abnormal movement can be triggered by voluntary movement and occasionally cutaneous stimuli

- Hereditary chin quivering (geniospasm): autosomal dominant childhood onset disorder (linkage to chromosome 9q) with episodic, usually stress induced, trembling of the chin with a frequency of 8–10 Hz; responds well to local mentalis botulinus toxin injections.

- Belly dancers dyskinesias: undulating rhythmical movements of the abdominal wall causing distressing circular rotatory umbilical motion. Most reported cases follow in the wake of a laparotomy and pain may be associated. Diaphragmatic flutter should be eliminated

- Camptocormia: extreme flexion of the thoracolumbar spine which increases with walking and disappears in the recumbent position. Rarely this may be the most prominent and disabling feature of Parkinson’s disease and seems to have no clear association with drug treatment. It may be a form of axial flexion dystonia.

RECOMMENDED TRAINING AIDS

All trainee neurologists with a specialist interest in movement disorders should join the Movement Disorder Society (fellows rate US$100 a year) which provides an annual subscription to Movement Disorders and discretionary rates at the society’s conferences.

Some systemic disorders presenting with dyskinesias

- Thyrotoxicosis: tremor, chorea
- Renal failure: myoclonus, asterixis, restless legs
- Non-ketotic hyperglycaemia: chorea, myoclonus
- Polycythaemia: chorea
- Antiphospholipid syndrome and systemic lupus erythematosus: chorea, myoclonus
- Hypoponatremia: myoclonus, dystonia
- Streptococcal infections: chorea, tics
- Hypoglycaemia: tremor, myoclonus
- Liver failure and acquired hepatocerebral degeneration: myoclonus, asterixis, tremor, chorea
- Paraneoplasia–opsoclonus: myoclonus, chorea, dystonia
- AIDS: chorea, dystonia, myoclonus, tremor
- Hypoparathyroidism: dystonia, chorea

RECOMMENDED BOOKS


KEY REFERENCES


