



CLINICAL REPORT

Motor Delays: Early Identification and Evaluation

abstract



Pediatricians often encounter children with delays of motor development in their clinical practices. Earlier identification of motor delays allows for timely referral for developmental interventions as well as diagnostic evaluations and treatment planning. A multidisciplinary expert panel developed an algorithm for the surveillance and screening of children for motor delays within the medical home, offering guidance for the initial workup and referral of the child with possible delays in motor development. Highlights of this clinical report include suggestions for formal developmental screening at the 9-, 18-, 30-, and 48-month well-child visits; approaches to the neurologic examination, with emphasis on the assessment of muscle tone; and initial diagnostic approaches for medical home providers. Use of diagnostic tests to evaluate children with motor delays are described, including brain MRI for children with high muscle tone, and measuring serum creatine kinase concentration of those with decreased muscle tone. The importance of pursuing diagnostic tests while concurrently referring patients to early intervention programs is emphasized. *Pediatrics* 2013;131:e2016–e2027

INTRODUCTION

The American Academy of Pediatrics (AAP) recommends developmental surveillance at all preventive care visits and standardized developmental screening of all children at ages 9, 18, and 30 months.¹ Recently, developmental screening instruments and their clinical interpretations have emphasized the early detection of delays in language and social development, responsive to rising prevalence rates of autism spectrum disorders in US children.² The most commonly used developmental screening instruments have not been validated on children with motor delays.^{3,4} Recognizing the equal importance of surveillance and screening for motor development in the medical home, this clinical report reviews the motor evaluation of children and offers guidelines to the pediatrician regarding an approach to children who demonstrate motor delays and variations in muscle tone. (This report is aimed at all pediatric primary care providers, including pediatricians, family physicians, nurse practitioners, and physician assistants. Generic terms, such as clinician and provider, are intended to encompass all pediatric primary care providers.)

RATIONALE

Gross motor development follows a predictable sequence, reflecting the functional head-to-toe maturation of the central nervous system.

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KEY WORDS

motor delays, development, screening, neurologic examination, early intervention

ABBREVIATIONS

AAP—American Academy of Pediatrics

CK—creatine phosphokinase

CPT—*Current Procedural Terminology*

DCD—developmental coordination disorder

DMD—Duchenne muscular dystrophy

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The guidance in this report does not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

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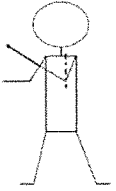
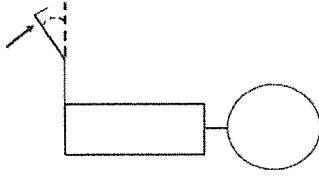
Physical Examination Maneuver	 Scarf sign for tone assessment of shoulder girdle	 Popliteal angle for hamstring tone assessment
Description	With child supine or sitting (if able), pull arm across to the opposite shoulder until resistance occurs. Note elbow position in relation to the bilateral midclavicular line and the midline of the chest.	With child supine, flex the hip to 90°, with child's torso supine on the table. Slowly extend leg at knee until resistance occurs. Note the angle between the lower leg and vertical.
IMPLICATIONS	FINDINGS^a	FINDINGS
Suspect normal tone	The elbow position is between the bilateral midclavicular lines	6° in children 1–3 y; 17°–27° in children 4 y, and 26° in children ≥5 y
Suspect low tone	The elbow crosses the midline to the contralateral midclavicular line	<6° in children ≥1 y
Suspect high tone	The elbow does not cross the ipsilateral midclavicular line	≥50° in children ≥1 y

FIGURE 2

Examples of physical examination maneuvers. Adapted from Mercuri E, Haataja L, Dubowitz L. Neurological assessment in normal young infants. In: Cioni C, Mercuri E, eds. *Neurological Assessment in the First Two Years of Life*. London, UK: Mac Keith Press; 2007:30–31. ^a In term infants, these findings do not change significantly after 3 months of age.

