Good Practice Points for Health Visitors

Duchenne Muscular Dystrophy - recognition, diagnosis and clinical care

What is Duchenne Muscular Dystrophy (DMD)?

- Duchenne muscular dystrophy (DMD) is a genetic condition which causes progressive weakness of most muscles in the body[1].
- DMD affects 1 in 3,500-5,000 boys[1]. This means that, in the UK, about 100 boys are born with DMD each year and about 2,500 boys and men are living with it.
- Affected boys do not have signs of muscle weakness at birth but they usually present with the first manifestations between the age of 18 months and 3 years. As they get older, they show difficulties in walking, running and climbing. With time, all boys lose the ability to walk independently and need help with most activities of daily life.
- The condition also affects the heart and breathing muscles, and life expectancy is significantly reduced.
- DMD can affect any child without anyone in the family being affected. But, in most cases, it is passed on from the mother. It is an X-linked condition, this means that females carry the genetic fault which causes DMD but they usually do not have any muscle weakness. Carriers have a 50% chance of having a son with DMD and 50% chance of having a daughter who is a carrier.
- Female carriers are usually asymptomatic but they are still at risk of developing cardiomyopathy, and cardiac surveillance is required.

Signs and symptoms

- Symptoms depend on the age of the child but typically become clear between the age of 18 months and 3 years[2].
- First signs can be delayed motor milestones:
  - Poor head control (i.e., persistent head lag)
  - Slow to sit, crawl, stand and/or walk
- In some children, there is also speech delay and learning or behavioural difficulties (e.g., autism, ADHD).
- In toddler years, boys often fall frequently and might develop an unusual gait (tiptoe or waddling gait).
- School-age boys typically present with:
  - Unusual gait
  - Difficulty climbing steps (needing bannisters to haul themselves up, due to weakness in legs) and running (i.e., lifting both feet off the floor)
  - Unable to jump (i.e., clearing the floor)
  - Difficulties getting up from the floor - Gowers' manoeuvre (see Fig.1)

Figure 1: This boy* with DMD struggles to get up from the floor, he shows the typical "Gowers' manoeuvre" to get up.

*Parents of children photographed have kindly given the author/s permission to share for educational purposes.

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The information in this resource was updated on 20/04/2023.

Signs and symptoms continued

- **Gowers' manoeuvre**: boys with DMD struggle to get up from the floor quickly due to muscle weakness in their legs. They typically turn around on their tummy first and then push themselves up on their legs before they can stand up (see Fig.1).
- Some children complain of pain or cramps in their legs.
- The calf muscles can look very big (like body builder muscles, see Fig.2).
- Some children can experience episodes of very dark (coca-cola coloured) urine after exercise – this is a sign of muscle damage and can be associated with muscle pains and cramp.
- Some boys are more severely affected than others, but all show a predictable pattern of ***progressive*** muscle weakness.

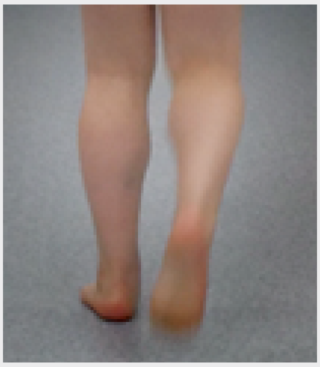

Figure 2: Abnormal big calf muscles

Identification

- Recognising the early signs and symptoms of DMD in children is very important. Aiming for a diagnosis of DMD as early as possible and definitely before the child goes to school:
  - Allows the child to receive prompt clinical care (physiotherapy, corticosteroid treatment, cardiac surveillance) – this is associated with a better long-term outcome[3]
  - Allows the family to understand the genetic implications of the disease and access counselling. Antenatal screening can be offered for future pregnancies to find out if the new baby has DMD before he is born
  - Gives the opportunity to receive advanced therapies and to take part in clinical trials and research with new therapies
  - Allows the family and the child to receive adequate support if required) (e.g., speech and language therapy, educational healthcare plan, behaviour management and treatment)
  - Allows access to genetic counselling to other female family members who might be at risk of being asymptomatic carriers of DMD.
  - Ensures that other family members at risk of developing symptoms are offered adequate and timed surveillance (e.g., cardiac assessments for female carriers)

Investigations

- A blood test, called **creatine kinase (CK)**, can be done as a first step in the diagnosis of DMD. This is a cheap and quick test (results can be available within 24 hours). The CK levels are significantly raised (>1000IU) in all boys with DMD.
- A blood test for CK levels can be done by the GP or local paediatric services.

When to check the CK levels:

- Any boy with motor or general development delay (see red flags below).
- Any boy with abnormal motor skills **at any age**, in particular if associated with speech delay or behavioural difficulties.

