

Motor Stereotypies

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Stereotypic movements are ubiquitous, occur in a variety of forms, and exist in different populations, ranging from individuals with autism to typically developing children. Although such movements are required to be restricted, repetitive, and purposeless, their definition and included activities remain broad and imprecise. Movements are typically classified into 2 groups, primary (physiological) and secondary (pathological), depending upon the presence of additional signs or symptoms. Although some view these movements as behaviors produced to alter a state of arousal, there is increasing evidence to support a neurobiological mechanism. Behavioral and pharmacological therapies have been used with varying effect. Semin Pediatr Neurol 16:77–81 © 2009 Published by Elsevier Inc.

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Terminology

Stereotypies lack a clearly defined terminology; the term is applied to activities involving specific movements (eg, rocking and hand flapping or waving) or more broadly to heterogeneous self-directed, repetitive behaviors, activities, and interests (eg, covering ears, staring at an object, pacing, object fixation, playing in a fixed pattern, picking skin). When describing a movement, frequently used descriptors include involuntary, bizarre, repetitive, rhythmic, coordinated, patterned, and predictable (form, amplitude, and location), but purposeless.^{1,2} As a behavior, especially in the context of autism or developmental delay, its defining features overlap with terms such as repetition, rigidity, invariance, and inappropriateness.³ Other applied diagnostic terminology includes abnormal repetitive behaviors, preoccupations, circumscribed interest patterns, abnormal object attachments, cognitive rigidity, unusual sensory responses, and social communication difficulties.^{4,5} Further confusion is generated by the fact that similar stereotypic movements can occur in typically developing children as well as in those with secondary etiologies such as autistic spectrum disorder, mental retardation, and sensory deprivation. The Diagnostic and Statistical Manual of Mental Disorders (DSM-IV) criteria for stereotypies requires repetitive, nonfunctioning behaviors that are present for greater than 4 weeks and that interfere with nor-

mal activities or result in self injury.⁶ Suggestions have been made to place all clinically significant, repetitive, body-focused behaviors (including trichotillomania, body rocking, self biting, and skin picking) into a separate “body-focused” category.⁷ In neurodevelopmentally delayed populations, the differentiation of stereotypies, tics, repetitive behaviors, and compulsions has been variable and often dependent on the bias of the evaluator.^{8,9} Clearly, more definitive terminology will be required if specific movements/behaviors are to be ascribed to distinct neuroanatomical localizations and biological mechanisms.

Movement Characteristics

Movements labeled as stereotypies have been variable and wide ranging. In infants and young children, they tend to be more complex motor activities (head nodding, arm flapping, finger wiggling, body rocking), whereas in college students they include nail biting, tapping one’s feet or pencils, hair twirling, and smoking. Motor stereotypies typically begin within the first 3 years of life. In a group of normal children with complex motor stereotypies, about 80% began before 24 months of age, 12% between 24 and 35 months, and 8% at 36 months or older.¹⁰ Movements last from seconds to minutes, appear multiple times a day, and are associated with periods of engrossment, excitement, stress, fatigue, or boredom. Each child has his/her own repertoire, which can evolve with time. Nevertheless, several primary movements, such as bilateral flapping or rotating the hands, fluttering fingers in front of the face, flapping/waving arm movements, and head nodding, tend to predominate. These activities frequently occur in conjunction with other movements such as mouth opening, neck stretching, or even a vocalization. They can be

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readily suppressed by a sensory stimulus or distraction, especially in children with normal cognition. Occasionally, patients report that they enjoy performing the movement, although most patients are unaware of its presence. Stereotypies are frequently upsetting to parents because of concerns about disruptions and social stigmatization. They are, however, usually of little concern to the child whose daily routine is rarely affected. The incidence of attention deficit hyperactivity disorder, learning disabilities, or obsessive-compulsive behaviors is higher in typically developing children with complex arm and hand movements.¹⁰ Assumptions that behaviors resolve in early childhood are often incorrect.¹⁰ Several scales have been used for the assessment of stereotypy severity, including the Stereotypy Severity Scale,¹¹ the Repetitive Behavior Scale-Revised,¹² and the Behavior Problems Inventory.¹³

