Introduction
Patients with movement disorders typically present with a change in their overall pattern of movements: this may represent an increase of movement (hyperkinetic), decrease (hypo- or akinetic), uncoordinated movement (ataxia), or a combination of the aforementioned. The initial task is to properly categorize the appearance or “phenomenology” of the movement disorder, as this is the essential step to guide the clinician in developing a differential diagnosis and treatment plan. Given recent advances in neurology, the majority of movement disorder patients are candidates for treatment, such as medication, physical therapy, or surgical interventions.

The first part of this book provides a short chapter on non-parkinsonian hypokinetic movement disorders; parkinsonian disorders are covered in another volume in this series. The second part includes hyperkinetic disorders. Part three includes various syndromes that do not fit into the other categories or that overlap between categories. Broader chapters in part four, on genetics, neuroimaging, rating scales, and videotaping suggestions, are intended to serve as clinician resources.

This introductory chapter provides an approach that will facilitate the evaluation of a movement disorder patient. The phenomenological categorization of the most common movement disorders falls into seven major categories: parkinsonism, tremor, dystonia, myoclonus, chorea, ataxia, and tics. Most of the commonly encountered disorders can be classified into one of these categories, but given the breadth of the diseases in the field, there are many unusual or rare types of movement that may not be easily categorized or may be consistent with more than one phenomenological category. A thorough history and examination are essential to defining the phenomenology. Home videotapes of the patient may also be useful if the movements are intermittent, variable, or not seen clearly in the office. Laboratory testing and imaging are necessary in some movement disorders, but are less helpful in many circumstances given that the disorders are diagnosed mainly on history and examination.

History
Start by asking six questions in the history.

1. Can you describe the movements?
Patients will usually be able to describe a decrease or increase (or both) in their overall movement from baseline, although often hyperkinetic aspects of abnormal movements can overshadow the hypokinetic movements from the patient’s perspective. Hypokinetic movement disorders, also termed bradykinesia (slowed movement) or akinesia (loss of movement) are characterized by an overall decrease in the speed or amplitude of movement in any area of the body. Signs and symptoms could include decreased facial expression, slowed speech, reduced...
dexterity of the extremities, decreased arm swing, and slowed walking speed. Hyperkinetic movement disorders, also generally termed dyskinesia (abnormal movements), are characterized by an increase in baseline movements. Hyperkinetic movement disorders have highly variable manifestations, ranging from increased eye closure to arm flailing to jerking of the legs. Lastly, patients may complain of a change in the character of voluntary movements, such as becoming clumsy or unsteady with walking, which may be seen in ataxic disorders.

Certain features of abnormal movements are very important to elicit in the patient’s description. Defining the conditions under which the movement occurs, such as with rest or with action, is necessary for accurate diagnosis and categorization of tremor. An ability to suppress the movement or an increase in the movement with suggestion are features common to tics. Specific triggers of the movements, especially with certain tasks, may be reported in dystonic disorders or paroxysmal movement disorders. Myoclonus can be triggered by startle. Asking about worsening of the disorder or improvement with certain foods or alcohol can narrow the differential diagnosis in forms of dystonia, myoclonus, or tremor disorders. A history of falls, especially the temporal course, is helpful in disorders that affect gait and balance, as falls are seen earlier or more frequently in some disorders as opposed to others.

2. When did the movements start and how have they changed over time?
Most movement disorders are subacute or chronic in nature. An acute onset is less common and may signify a secondary movement disorder related to an underlying inciting event, such as a stroke or medication change. Acute onset of movement disorders at maximal severity is also commonly seen in functional movement disorders, where patients will often present to emergency departments from the start. Most hypokinetic, hyperkinetic, and ataxic movement disorders will slowly worsen over time. Disorders that improve over time are less common; for example, tic disorders will typically improve from childhood into adolescence and adulthood. Static movement disorders may occur with birth injury or some dystonic disorders.

