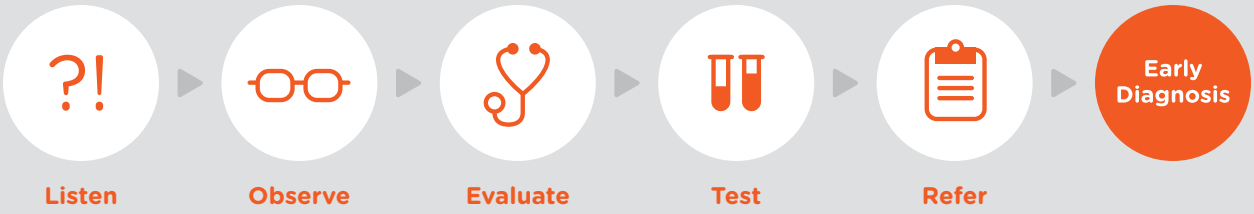
Early diagnosis makes a difference.

Learn the steps to identify pediatric muscle weakness and signs of neuromuscular disease.



Guide for primary care providers includes:

Surveillance Aid: Assessing Weakness by Age
Clinical Evaluation for Muscle Weakness
Developmental Delay, Do a CK
Motor Delay Algorithm

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Questions and comments to: info@ChildMuscleWeakness.org

Surveillance Aid: Assessing Weakness by Age

When it comes to child muscle weakness, early diagnosis makes a difference. Carefully monitoring motor development can help you identify weakness earlier, speeding diagnosis of pediatric neuromuscular disorders and access to treatment.

This surveillance aid will facilitate the identification of motor weakness that presents between birth and five years of age. According to the American Academy of Pediatrics and *Bright Futures* guidelines, providers should incorporate developmental surveillance at every health supervision visit. Developmental screening is recommended at the 9-month, 18-month, and 24- or 30-month health supervision visits, or when surveillance raises a concern. This aid is designed for children who were born on or after 38 weeks of gestation. If a child is born prior to 38 weeks of gestation, please use adjusted age for developmental milestones.

The assessment of milestones recommended in this aid is consistent with the *Bright Futures* guidelines. The exception is "Rise to Stand from Floor," which is not included as a *Bright Futures* milestone but is a quick, easy, and important way to identify motor weakness. See the *Motor Delay Algorithm* on page 10 for guidance if milestones are not achieved.

Milestone: Pull to Sit (Infant+)

Description	Evaluate pull to sit with attention to head lag, until achieved.
Surveillance & Next Steps	If a child has head lag at four months, carefully evaluate other age-appropriate motor milestones (e.g., rolling) and refer to early intervention for developmental stimulation. Re-evaluate in one month. If child is still not age-appropriate, CK test and referral are recommended. See the *Motor Delay Algorithm*, page 10.
Developmental Norms	50% by 3.5 months 75% by 4 months 90% by 6.5 months *Source: Denver II*

Milestone: Sitting (6+ Months)

Description

Evaluate sitting without support at 6-month visit and getting into sitting position at 9-month visit, or until achieved.

Surveillance & Next Steps

A child who is not sitting independently by 7 months or getting into a sitting position at 9 months should receive a CK test and referral. See the *Motor Delay Algorithm* on page 10.

Developmental Norms

Sitting without Support	*Getting into Sitting Position*
50% by 5.9 months	50% by 8.5 months
75% by 6.7 months	75% by 9.5 months
90% by 7.5 months	90% by 10 months
Source: WHO Motor Development Study	*Source: Denver II*

Milestone: Gait (12+ Months)

Description

Watch child walk after parents report s/he can walk independently. Watch or ask about ability to run at 18-month and 24-month visits or until running is achieved. Ask about any concerns with walking, running, or frequent falls at all visits after milestone is achieved.

Surveillance & Next Steps

Walking

If a child does not walk at 15 months, consider referral for early intervention and physical therapy for developmental stimulation, taking into account overall motor development, and re-evaluate within 2–3 months. A child who does not walk well at 18 months, or shows regression in ability to walk, needs further evaluation and should receive a CK test and referral. See the *Motor Delay Algorithm* on page 10.

Running

If a child does not run at 20 months, consider referral for early intervention and physical therapy for developmental stimulation, taking into account overall motor development, and re-evaluate within 2–3 months. Particularly note the quality of running, especially if there are other motor concerns. A child who does not run at 24 months, or shows regression in ability to run, needs further evaluation and should receive a CK test and referral. See the *Motor Delay Algorithm* on page 10.