Differentiating Stereotypies

The diagnosis of stereotypies requires the exclusion of other disorders or causes, such as habits, mannerisms, complex motor tics, obsessive-compulsive behaviors, and paroxysmal dyskinesias. Most frequently, stereotypies are misdiagnosed as complex motor tics. Several characteristics are helpful in differentiating these 2 conditions, though both may occur in the same individual. Stereotypies have an earlier age of onset (<3 years) than do tics (mean onset 5-7 years). They are consistent and fixed in their pattern, compared with the frequent addition and subtraction of tics. In terms of body location, stereotypies frequently involve arms, hands, or the entire body, rather than the more common tic locations of the eyes, face, head, and shoulders. Stereotypies are more fixed, rhythmic, and prolonged in duration than tics, which, except for the occasional dystonic tic, are brief, rapid, random, and fluctuating. Stereotypies, in contrast to tics, are not associated with premonitory urges, preceding sensations, or an internal desire to perform. Both occur during periods of anxiety, excitement, or fatigue, but stereotypic movements are also common when the child is engrossed in an activity. Tics and stereotypic movements are both reduced by distraction, but the effect on stereotypic movements is more instantaneous and dramatic. When evaluated with a force-sensitive platform, temporal measures, and spectral analysis, stereotypic movements differ from tics both quantitatively and qualitatively.¹⁴

Pathophysiology

The underlying pathophysiological mechanism for stereotypies in both primary and secondary categories is unknown, with suggestions ranging from psychological concerns to neurobiological abnormalities. The observation of a higher frequency of stereotypic behaviors in situations of altered arousal has led some investigators to suggest that movements act to maintain an optimal state of arousal.¹⁵

Proponents of a psychogenic mechanism tend to suggest the following possibilities:

1. A form of sensory self stimulation or automatic reinforcement (ie, the reinforcer and the behavior are one and the same) in which the stimulation is designed to compensate for a deficit of external arousal, eg, congenital blindness, deafness, autism, or mental retardation,^{15,16} or those that develop when an animal is caged or a human is placed in solitary confinement.
2. An attempt to deplete aversive stimuli, use up excess attentional capacity, or reduce external distractions or demands by channeling thoughts and actions into movements.¹⁷
3. Substitution behaviors to take the place of imaginative activities.¹⁸
4. A component of obsessive-compulsive disorder,¹⁹⁻²¹ general anxiety disorder,²² perfectionism, or impulse dyscontrol.²⁰ These later suggestions are based, in part, on studies of the occurrence of common stereotypies in college students.

Several lines of evidence support a neurobiological basis for stereotypies, including its correlation with the severity of autism and cognitive impairment,²³ association with disorders such as Rett syndrome,²⁴ pharmacological induction in animal models and humans, and abnormal findings on neuroimaging. The precise neuroanatomical localization for motor stereotypies is unknown. Investigators have suggested that these behaviors can involve neural circuits interconnecting the neocortex with the striatum and areas of the basal ganglia.²⁵ Animal studies of drug-induced stereotypies have shown that the ventral striatum, in particular, is a prominent site. Furthermore, the strength of induced repetitive behaviors, as measured by early-response gene assays, correlates with the differential activation of the striatal striosomal compartment.²⁶⁻²⁸ Striosomes in the anterior striatum receive inputs from the orbital frontal cortex and anterior cingulate gyrus and send projections directly or indirectly (via the lateral habenular and pallidum) to the substantia nigra.²⁹

Magnetic resonance volumetric studies have been performed in individuals with primary and secondary motor stereotypies. In nonautistic boys, there were reductions in frontal and temporal white matter and the size of the caudate nucleus.³⁰ In children with autism, stereotypic behaviors negatively correlated with the size of the cerebellar vermal lobules VI and VII and positively correlated with frontal lobe volumes.³¹ In Down syndrome, cerebellar white matter volumes positively correlated with the severity of stereotypic behaviors.³² Stereotypic movements have spontaneously appeared in a patient after meningoencephalitis with bilateral frontoparietal cortical lesions,³³ in patients with frontotemporal dementia,³⁴ and in association with strokes involving either the right putamen,³⁵ the right lenticular nucleus,³⁶ or bilateral paramedian thalamic and midbrain regions.³⁷