Step 7. Are the History or Examination Results Concerning?

After identifying concerns of motor development, primary care clinicians can perform key diagnostic tests. All testing should be performed in the context of the child's past medical history, including prenatal complications and exposures, perinatal problems, feeding, and growth. Family history is also important to identify any other relatives with developmental or motor issues, recurrent pregnancy loss, stillbirth, or infant death, which may lead to identification of an underlying genetic etiology. Findings on physical examination, such as unusual facial features or other known visceral anomalies, may suggest a specific genetic condition. The state-mandated newborn screening laboratory results should be reviewed, because normal results exclude many disorders and avoid unnecessary testing. Although newborn screening is

comprehensive, it does not test for all inborn biochemical disorders.

The algorithm (Fig 1) can be used to help guide appropriate initial testing. Table 3 lists "red flags" that should prompt the primary care pediatrician to expedite referral to diagnostic resources.

Step 8. High, Normal, or Low Tone?

Step 9a. Consider Neuroimaging

Increased tone in a child with neuro-motor delay suggests an upper motor neuron problem, such as cerebral palsy. The American Academy of Neurology recommends imaging of the brain, preferably by MRI, for patients suspected of having cerebral palsy.²² This test can be ordered within the medical home at the same time the patient is referred for specialist consultation for diagnosis.

Step 9b. Measure Creatine Phosphokinase and Thyroid-Stimulating Hormone Concentrations

When low to normal tone is identified, especially with concomitant weakness, investigations should target diseases of the lower motor neurons or muscles. Among the most common is Duchenne muscular dystrophy (DMD), characterized by weakness, calf hypertrophy, and sometimes cognitive or social delays. DMD usually presents at 2 to 4 years of age, but signs of weakness may be evident earlier. Becker muscular dystrophy is allelic to DMD but typically presents in older children and with a milder phenotype. Initial testing for all children with motor delay and low tone can be performed within the medical home by measuring the serum creatine phosphokinase (CK) concentration. The CK concentration is significantly elevated in DMD, usually >1000 U/L. As an X-linked disorder, there may be a family history of other affected male family members on the maternal side. However, DMD often presents in the absence of a family history for this disorder, with approximately one-third of cases being new mutations.²³ If the CK concentration is elevated, the diagnosis of DMD can usually be confirmed with molecular sequencing of the DMD gene. Other neuromuscular disorders include diseases of the peripheral motor nerves or muscles, such as myotonic dystrophy, spinal muscular atrophy, mitochondrial disorders, and congenital myasthenia gravis. Testing for these diseases should be performed by subspecialists, because these patients often require electrodiagnostic or specific genetic testing. Although congenital hypothyroidism will be identified by newborn screening, acquired hypothyroidism and hyperthyroidism can present in later infancy or childhood with motor delay

TABLE 3 "Red Flags" in the Evaluation of a Child With Neuromotor Delay

Red Flags: Indications for Prompt Referral	Implications
Elevated CK to greater than 3x normal values (boys and girls)	Muscle destruction, such as in DMD, Becker muscular dystrophy, other disorders of muscles
Fasciculations (most often but not exclusively seen in the tongue)	Lower motor neuron disorders (spinal muscular atrophy; risk of rapid deterioration in acute illness)
Facial dysmorphism, organomegaly, signs of heart failure, and early joint contractures	Glycogen storage diseases (mucopolysaccharidosis, Pompe disease may improve with early enzyme therapy)
Abnormalities on brain MRI	Neurosurgical consultation if hydrocephalus or another surgical condition is suspected
Respiratory insufficiency with generalized weakness	Neuromuscular disorders with high risk of respiratory failure during acute illness (consider inpatient evaluation)
Loss of motor milestones	Suggestive of neurodegenerative process
Motor delays present during minor acute illness	Mitochondrial myopathies often present during metabolic stress

and low to normal tone. It is reasonable to perform thyroid function studies (thyroxine [T4] and thyroid-stimulating hormone) as part of the general laboratory evaluation for children with low tone or neuromuscular weakness, even without classic signs of thyroid disease.