Developmental Norms

Walking Alone

50% by 12 months
75% by 13.1 months
90% by 14.4 months

Source: WHO Motor Development Study

Running

50% by 16 months
75% by 18.5 months
90% by 21 months

Source: Denver II

Milestone: Rise to Stand (12+ Months)

Description	Watch for independent rise from floor from a supine position after child is able to walk well without assistance (generally 12–16 months, see above). Watch to see if child uses a Gowers maneuver (full or modified, by putting hands even briefly on knees or thighs) or cannot rise without pulling up. Repeat any time concerns are raised about walking or other motor function, to evaluate for regression.
Surveillance & Next Steps	A child who cannot rise from floor to stand without support (including without using hands on knees or thighs to push up) by 18 months or shows regression in rise to stand should receive a CK test and referral. See the *Motor Delay Algorithm* on page 10.
Developmental Norms	Corresponds with time of independent walking.

Clinical Evaluation of Muscle Weakness

If a child shows signs of muscle weakness during routine assessment of motor milestones, a thorough clinical evaluation is the next step. This guide will help you determine whether the child has delayed motor development and what the next steps should be to determine the cause.

Taking a History

Patient History

Review all developmental milestones, including social and language. Specific questions to ask about motor function include:

- Loss of skills (to evaluate for loss of skills, compare to what the child was doing ¼ of his/her life ago, e.g., if a patient is 12 months, compare to what he/she was doing at 9 months)
- Difficulty keeping up with peers (e.g., when sitting, walking, running, climbing stairs)
- Falling/clumsiness
- Whether child "slips through the hands" when held suspended
- Difficulty feeding

Family History

A negative family history does not rule out a genetic neuromuscular disease. Genetic neuromuscular disorders have a range of inheritance patterns, some of which allow transmission of a gene mutation through unaffected carriers and may result in an affected child with no previous family history. In addition, gene mutations that cause neuromuscular disorders may happen for the first time *(de novo)* in an affected child.

A family history of neuromuscular disease may be critical in your evaluation. A complete family history includes:

- Information about at least 3 generations
- A question about consanguinity in the parents
- A summary question about whether anyone else in the family had muscle concerns or weakness

A three-generation family history in the pediatric context includes information on the child's generation (siblings and cousins), the parents' generation (the parents and their siblings and cousins), and the grandparents' generation (the grandparents and their siblings). Asking about consanguinity (whether the parents are related) is important because this increases suspicion of an autosomal recessive disorder.

Neuromuscular diseases have different inheritance patterns. They may be autosomal dominant (passing from affected parent to child 50% of the time), autosomal recessive (passing from two unaffected, carrier parents to child 25% of the time), or X-linked (passing from an unaffected carrier mother to a child—almost always a son—50% of the time).

For those diseases that are X-linked (such as Duchenne and Emery Dreifuss muscular dystrophies and some of the Charcot Marie Tooth disorders), pay particular attention to the extended maternal family (i.e., ask if the mother and maternal grandmother have brothers, and are there any concerns?).

Doing an Exam

What to Look For

Examination for signs of proximal muscle weakness includes:

- Watching for abdominal breathing or accessory muscle use
- Evaluating for head lag when pulled to sit (also note biceps resistance—failure of child pulling back when pulled to sit) or inability to voluntarily flex neck when supine
- Evaluating whether child "slips through the hands" when held suspended with examiner's hands under the armpits
- Stimulating foot and evaluate force of withdrawal movement
- Watching for difficulty rising from floor (including Gowers maneuver, full or modified)

Look for muscle hypertrophy or atrophy, particularly tongue and calves (calves may also feel abnormally full).

It is important to test for signs of motor weakness over time. If a child does not achieve a motor milestone at the expected time, follow up until the milestone is achieved or until there is sufficient concern or referral. Ask about and evaluate for signs of motor regression.

Muscle Tone vs. Weakness

All weak children are hypotonic, but not all hypotonic children are weak. The exam noted above helps distinguish weakness. A weak child likely has a neuromuscular disorder. Hypotonia by itself does not imply a neuromuscular disorder, but both weakness and hypotonia require referral for evaluation.

Peripheral and Central Causes

Other physical findings may help distinguish neuromuscular disease (peripheral cause, such as Duchenne muscular dystrophy) from diseases of the brain (central cause, such as cerebral palsy). The table on the following page shows general guidelines, though evaluation requires use of clinical judgment related to the child's overall developmental history.

Sign	Peripheral Cause	Central Cause
Chest Size	May be small with bell shape	Usually normal
Facial Movement	Often weak “myopathic” with high arched palate	Usually normal
Tongue Fasciculation	May be present, particularly in SMA	Absent
Muscle Tone	Reduced tone	Reduced tone or increased tone with scissoring
Deep Tendon Reflexes	Decreased or absent	Increased, may have clonus
Gait	Toe walking Waddling Hyperlordotic	Toe walking Hemiparetic Spastic

Ordering Lab Tests

CK Testing

If you suspect neuromuscular weakness, include a CK as part of your evaluation. See the *Developmental Delay, Do a CK* guidance on the next page.

