In nature, there exist common traits which identify phenomena. The fundamental principles are often so basic that uneducated individuals can easily recognize the traits with little instruction. A goal of scientific research is to observe and define the characteristic features of the object of interest. For example, there are aspects which distinguish some plants as follows:

Rose is a rose is a rose is a rose (1).

There are particular qualities, such as the scent and the configuration of the petals and stem, that allow most people to identify roses and to distinguish roses from daffodils, tulips, dandelions, and other flowers. It does not matter if the rose comes from Australia, Brazil, or Mongolia. The fundamental features of the rose are the same.

By the same token, in medicine one strives to characterize the patterns that distinguish one disease from another. That is, one seeks to identify syndromes. A syndrome is a constellation of symptoms, the subjective complaints of the patient; signs, the objective findings of the examiner; laboratory values; family history; and natural history. The causes of syndromes may be multiple or unknown. Knowledge of the existence of a syndrome allows prediction of the prognosis of the syndrome and the likely response to possible treatments. Thus, establishing that a patient has the diagnosis of a syndrome is valuable to predict the course of the illness and the outcome of therapy. For example, neurologic syndromes include upper motor neuron syndrome, amnesic (Korsakoff) syndrome (2), and parkinsonism (3,4), and psychiatric syndromes include schizophrenic disorders, somatization disorder (hysteria) (5), and autistic disorder (6). Some syndromes are fully defined by the abnormalities in movement. As an example, a trained examiner, who observes a complete stranger walking across a street exhibiting a pill-rolling tremor, rigidity, akinesia, and postural instability, can diagnose the syndrome of parkinsonism (4), although the cause of the syndrome would be unknown without additional information. Thus, in some individuals the existence of the movements uniquely seen in parkinsonism implies the presence of the syndrome of parkinsonism. In this manner some movement disorders are pathognomonic of a syndrome.

We hypothesize that some movements are similarly pathognomonic of autistic disorder in some individuals. Autistic disorder is an extremely disabling syndrome with onset in early childhood, is associated with multiple comorbid conditions. Although autistic disorder is heterogeneous in its manifestations, there is a subgroup of individuals with autistic disorder who display movements that appear to be unique for the disorders. Hand flapping and a variety of movements termed stereotypes may be pathognomonic of autistic disorder. Therefore, identification of a movement disorder characteristic of autistic disorder may imply that the individual has autistic disorder.

Summary  Autistic disorder, an extremely disabling syndrome with onset in early childhood, is associated with multiple comorbid conditions. Although autistic disorder is heterogeneous in its manifestations, there is a subgroup of individuals with autistic disorder who display movements that appear to be unique for the disorders. Hand flapping and a variety of movements termed stereotypes may be pathognomonic of autistic disorder. Therefore, identification of a movement disorder characteristic of autistic disorder may imply that the individual has autistic disorder.

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Movements in autistic disorder

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In nature, there exist common traits which identify phenomena. The fundamental principles are often so basic that uneducated individuals can easily recognize the traits with little instruction. A goal of scientific research is to observe and define the characteristic features of the object of interest. For example, there are aspects which distinguish some plants as follows:

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We hypothesize that some movements are similarly pathognomonic of autistic disorder in some individuals. Autistic disorder is a condition presenting in early childhood manifested by marked abnormalities in social interactions and relationships, communication and language, and range and repertoire of interests and activities. The cause of most cases of autistic disorder is unknown. Most individuals with autistic disorder have mental retardation. Many individuals with autistic disorder have seizure disorders (7). A wide variety of genetic and other comorbid disorders have been reported in many people with autistic disorder (6). Neuropathological and neuroimaging studies have demonstrated abnormalities in several different regions in various individuals with autistic disorder. Hypotheses about the basis of the syndrome have included the neocortex, the limbic system, the cerebellum, the diencephalon, and the brain stem. Hypotheses about neurochemical disorders in autistic disorder include the serotonergic (8–10), dopaminergic (10), histaminergic (11), and endogenous opioid systems (12).
Although neurological examination of a group of 28 children with autistic disorder revealed no individual with a movement disorder (13), there are many reports of adults and children with autistic disorder who manifest movement disorders. Some adults with autistic disorder exhibit co-ordination abnormalities, including mild clumsiness and mild cerebellar ataxia; gait abnormalities, such as decreased ankle range of motion, and balance abnormalities (14). Children with autistic disorder have demonstrated abnormalities in the development of the normal sleep–wake cycle (15), the determination of hand dominance, and the utterance of meaningful words, which have been hypothesized to be mediated by the serotonergic neurons of the brainstem (9,10). Children with autistic disorder have also demonstrated dysdiadochokinesia, abnormalities in crawling and stepping, absent or poor induction of optokinetic nystagmus, difficulty in voluntary eye closure, mouthing phenomenon, and absence of the glabellar tap. Children with autistic disorder have also exhibited the presence of pathologic reflexes including the toe grasping reflex, the tonic foot reflex, and the palmmontal reflex (10). Motor abnormalities observed in children with autistic disorder (10) include akathisia, tics, self-injurious behavior, and other movement disorders (8,16–22). In particular, stereotypies, patterned and repetitive movements, posture, and utterances, have been associated with autistic disorder. For example, hand-flapping, a behavior in which both hands rhythmically move up and down in association with the forearm with limp wrists as the elbows are repeatedly flexed and extended, is uniquely seen in people with autistic disorder. Thus, we propose that some movements are pathognomonic of autistic disorder (21).

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