

Clinical Pearls in the Evaluation of the Child with Developmental Delay and Muscle Weakness

1. Red flags for prompt referral to a specialist (consider pediatric neurology) include:

- Tongue fasciculation
- Loss of motor milestones
- CK of greater than 3x normal

Noting one or more red flags will often expedite a neurology referral.

2. Signs of proximal muscle weakness include:

- Abdominal breathing or accessory muscle use
- A feeling of “slipping through hands” when held suspended by examiner under armpits
- Inability to voluntarily flex neck when supine or head lag when pulled to sit
- Difficulty rising from floor (including Gowers maneuver, full or modified)

3. Listen to the parents. 80% of parent concerns are correct and accurate.*

4. Early diagnosis can help target interventions, improve outcomes, reduce family distress related to the diagnostic process, and provide the opportunity for accurate genetic counseling

5. Children can make developmental progress, even with progressive weakness. Developmental progress does not exclude the possibility of an underlying neuromuscular condition.

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Written by Administrator

Friday, 04 February 2011 05:37 - Last Updated Tuesday, 05 February 2013 23:10

6. **Neuromuscular diseases can involve the brain and cognition.**

7. **If you evaluate a child with developmental delay, do a [CK](#).**

8. **Increased CK (especially if CK is greater than 3x normal) indicates the need for prompt referral and diagnosis.**

9. **Children with some neuromuscular disorders have a normal CK.** Normal or near normal CK should not be reassuring if a child is weak or losing milestones.

10. **A negative family history does not rule out a genetic neuromuscular disorder.**

11. **Genetic/genomic tests (such as karyotype, microarray, or sequencing) may diagnose or identify risk for a neuromuscular disorder before there are obvious symptoms.** Urgent telephone consultation with a geneticist or neurologist will inform an appropriate referral.

12. **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.

13. **Toe walking, while often a normal variant, may be an early sign of a neuromuscular condition** or a central condition such as cerebral palsy.

14. **Many neuromuscular conditions increase risk of malignant hyperthermia and other anesthetic catastrophies with anesthesia use.** Anticipated surgery should increase the urgency of a diagnostic evaluation.

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*Source: National Center for Medical Home Implementation, AAP