Although numerous neurotransmitter systems coexist within corticostriatal pathways, evidence suggests involvement of the dopaminergic system. In rodent models, repetitive sequences of behaviors, such as sniffing, chewing, rearing, or grooming, can be induced in response to low doses of stimulants (release dopamine) and cocaine (block dopamine

reuptake).³⁸⁻⁴⁰ Stereotypic behaviors characterized by a fascination with repetitive, meaningless movements (punding) have been linked to the stimulation of dopamine receptors with levodopa, dopamine agonists, and, rarely, dopamine receptor blockers.^{41,42} Plasma concentrations of homovanillic acid, a dopamine metabolite, are reduced in adults with high rates of body rocking.⁴³ Finally, using the eye-blinking rate as a noninvasive indicator of dopamine function, the mean eye-blink rate was lower in adults with stereotypic behaviors studied in a state mental facility.⁴⁴

A pattern of Mendelian inheritance has been suggested for primary stereotypies based on a report of 100 typically developing children with complex motor stereotypies.¹⁰ Seventeen had a first-degree relative (parent or sibling) with similar movements, and 25 had at least 1 relative with motor stereotypies. Family history was confined to either the maternal or paternal side, discouraging a suggestion of sex linkage. A relatively equal distribution occurred between maternal and paternal lines. A parametric linkage analysis is currently in progress.

Classification

Primary

Stereotypies have been reported in multiple studies of typically developing children.^{2,10,19,45-50} This category implies that there is no specific cause for the stereotypy as it occurs in an otherwise normal individual, although mild delays in either language or motor development may be present.^{2,10,47} Primary stereotypies are classified into 3 groups: common behaviors (eg, rocking, head banging, finger drumming, pencil tapping, hair twisting) and 2 forms with atypical or complex behaviors—head nodding and complex motor movements (eg, hand and arm flapping/waving), respectively. Precise estimates of the prevalence of stereotypic movements in the typically developing child are unknown. About 20% of healthy children have been estimated to exhibit stereotypies, with most being of the common type.⁵⁰ The outcome of stereotypies in this group has been controversial, declining in some studies after age 4,⁴⁹ persisting in others,¹⁰ and seen in adults who are bored or stressed.⁵¹

Common Stereotypies

Behaviors such as thumb sucking, nail/lip biting, hair twirling, body rocking, self biting, and head banging, sometimes called habits, are relatively common in childhood, and generally, most regress.^{46,52,53} Some children have an evolving pattern of stereotypic behaviors with thumb and hand sucking at a younger age, replaced by body rocking and head banging and later by nail biting and finger and foot tapping.⁵⁴ Investigations of stereotypies in college students have identified a variety of common movements (touch face; play with hair, pens, or jewelry; shake leg; tap fingers; scratch head), but most were not time-consuming or disruptive.²⁰ The prevalence of body rocking has varied from 3% to 25%, depending on the identifying methodology.⁵⁵ Stereotypies in college students are often accompanied by general distress, anxiety, obsessive-compulsive symptoms, and impulsive aggressive

traits.¹⁹⁻²¹ Whether body rocking should be considered a separate entity, based on a high frequency in first-degree relatives with similar movements and without evidence of secondary issues,^{19,22} is controversial.