3. Are the movements continuous or intermittent?
Although many movement disorders start out as intermittent or suppressible, they tend to become more continuous or constant when they progress over time. The rest tremor seen in parkinsonian disorders is a classic example, where the tremor starts intermittently in a limb before becoming more regular and spreading to other limbs. Early on, this type of tremor can be sometimes voluntarily suppressed or decreased with movement, but later the tremor is continuous. Episodic movement disorders are much less common. Paroxysmal disorders, which are typically choreic or dystonic in nature, can many times be diagnosed by history alone if specific triggers such as sudden movements cause the disorder to occur. Functional (psychogenic) movement disorders are also frequently episodic. The circumstances under which the movement occurs can be particularly helpful. For example, restless legs syndrome worsens at night when the patient is laying down.

4. Is there a family history?
All modes of inheritance patterns are seen in movement disorders and the genetic basis of these disorders is rapidly being discovered. It is not sufficient to inquire only about the particular movement disorder seen in the patient, since broadening the questioning to other biological family members may yield additional important clues. For example, patients with grandchildren with intellectual disabilities may be at risk for fragile X-associated disorders. Tic patients may have associated diagnoses in the family, such as attention deficit hyperactivity disorder.

5. Are there other medical illnesses?
The majority of movement disorders are restricted to the nervous system, but systemic organ involvement may provide diagnostic clues. For example, patients with underlying cancers may be at risk for paraneoplastic disorders and iron deficiency anemia or diabetes may predispose to restless legs syndrome. The presence of cardiomyopathy is associated with Friedreich ataxia or mitochondrial disorders. Enlargement of visceral organs (spleen, liver) may suggest a lysosomal storage disease.

6. Have the movements been treated in the past and what was the response to treatment?
A response to dopamine medications may facilitate diagnosis of dopa-response dystonia. Paroxysmal movement disorders may be exquisitely responsive to antiepileptic medications. Other substances may improve movements, such as the improvement
of essential tremor, essential myoclonus, and myoclonus-dystonia with alcohol.

Examination

Depending on the movement disorder, abnormal movements may be present in focal or contiguous areas of the body or may be generalized. By determining the location and phenomenology of the movement, most patients can be placed into one of seven distinct patterns of abnormal movement.

Parkinsonism

The main features of parkinsonism are tremor at rest, bradykinesia or akinesia, rigidity, loss of postural reflexes, flexed posture, and freezing. Parkinsonism, in particular, Parkinson disease, is the most common disorder seen in movement disorder clinics and is covered by another volume of this series.

Tremor

This pattern is typically rhythmic and oscillatory and may affect more than one body part. Tremor should be classified on examination by the conditions under which it is activated: at rest, with posture, or with action. Tremor may be present in multiple conditions, for example, essential tremor, which is frequently seen with posture and action or intention. Tremors may also be task specific, such as the dystonic tremor of writer’s cramp.

Chorea

Choreic movement is random in nature and is purposeless, non-rhythmic, and unsustained. It may appear to flow from one body part to another. Huntington disease is a frequent cause of chorea and manifests with brief, irregular movements. Chorea can be suppressed or camouflaged. It can be accompanied by “negative chorea” or motor impersistence.

Dystonia

In dystonia, agonist and antagonist muscles contract simultaneously causing twisting movements that are frequently sustained. The speed of the movement is variable and when sustained, can lead to abnormal postures and contractures. Dystonia is typically worsened with action, sometimes only occurring with specific actions. It can be classified by location, age of onset, and etiology, and the classification system has recently been revised.

Myoclonus

This pattern consists of brief, sudden, typically irregular jerks from muscle contraction. Myoclonus may be synchronized and triggered by action or startle. Negative myoclonus is caused by inhibition of the muscles, with the classic example being asterixis. Myoclonus can be rhythmic or oscillatory and occur in various parts of the body, either focally or generally.

Tics

Tics are abnormal movements (motor) or sounds (phonic) that are abrupt, usually transient, and can be simple or complex. Tics can vary over time and can be accompanied by an uncomfortable urge or feeling. Tics may be suppressible, although severe tics may be continuous. Gilles de la Tourette syndrome is characterized by the presence of both motor and phonic tics, present for more than one year, with young onset.

