



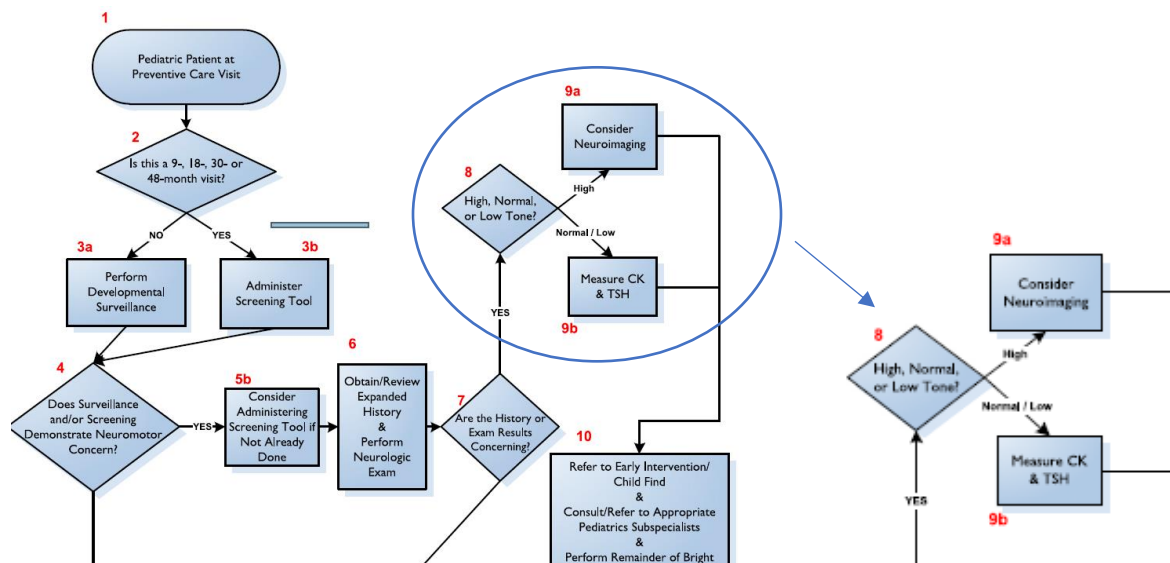
Motor Delays and Hypotonia

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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 visits and children have low to normal tone, please consider ordering a CK and TSH.
- If not walking by 15 months, please consider CK and TSH.
- Elevated Transaminases, please order CK or repeat transaminases.

AAP Recommendations:



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.

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.

[The Hidden Disease: Delayed Diagnosis in Duchenne Muscular Dystrophy and Co-Occurring Conditions - PubMed](#)

“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 1-424-924-KIDS.”