Head Nodding Stereotypies

Rhythmic, regular head movements (either a side-to-side “no” movement, an up-and-down “yes” movement, or a shoulder-to-shoulder movement) with a frequency of 1-2 seconds, that can be stopped voluntarily have been reported in normal children as a form of stereotypy.^{10,56} Upgaze eye deviations or movements of the hands/feet occasionally accompany the head shaking. Although these movements may occur in typical children, secondary etiologies have included a congenital brainstem-cerebellar abnormality, Sandifer syndrome, spasmus nutans, bobble-head doll syndrome, congenital nystagmus, oculomotor apraxia, and jactatio capitis nocturna. In a study following 8 children with typical development and head nodding, 3 children stopped entirely after exhibiting symptoms for less than 6 months. These data suggest that outcomes in normal children with head nodding may differ when compared with those in children with complex motor stereotypies.¹⁰

Complex Hand and Arm Movement Stereotypies

Movements in this group include hand shaking, posturing, flapping or waving, opening and closing of the hands, finger writhing, arm flapping, and flexion and extension of the wrists. They may occur in conjunction with other activities (eg, body rocking, leg shaking or kicking, facial grimacing, mouth opening, neck extension, and involuntary noises), but the hand/arm movements are dominant.^{10,47} Although several small studies have attempted to compare stereotypic movements of children in the general population with those in children with autism,^{49,57} most investigators suggest that the complex stereotypies seen in typical children can be prolonged, include complex motor patterns, and resemble those in the autistic population.

In a study involving 90 children with complex motor stereotypies, the gender ratio was 1.8:1 (male:female), and onset of movements occurred before age 3 in 91%.¹⁰ The duration of movements was less than 10 seconds in 37%, 11-60 seconds in 27%, and more than 1 minute in 37%. Complex movements occurred more than once a day in about 80%. Movements were exacerbated when the child was excited/happy (86%), focused/engrossed (34%), anxious/stressed (27%), and tired/fatigued (21%); more than 1 trigger was commonly identified. When distracted, in all but a single case, the movement ceased. In 57 subjects aged 7 years and older, 32% had deficit hyperactivity disorder, 16% had tics, and 9% had obsessive-compulsive behaviors. In terms of long-term outcome, consistent with a prior report,⁴⁷ most motor stereotypies persisted, that is, only 3% stopped; of those that continued, 28% were better, 61% remained stable, and 11% worsened.¹⁰ Hence, suggestions to parents that complex motor stereotypies are brief and transient appear incorrect.

Secondary

The secondary stereotypies category implies the presence of an additional diagnosis with behavioral or neurological signs and symptoms, including autistic spectrum disorder, mental retardation, sensory deprivation, Rett syndrome, neurodegenerative disorders, inborn errors of metabolism, drug-induced conditions, infection, tumor, or psychiatric conditions.

As discussed under the terminology section, repetitive behaviors are a major diagnostic feature of autistic disorder, ie, "restricted repetitive and stereotypic patterns of behavior, interests, and activities."⁶ Children with autism have more stereotypies than do equally delayed children without autism,^{23,58} and their severity and frequency positively relate to severity of illness,^{12,59} cognitive deficiency,^{23,60,61} impairment of adaptive functioning,^{62,63} and symbolic play.¹⁸ On the basis of the scoring of motor stereotypies observed on videos of standardized play sessions, children with low functioning autism had a greater prevalence and an increased number and variety of tics compared to those with high functioning autism, developmental language disorder, and non-autism-related low intelligence quotient.²³ The authors further suggested that the stereotypy of gazing atypically at fingers or objects is rare and virtually limited to the autism group. Other investigators, however, have suggested that visual fixation/staring at objects was more common in children with developmental delay than autism,⁶⁴ and "hands to ears" (abducting and externally rotating the arms with the hands close to ears) was more common in children with autistic spectrum disorder than controls.⁶⁵ A videotaped review of play sessions has shown that children with developmental delay exhibited "more bizarre" movements than nonhandicapped children.⁵⁷ Nevertheless, despite these suggestions of a possible behavioral marker, most researchers emphasize the considerable overlap in repertoire of stereotypic movements among autistic, mentally retarded, and typically developing children.

Therapy

Evidenced-based therapy for the suppression of motor stereotypies is generally lacking, and the response of stereotypic movements to medications is largely inconsistent. Behavioral interventions in the autistic population^{16,66-68} have been used with varying success. In a small number of typically developing children, the combination of habit reversal and differential reinforcement of other behaviors was beneficial in reducing motor stereotypies.¹¹

Conclusions

Motor stereotypies are common, but poorly understood conditions that require greater scientific investigation.

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