Cerebral palsy classically presents with spasticity, dystonia, or athetosis, but may also result in hypotonia. Children with cerebral palsy may have a history of perinatal insult with concomitant abnormalities on brain imaging. Other causes of hypotonia should be considered before the diagnosis of hypotonic cerebral palsy is given to a child with an uneventful perinatal history and normal brain imaging.

DCD may be present when a child's motor coordination performance is significantly below norms for age and intellect, unrelated to a definable medical condition that affects neuromotor function (such as cerebral palsy, ataxia, or myopathy). It can affect gait, handwriting, sports and academic participation, and self-help skills. More than half of individuals with DCD remain symptomatic through adolescence and young adulthood. Intervention, especially task-oriented approaches, can improve motor ability.⁸

Children with neuromotor abnormalities, who also have failure to thrive, growth abnormalities, dysmorphic facial features, or other visceral anomalies, may have a chromosome

abnormality, either common or rare. The American College of Medical Genetics and Genomics recommends microarray testing as the first-line chromosome study.²⁴ Because of the difficulty often encountered in interpretation of results, this test is typically ordered by a subspecialist familiar with this testing. Routine chromosome testing may be appropriate for children with weakness suspected as having recognizable disorders, such as Down syndrome (including mosaic Down syndrome), Turner syndrome, and Klinefelter syndrome. Fragile X syndrome is the most common inherited cause of cognitive impairment, and children with fragile X syndrome may have some element of motor delay. Genetic testing for fragile X syndrome should be considered in both boys and girls, whether they have dysmorphic facial features or a family history.

Common genetic conditions may present with early motor delays (Table 4). The 22q11.2 deletion syndrome (velocardiofacial syndrome) may present with hypotonia and feeding disorder in infancy and delayed motor milestones.²⁵ Noonan syndrome is also a common disorder, and although it is classically associated with short stature, webbed neck, ptosis, and pulmonary stenosis, the phenotype is highly variable, and developmental delays, especially motor delays, are common. Noonan

syndrome is genetically heterogeneous and may be caused by mutations in genes in the *ras* pathway.²⁶ Neurofibromatosis type 1, associated with mutations in the *NF1* gene, can lead to developmental delays and hypotonia in infancy and early childhood. This condition should be suspected in children with hypotonia and multiple (greater than 6) café au lait spots.²⁷ Children with known or suspected genetic disorders may benefit from genetic consultation and genetic counseling for the family.

Step 10. Refer to Early Intervention/Child Find, and Consult/Refer to Appropriate Pediatric Subspecialists, and Perform Remainder of Bright Futures Health Supervision Examination

Observation

Mild abnormalities that are not accompanied by "red flag" findings (red flag conditions necessitate prompt referral) may be closely followed through "observation," but a plan for new or worsening symptoms as well as a time-definite follow-up plan must be developed. Families should understand that clinical changes should prompt urgent reevaluation. This includes regression of motor skills, loss of strength, or any concerns with respiration or swallowing. This ensures that the progressive disorders are brought to medical attention immediately.

TABLE 4 Common Genetic Disorders for Which Neuromotor Delays May Be a Presenting Feature