Milestone	Red flag age (in months)
Head control	3
Rolling both ways	6
Sitting independently	9
Cruising	15
Walking	18
Running	30
Jumping	36

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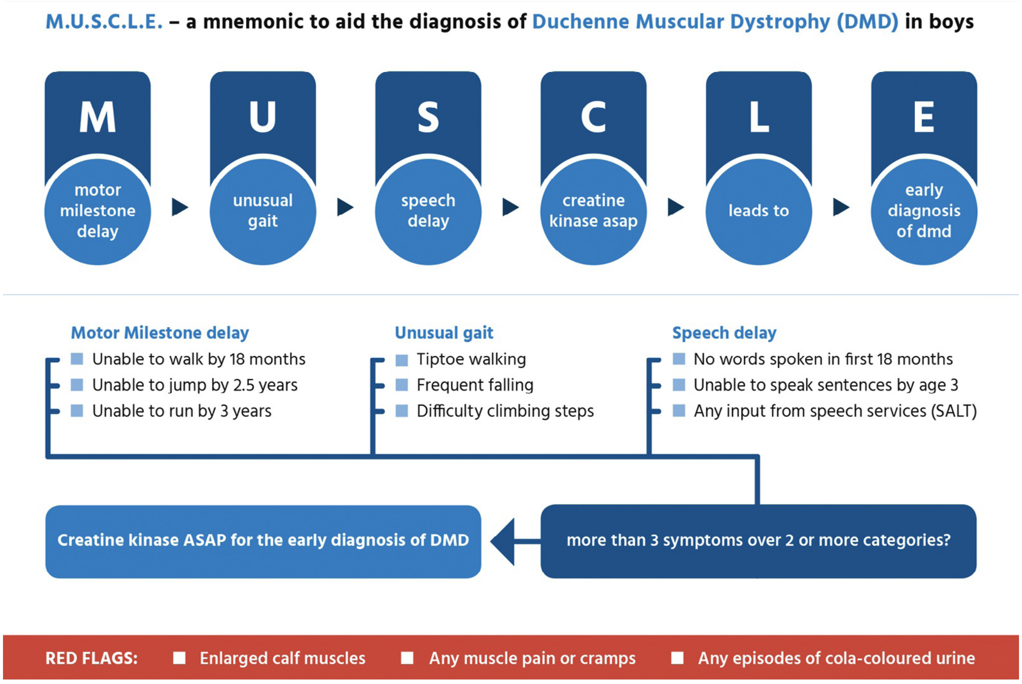
- In summary – remember MUSCLE



M.U.S.C.L.E. – a mnemonic to aid the diagnosis of Duchenne Muscular Dystrophy (DMD) in boys

Motor Milestone delay

- Unable to walk by 18 months
- Unable to jump by 2.5 years
- Unable to run by 3 years

Unusual gait

- Tiptoe walking
- Frequent falling
- Difficulty climbing steps

Speech delay

- No words spoken in first 18 months
- Unable to speak sentences by age 3
- Any input from speech services (SALT)

more than 3 symptoms over 2 or more categories?

Creatine kinase ASAP for the early diagnosis of DMD

RED FLAGS: ■ Enlarged calf muscles ■ Any muscle pain or cramps ■ Any episodes of cola-coloured urine

Referral Pathway

High CK

- Suggests there might be a problem with the child's muscles, this could be DMD:
    - Urgent referral (same day) to the local paediatric services or neuromuscular centre, if available
    - Explain to parents the reason for referral: i.e., the blood test suggests there might be a problem with their child's muscles and therefore you would like to ask for a specialist opinion
    - Please be aware that a high CK does not equal a diagnosis of DMD. This requires further genetic testing.

Normal CK (<200 IU)

- There could still be a problem with the child's muscles or nerves, but normal CK levels exclude a diagnosis of DMD.
- If abnormal motor development or red flags in motor development – refer directly to the local paediatric services.
- If in doubt – discuss with wider team (including GP).
    - Review the child in 4-6 weeks and monitor progress of motor development. If there are still concerns, consider referral to the local paediatric services.

Clinical care

- In the last few years, there has been huge progress in clinical care and development of new therapies for muscle diseases, in particular for DMD.
- Improved clinical care has increased quality of life and life expectancy of boys with DMD. Clinical care involves access to treatments, including: corticosteroids; early physiotherapy input; monitoring of heart and breathing function; and supportive care for muscles and bones[3].
- New treatments might be available for some patients with DMD and more treatments are currently tested in clinical trials.

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Good Practice Points for Health Visitors

- A diagnosis of DMD as early as possible can improve the outcome for affected boys and their families.
- Health visitors (HVs) can play a crucial role in supporting families to access appropriate services for diagnosis.
- Listen to parents' concerns - they may have noticed some signs in their child, such as difficulty in getting up from the floor where they have been playing.
- HVs may see babies and children with symptoms of DMD in their contacts, such as delayed milestones, and taking prompt action can really improve that child's outcomes.
- For help understanding genetic causes of rare diseases and the changes genomics is providing in healthcare, please see our GPP eXtra - Understanding Genomics.
- Remember the MUSCLE mnemonic to help consider DMD and act where concerned using local referral pathways.

Professional Resources

- **Recognising Neuromuscular Disorders: A practical approach**

  An e-learning module which covers child motor development, red flags in a child's motor development, types of neuromuscular disorder, and a plan of action for a child you are concerned about.

  This e-learning was developed for the Royal College of Paediatrics and Child Health (RCPCH) by the authors of this GPP.

  To access this course, visit https://bit.ly/40TZhmL

  The link will guide you through registering and then signing up to the RCPCH - you need to do this in order to access the learning section.

Patient organisations

- Action Duchenne: www.actionduchenne.org
- Muscular Dystrophy uk: www.musculardystrophyuk.org
- Duchenne UK: https://www.duchenneuk.org/

References

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2. Van Ruiten H, Straub V, Bushby K and Guglieri M Improving recognition of Duchenne muscular dystrophy: a retrospective case note review. *BMJ Archives of Disease in Childhood* 2014;99 (12) Available from: https://bit.ly/3LVK9B8 [Accessed 30/03/2023]
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