translarna™
ataluren

Because every moment counts

DMD is a progressive disease presenting early in childhood that needs an early and accurate diagnosis[1,5,6]

The following milestones could assist you in identifying a developmental delay in your patient:

0-6 months

- No head control at 2 months[1,2]
- Not making sounds at 4 months[2]
- Not reaching or grasping by 6 months[1,2]
- Not rolling over by 6 months[1,2]

6–18 months

- Not sitting independently at 9 months[2,3]
- Not crawling by 9–15 months[1,4]
- Not speaking first words by 12 months[2]
- Difficulty rising to stand by 18 months[1,2]
- Not walking well by 16–18 months[2,3,7]

2–3 years

- Gowers' sign from 2 years old[8]
- Not walking smoothly at 2 years old (tip-toe walking)[8]
- Not jumping at 2 years old[1]
- Difficulty runningor climbing at 3 years old[1,2]
- Not speaking in sentences at 3 years old[2]

Other signs and symptoms
Elevated serum CK or transaminases[9,10], cognitive delay[7], calf hypertrophy[7,11], abnormal gait[7], frequent falls[7,11]

CK, creatinine kinase; DMD, Duchenne muscular dystrophy.

When you think Developmental delay...
Do a CK![18,19]

A CK test should be carried out in patients with developmental delay[12,13]

Achieving a timely and accurate diagnosis of DMD is a crucial aspect of care[12]

Positive family history with suspicion of abnormal muscle function[12]

Developmental delay, such as difficulty rising to stand, or not walking well by 18 months[12,14–16]

Unexplained increases in transaminases[12]

PERFORM A CK TEST

Elevated CK* (>250 U/L)

Promptly refer to a neuromuscular specialist or geneticist[12,15,17]

Normal CK*

Normal or mildly elevated CK levels do not rule out neuromuscular disease

CK, creatine kinase; DMD, Duchenne muscular dystrophy; L, litre; U, unit. *The normal CK range is generally up to 250 U/L. Absolute values may differ between laboratories.4

References: 1. Noritz GH, et al. Pediatrics. 2013;131:e2016–e2027. 2. Centers for Disease Control and Prevention. Developmental milestones. Available at: https://www.cdc.gov/ncbddd/actearly/pdf/checklists/all_checklists.pdf [Accessed February 2021]. 3. Lurio JG, et al. Am Fam Physician. 2015;91:38–44. 4. van Dommelen P, et al. Dev Med Child Neurol. 2020; doi: 10.1111/dmcn.14623. 5. National Task Force for Early Identification of Childhood Neuromuscular Disorders. Child Muscle Weakness. 2019. Available at: childmuscleweakness.org [Accessed February 2021]. 6. Ciafaloni E, et al. J Pediatr. 2009;155:380–385. 7. Birnkrant D, et al. Lancet Neurol. 2018;17:251–267. 8. Parsons EP, et al. Eur J Paediatr Neurol. 2004;8:145–153. 9. Ardıçlı D, et al. Neuropediatrics. 2019;50:41–45. 10. Counterman KJ, et al. Muscle Nerve. 2020;61:36–43. 11. Aartsma-Rus A, et al. J Pediatr. 2019;2014:305–313.e14. 12. Birnkrant DJ, et al. Lancet Neurol. 2018;17:251–267. 13. National Task Force for Early Identification of Childhood Neuromuscular Disorders. Guide for primary care providers. Available at: https://childmuscleweakness.org/wp-content/uploads/2019/05/PrimaryCareProviderPacket.pdf [Accessed February 2021]. 14. WHO Multicentre Growth Reference Study Group. Acta Paediatr Suppl. 2006;450:86–95. 15. National Task Force for Early Identification of Childhood Neuromuscular Disorders. Child Muscle Weakness. 2019. Available at: childmuscleweakness.org [Accessed February 2021]. 16. Lurio JG, et al. Am Fam Physician. 2015;91:38–44. 17. Aartsma-Rus A, et al. J Pediatr. 2019;2014:305–313.e14.
18. van Ruiten HJ, et al. Arch Dis Child. 2014;99:1074–1077. 19. Birnkrant DJ, et al. Lancet Neurol. 2018;17:251–267. 3. Bushby K,et al. Lancet Neurol. 2010;9:77–93..

for full information please refer to the Israeli PI as approved by MOH

MEDISON

PTC THERAPEUTICS

DUC0004