Condition	Inheritance	Clinical Testing	Clinical Caveats
Angelman syndrome	Sporadic	Methylation testing for Prader-Willi/Angelman syndrome critical region, gene sequencing of UBE3A gene	Infantile hypotonia and delayed motor milestones, usually present with global delays; dysmorphic features are subtle in infancy.
Chromosome disorders	Many sporadic; high recurrence risk for unbalanced translocations if 1 parent has a balanced translocation	Chromosome analysis, single nucleotide polymorphism microarray	Some patients will have multiple anomalies and will have global developmental delays. Some may present in infancy or early childhood with delayed motor and/or speech milestones. Chromosome mosaicism also seen.
Down syndrome Klinefelter syndrome Rare deletions and duplications			
Deletion 22q11 syndrome (velocardiofacial syndrome)	Autosomal dominant (most cases new mutations)	Fluorescence in situ hybridization (FISH) for deletion 22q11.2	90% of cases new mutations. Feeding and speech disorders and cognitive impairment also seen. >50% will have a congenital heart defect.
DMD Becker muscular dystrophy	X-linked recessive	CK, sequencing of dystrophin gene	Becker and Duchenne muscular dystrophies are caused by mutations in different regions of the dystrophin gene. Becker muscular dystrophy has a later onset of symptoms with a less severe course; 67% of cases are inherited, 33% are new mutations.
Fragile X syndrome	X-linked	Gene sequencing and methylation analysis of FMR1 gene	Usually have global delays and cognitive impairment but may present in infancy or early childhood with predominantly motor delays. Males affected primarily, but females with FMR1 expansions may also be affected.
Mitochondrial myopathies	Autosomal recessive; X-linked recessive mitochondrial inheritance	Constitutional and mitochondrial genetic testing, lactate/pyruvate levels and ratio, serum amino acids	Genetic heterogeneity. May not present in infancy. Also at risk for cardiomyopathy, vision loss, hearing loss, cognitive disabilities.
Myotonic muscular dystrophy	Autosomal dominant	Gene sequencing for DMPK gene	May see anticipation with progression of phenotype in subsequent generations.
Neurofibromatosis type 1 (NF1)	Autosomal dominant	Usually a clinical diagnosis, gene sequencing NF1 gene	50% new mutations. Hypotonia most evident in infancy and early childhood. Suspect NF1 if hypotonia seen with multiple café au lait spots.
Noonan syndrome	Autosomal dominant	Gene sequencing for PTPN11 gene, genetically heterogeneous and multiple gene sequencing panels are available	Genetic heterogeneity. Commonly associated with short stature, ptosis, learning and developmental delays, hypotonia, pulmonary stenosis, cryptorchidism, cardiomyopathy.
Prader-Willi syndrome	Sporadic	DNA methylation testing for Prader-Willi/Angelman syndrome critical region	Hypogonadism, especially in boys. Hypotonia most evident in infancy and may be profound.
Spinal muscular atrophy, including congenital axonal neuropathy, Werdnig-Hoffmann disease, Kugelberg-Welander disease	Autosomal recessive	Gene deletion or truncation studies for SMN1 gene (95% to 98% of cases)	Usually presents in early infancy with severe hypotonia. Milder forms identified at later ages.

Depending on the nature of the suspected condition and the age of the child, it may be appropriate to have the child return to his or her medical home for a follow-up visit before the next Bright Futures health supervision visit. This will afford the opportunity

for an interval review of noted symptoms, new concerns, and changes in physical examination or other developmental findings.

Education with the family should not be overlooked or delayed, as a suspected condition can cause significant

anxiety.²⁸ Although the discussion may not be as in-depth as a situation in which diagnostic studies or referral is involved, families deserve a cogent and appropriate discussion of the findings that are being evaluated and what developmental trajectory is expected.

This may help assuage fears and increase compliance with follow-up plans.

Resources

All children with suspected neuromotor delay should be referred to early intervention or special education resources. Additionally, concurrent referrals should be made to physical and/or occupational therapists while diagnostic investigations are proceeding.²⁹ Even when a specific neuromotor diagnosis has not been identified, children with motor delays benefit from educationally and medically based therapies.