Ataxia

Lack of coordination of movement distinguishes ataxia from other movement disorders. The pattern of atactic movement varies, but may include clumsy limb movements (dysmetria), dysarthria, ataxic eye findings such as abnormal pursuit, and uncoordinated walking. Kinetic tremor can also accompany ataxic signs. Ataxia can be localized to the peripheral or central nervous system so a thorough sensory and vestibular examination is necessary in these patients.

Other patterns of movements

There are several other types of abnormal movements that, despite being distinctly recognizable, do not fit well into the preceding patterns. These include stiff-muscles, akathetic movements, myokymia, paroxysmal dyskinesias, restless legs, and stereotypy. In addition, some movement disorders have more than one pattern of movement, such as in the myoclonus-dystonia disorders. Functional movement disorders frequently do not fit well into the above-described patterns, but caution must be maintained, since many unusual movement disorders can be labeled functional.

Diagnostic testing

Accurate description of the phenomenology of the abnormal movements as a result of the history and examination is the first and most fundamental step
in diagnosis of movement disorders. Additional diagnostic testing is not warranted in many situations, for example, in the classic appearance of Tourette syndrome. However, there are some studies that may enhance or confirm clinical diagnosis. For example, laboratory studies can be useful particularly with tremor. Abnormalities of the thyroid, evidenced by elevated or low thyroid stimulating hormone (TSH), may cause or worsen tremor. Wilson disease, diagnosed by abnormal copper levels (in serum and/or urine), low ceruloplasmin, and the presence of Kaiser–Fleischer rings; should be considered in younger patients who present with bizarre tremors or other unusual movement patterns/combinations.

Genetic testing is available for many movement disorders and is driven by family history, age of the patient, and financial resources. For the more rare movement disorders, such as the inherited ataxias and Huntington disease, it may be the only testing that can give a definitive diagnosis. For individuals who are considering family planning, it may be necessary that genetic testing be accompanied by genetic counseling.

Neurophysiological assessment may be helpful in myoclonus, where myoclonic jerks show brief electromyography (EMG) bursts of 10–50 milliseconds. Rhythmicity in tremor can be demonstrated on EMG, but this is not frequently ordered by clinicians when evaluating a patient with tremor. Electromyography may also be helpful therapeutically in dystonic patients when used in conjunction with botulinum toxin treatment. Nerve conduction studies may be used to evaluate atactic individuals for sensory abnormalities in peripheral nerves.

Imaging can be valuable in movement disorders that do not fit classic patterns or presentations. The most common movement disorders typically show normal basal ganglia structures on routine imaging, as in essential tremor, and dystonia. However, patients with movement disorders that are localized to one side of the body, that have abrupt stroke-like onset, or that include ataxia should be imaged with computed tomography or preferably, magnetic resonance imaging. Atrophy of specific structures, such as the striatum in Huntington disease, or the cerebellum in degenerative ataxias, may support the clinical diagnosis. Functional or nuclear medicine imaging is playing an increasingly important role in diagnostics.

**Treatment**

The majority of treatment options in movement disorders are symptomatic, not curative. However, in a few circumstances, early intervention of treatable forms of movement disorders may be curative or halt the progression of the disease. While rare, such conditions should be considered in patients with particular disease profiles; examples include patients with young onset tremor, dystonia or parkinsonism (Wilson disease), or fluctuating dystonic and parkinsonian features (dopa-responsive dystonia).

The approach described in this chapter offers a straightforward approach to evaluating a movement disorder patient. Questions about the movements, course, family history, medical illnesses, and medication response will help the clinician with the evaluation. Correctly describing the phenomenology is key to narrowing the list of diagnostic possibilities and guides the need for additional testing. The subsequent chapters will fill in the details, and with this framework, the reader will gain an ease of diagnosis and treatment of movement disorders.

**Further Readings**