Note of warning: If Transaminases (AST and ALT) are elevated, check CK. Since AST/ALT can come from muscle or the liver, while CK comes only from muscle, this test will help localize the child’s problem and may prevent unnecessary liver tests.

Brain MRI Findings

Brain MRI is not an initial or routine component of the evaluation of a weak child. An abnormal MRI does not exclude a neuromuscular disease. Most neuromuscular diseases have normal MRI of brain, but some have characteristic abnormalities.

Developmental Delay, Do a CK

If a child shows delayed motor development and you suspect a peripheral neuromuscular cause, include a CK test as part of your evaluation. This guide provides indications for CK testing in children between 6 months and 5 years of age as well as information on interpeting results and next steps.

About CK Testing

What is a CK (CPK)?

Creatine phosphokinase (CK) is an enzyme found mainly in the skeletal muscle, but also in the heart and brain. Higher-than-expected serum CK indicates leakage of CK through the muscle membrane, suggesting muscle damage either from cardiac or skeletal muscle. In children, elevated CK almost always reflects skeletal muscle damage.

Rationale for CK Testing

A CK test is a starting point in the evaluation of a child with motor delay, even if cognitive delay is more of a concern, and helps focus further testing and referrals. Benefits of a CK test include:

- CK testing is quick and inexpensive.
- CK results can help clinicians differentiate between various disorders that cause weakness.
- The CK can help distinguish between central (where there is normal CK) and peripheral (where CK may be elevated) causes of motor delay.

There are many neuromuscular conditions where the CK is always elevated from birth (e.g., in Duchenne and Becker muscular dystrophies, and in some of the congenital muscular dystrophies, and some limb girdle muscular dystrophies) and other conditions where CK is mildly elevated or normal (e,g., spinal muscular atrophy, neuropathies, and congenital myopathies).

When to Order a CK Test

In Children with Developmental Delay

Evaluate motor milestones in any child who has mild to moderate developmental delay of unknown etiology. (Note that some children with neuromuscular disorders have non-motor developmental delays; for example, delayed language. This may be the feature that first brings them to clinical attention.) If you have any concerns about motor development, include a CK in your screening process.

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In Children with Motor Delay

Order a CK for a child who has unexplained gross motor delay. Findings that should always trigger a CK include:

- Signs of proximal muscle weakness, including:
  - Slipping through hands when held suspended
  - Inability to voluntarily flex neck when supine or head lag when pulled to sit
  - Difficulty rising from floor (including Gowers maneuver, full or modified)
- Loss of motor milestones
- Isolated gross motor delay without other developmental difficulties

Coding and Reimbursement

ICD code for muscle weakness 728.87. Using this code is the best way to ensure reimbursement for CK testing.

Interpreting CK Results

The amount of CK in the serum is reported in units (U) of enzyme activity per liter (L) of serum. In a healthy adult, the serum CK level varies with a number of factors (gender, race and activity). The normal range is generally up to 250 U/L (units per liter) but varies by laboratory.

CK levels can be mildly elevated (~500 U/L) in neuropathies like Charcot-Marie-Tooth disease or spinal muscular atrophy, or grossly elevated (~3,000 to >30,000 U/L) in Duchenne muscular dystrophy or some other muscular dystrophies. CK levels do not reflect the level of functional impairment.

Elevated CK

Elevated CK warrants prompt referral to neurology. In many specialty clinics, an elevated CK level reduces wait time for consultation. A personal phone call may also be helpful.

During episodes of acute muscle breakdown (i.e., rhabdomyolysis), CK levels can temporarily go off the scale, topping out at 50,000 to 200,000 U/L. This is a medical emergency and patients should be referred to the nearest Emergency Department immediately.