Each medical home must develop its own local resources and network of subspecialists for assistance with the diagnosis and management of young children with suspected motor delay. Depending on the setting, such subspecialists may include neurologists, developmental pediatricians, geneticists, physiatrists, or orthopedists. In some areas, availability of these resources may be limited, and waiting times may be long.³⁰ Direct physician-to-physician communication is recommended when red flags are identified (Table 3). Sharing digital photographs via a secure Internet connection may further expedite evaluations. However, the absence of red flags does not rule out the presence of significant neuromotor disease, and all children with motor delays should be thoroughly and serially evaluated.

Step 11. Is a Developmental Disorder Identified?

If a developmental disorder is identified, the child should be identified as a child with special health care needs, and chronic-condition management should be initiated (see Step 12b).

Step 12a. Ongoing Developmental Monitoring

If a developmental disorder is not identified through medical and developmental evaluation, the child should be

scheduled for an early return visit for further surveillance, as mentioned previously. More frequent visits, with particular attention paid to areas of concern, will facilitate prompt referrals for further evaluation when indicated.

Step 12b. Identify as a Child With Special Health Care Needs and Initiate Chronic Condition Management

When a child has delays of motor development, that child is identified as a child with special health care needs even if that child does not have a specific disease etiology. Children with special health care needs are defined by the Department of Health and Human Services, Health Resources and Services Administration, Maternal and Child Health Bureau as "...those who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally."³¹

Children with special health care needs benefit from chronic-condition management, coordination of care, and regular monitoring in the context of their medical homes. Primary care practices are encouraged to create and maintain a registry for the children in the practice who have special health care needs. The medical home provides a triad of key primary care services, including preventive care, acute illness management, and chronic-condition management. A program of chronic-condition management provides proactive care for children and youth with special health care needs, including condition-related office visits, written care plans, explicit comanagement with specialists, appropriate patient education, and effective information systems for monitoring and tracking. Management plans should be based on a comprehensive

needs assessment conducted with the family. Management plans should include relevant, measurable, and valid outcomes. These plans should be reviewed and updated regularly. The clinician should actively participate in all care-coordination activities for children with identified motor disorders. Evidence-based decisions regarding appropriate therapies and their scope and intensity should be determined in consultation with the child's family, therapists, pediatric medical subspecialists, and educators (including early intervention or school-based programs).

Children with established motor disorders often benefit from referral to community-based family-support services, such as respite care, parent-to-parent programs, and advocacy organizations. Some children may qualify for additional benefits, such as supplemental security income, public insurance, waiver programs, and state programs for children and youth with special health care needs (Title V). Parent organizations, such as Family Voices, and condition-specific associations can provide parents with information and support and can also provide an opportunity for advocacy.

RESOURCES

Internet resources are available (www.childmuscleweakness.org) for clinicians to view both typical and atypical motor findings. The identification of motor delays (or any chronic condition) in a child can trigger significant psychosocial stress for families.³² The effects of repeated medical visits, testing, and modifications to home and school environments can place a significant burden on even well-functioning families.³³ Appropriate psychological support should be implemented early. A consumer health librarian or medical librarian can be used by families to provide specific resources tailored to

individual needs (<http://www.nlm.nih.gov/medlineplus/libraries.html>).

For conditions with genetic basis or implications for family planning, medical genetics consultation and genetic counseling should be recommended. An international directory of genetics and prenatal diagnosis clinics can be found at <http://www.ncbi.nlm.nih.gov/sites/>

GeneTests/. Additional Web sites, such as www.rarediseases.org, offer information for both physicians and families.

Information on financial assistance programs should also be provided to families of children with established developmental disorders. They may qualify for benefits, such as supplemental

security income (<http://www.ssa.gov/pgm/ssi.htm>), public insurance (<http://www.medicaid.gov>), and Title V programs for children and youth with special health care needs (<http://internet.dssc.uic.edu/dsccroot/titlev.asp>). There also may be local community programs that can provide transportation and other assistance.

