

Ask the Expert: Pediatric Motor Delays and Hypotonia

General Concerns

Motor delay and hypotonia are common concerns in the pediatric population and often accompany other developmental delays or differences (such as language delay or Autism Spectrum Disorder).

- If a delay is noticed at well child visit and the child has low to normal tone, please consider ordering a CK and TSH.
- If not walking by 15 months, please consider ordering a CK and a TSH.
- If there are concerns for motor delay/hypotonia and the child is incidentally found to have elevated transaminases, please consider ordering a CK and repeat transaminases.

Duchenne Muscular Dystrophy

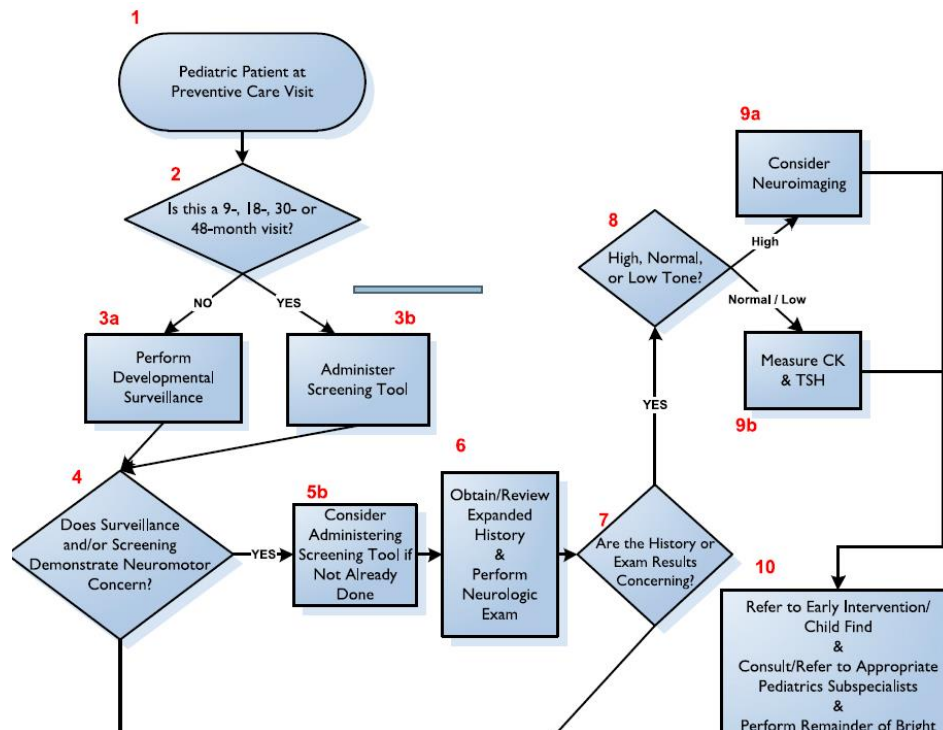
Duchenne Muscular Dystrophy is a “common” rare disease, impacting 1/3500 boys in the US.

- Standard of care is steroids. <https://www.parentprojectmd.org/wp-content/uploads/2018/03/PJ-Nicholoff-Steroid-Protocol.pdf>
- Emerging therapies include exon skipping and gene-replacement, which require close follow-up to monitor for liver damage, immune mediated myositis, and myocarditis.
- More states offering on newborn screen.
- The Hidden Disease: Delayed Diagnosis in Duchenne Muscular Dystrophy and Co-Occurring Conditions: <https://pubmed.ncbi.nlm.nih.gov/35943375/>

Spinal Muscular Atrophy

- On newborn screen in all 50 states.
- However, there are still children with milder forms who were not picked up on newborn screen, so consider in children with low tone and motor differences.
- Now has multiple life-extending therapies including gene replacement and gene enhancing therapies.

AAP Algorithm/Recommendations for PCP Evaluation of Motor Delays/Hypotonia



The information provided in this document is intended as general educational guidance to be considered by you in the specific context of your patients' medical needs. As guidance, it is not intended as a substitute for consultation with a specialist. If you have questions or wish to speak with one of our pediatric specialists at UVA Health Children's Hospital, please call 424-924-KIDS.