Mildly Elevated CK

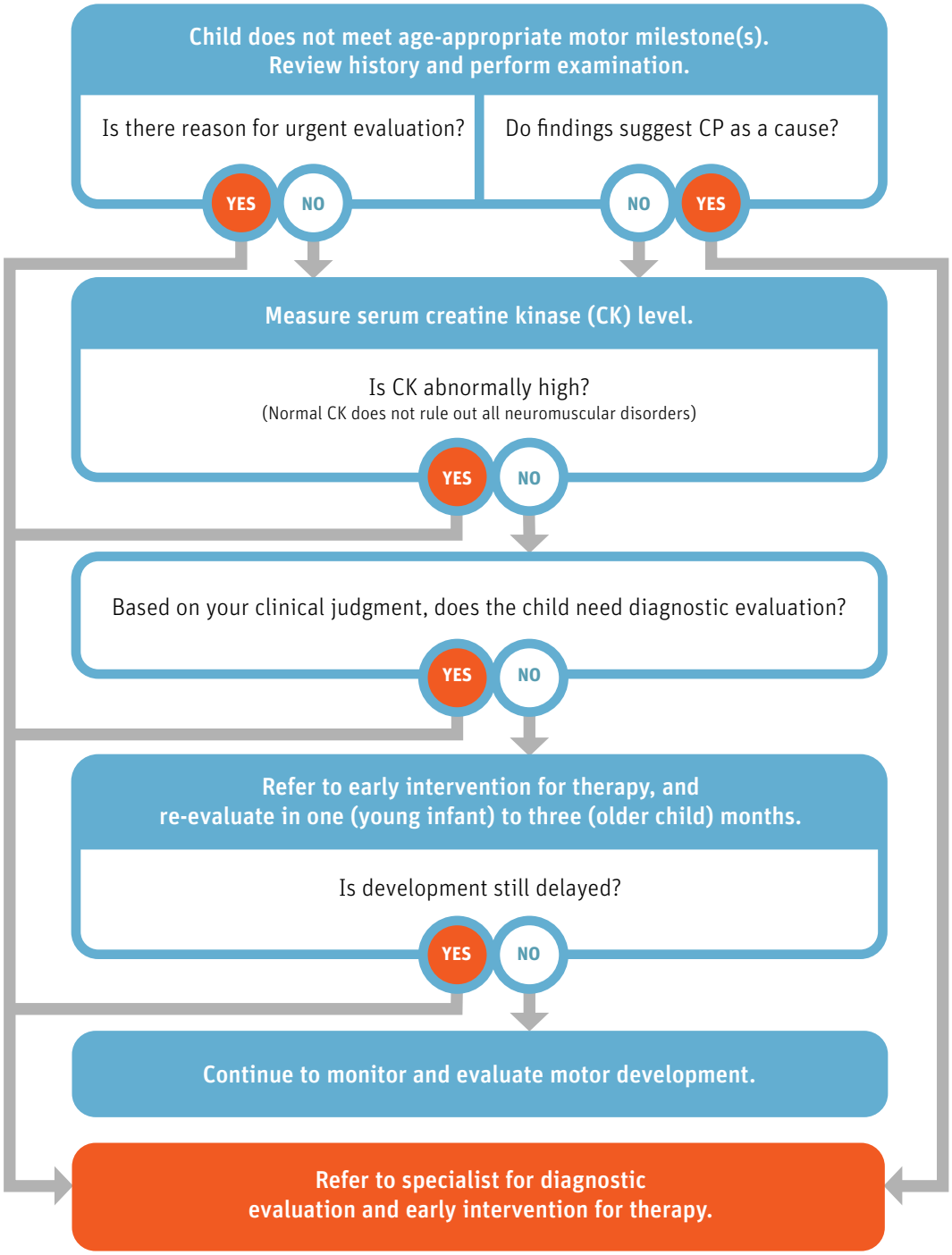
Mildly elevated CK (1–2x normal, <500) should be followed up as it could be temporarily elevated as a result of self-limited conditions such as recent immunization, muscle trauma, or viral infection. Repeat the test after several weeks, and if you have concerns, consult with a neurologist.

Normal CK

Normal CK does not rule out neuromuscular disease. Referrals to specialists, physical therapy, and early intervention are warranted for a child with motor delay, even with normal CK levels.

Motor Delay Algorithm

If a child does not meet age-appropriate motor development milestones, it may be a sign of a serious neurological or neuromuscular disorder. This chart will guide you through the appropriate tests and next steps for referral.



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Urgent Referral

The following factors indicate the need for urgent referral to a neurologist:

- Tongue fasciculation
- Loss of motor milestones
- CK>3x/nl
- Anticipated surgery, due to anesthesia concerns in children with neuromuscular disorders

If a child has an indication for urgent referral, you should consider direct contact with the neurologist's office to share concerns, as this may expedite the appointment.

Note: If a child's urine is the color of cola/tea, especially with muscle pain, send child immediately to the ER for emergency intervention.

Non-urgent Referral

When referral to neurology is not urgent, consider these additional referrals:

Therapy Services (PT, OT, speech therapy) and Early Intervention

- Therapists will perform a more detailed motor evaluation.
- Therapists do not make diagnoses.
- Therapists may detect a need for additional referrals.
- Therapists monitor progress over time.
- In benign forms of motor delay, physical and occupational therapy may promote acquisition of skills.
- Therapists may help parents feel more comfortable working with their children and achieve a greater sense of control.

Developmental Pediatrician or Pediatric Rehab

Referral to developmental pediatrician or pediatric rehab specialty may be an appropriate first referral for patients who do not have an indication for urgent referral.

After Referral

The referring practitioner should continue to evaluate the child's motor development over time, in partnership with therapists and other specialists. Evaluate whether the child improves, stays the same, or regresses. Referral to a neurologist is warranted for children without improvement after 6 months. Appreciate that a diagnosis will inform the care given by therapists and other specialists.