TABLE 5 CPT Codes for Developmental Screening

Services/Step in Algorithm	Notes	CPT Code	Comments
Pediatric preventive care visit	All preventive care visits should include developmental surveillance; screening is performed as needed or at periodic intervals	99381–99394 (EPSDT)	
Developmental/medical evaluation: Office or Other Outpatient Services Codes; New Patient	If performed by the physician as a new patient outpatient office visit	99201-99205	99204 is used for evaluations performed by the physician that are detailed and moderately complex or take at least 45 min (with more than half spent counseling); 99205 is used for evaluations that are comprehensive and highly complex or take 60 min (with more than half spent counseling)
Developmental/medical evaluation: Office or Other Outpatient Services Codes; Established Patient	If performed by the physician as an outpatient office visit	99210–99215	99214 is used for evaluations performed by the physician that are detailed and moderately complex or take at least 25 min (with more than half spent counseling); 99215 is used for evaluations that are comprehensive and highly complex or take 40 min (with more than half spent counseling)
Developmental/medical evaluation/Office or Other Outpatient Consultations Codes	If performed by the physician as an outpatient office visit	99241–99245	99244 is used for “moderate activities” of up to 60 min (with more than half spent counseling); 99245 is used for “high” activity of up to 80 min (with more than half spent counseling)
Developmental screening	Does not require any physician work; rather, clinical staff can score results and provide to physician for interpretation as part of E/M	96110	Reported in addition to E/M services provided on the same date
Developmental testing	Used for extended developmental testing typically provided by the medical provider (often up to 1 h), including the evaluation interpretation and report	96111	Reported in addition to E/M services provided on the same date
Identify as a child with special health care needs, and initiate chronic condition management	Children with special health care needs are likely to require expanded time and a higher level of medical decision-making found in these “higher-level” outpatient codes; these codes are appropriate for services in the office and for outpatient facility services for established patients; these codes may be reported using time alone as the factor if more than half of the reported time is spent in counseling	99211–99215	As above
Prolonged services	At any point during the algorithm when outpatient office or consultation codes are used, prolonged physician service codes may be reported in addition when visits require considerably more time than typical for the base code alone; both face-to-face and non-face-to-face codes are available in CPT	99354 99355 99358 99359	99354 for first 30–74 min of outpatient face-to-face prolonged services 99355 for each additional 30 min 99358 for first 30–74 min of non-face-to-face prolonged services 99359 for each additional 30 min

E/M, evaluation and management; EPSDT, Early and Periodic Screening, Diagnostic and Treatment program.

DEVELOPMENTAL SCREENING BILLING AND CODING

Separate *Current Procedural Terminology* (CPT) codes exist for developmental screening (96110: developmental screening) and testing (96111: developmental testing) when completing neuromotor screening and assessment. The relative values for these codes are published in the Medicare Resource-Based Relative Value Scale and reflect physician work, practice expenses, and professional liability expenses. Table 5 outlines the appropriate codes to use when billing for the processes described in the algorithm. Billing processes related to developmental screening and surveillance should be carefully reviewed to ensure that appropriate CPT codes are used to document screening procedures and ensure proper payment. CPT code 96110 does not include any payment for medical provider services. The expectation is that a nonphysician will administer the screening tool(s) to the parent and score the responses. The physician reviews and interprets the screening results; the physician's work is included in the evaluation and management code used for the child's visit. The preventive care (or new, consultative, or return visit) code is used with

the modifier 25 appended and 96110 listed for each screening tool administered. The CPT code 96111 includes medical provider work. This code would more appropriately be used when the medical provider observes the child performing a neuromotor task and demonstrating a specific developmental skill, using a standardized developmental tool.

CONCLUSIONS

The initial responsibility for identifying a child with motor delay rests with the medical home. By using the algorithm presented here, the medical home provider can begin the diagnostic process and make referrals as appropriate. Both during and after diagnosis, communication between the medical home and subspecialists is important,³⁴ and the medical home should remain fully engaged with the child's care as an integral part of chronic-condition management.

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