DUCHENNE & BECKER FREQUENTLY ASKED QUESTIONS

The following information was developed by The Royal Children's Hospital Melbourne (RCH) Neurology department with input from Muscular Dystrophy Australia and RCH consumers and carers to inform parents, families, children, carers and the wider Duchenne and Becker community on answers to questions asked regularly to professionals in the field.

Duchenne muscular dystrophy (DMD)

Muscular dystrophies (MD) are common neuromuscular (nerve and muscle) disorders, characterised by loss of muscle strength and bulk. There are several different types of MD, and each type has its own pattern of muscle weakness and progression over time.

Duchenne (doo-shen) muscular dystrophy (DMD) is the most common type of muscular dystrophy, and it affects about one in 3500 boys. Girls do not usually develop DMD.

DMD is a progressive disease. The muscle degeneration in DMD affects all muscles of the body, including the breathing muscles and the muscles that hold the spine straight. Children affected by DMD generally lose the ability to walk by 12 years of age, after which they need to use a wheelchair.

Signs & Symptoms of DMD

DMD is usually first diagnosed when a child is three to four years old, although symptoms are common earlier than this. Early signs of DMD include:

- toe-walking – children start walking on their tip toes
- larger than normal calf muscles, which is called pseudohypertrophy (see-you-doe-hyper-truh-fee)
- a waddling type of walk
- inability to run or climb stairs
- an unusual way of getting off the floor, called a Gowers sign.

Some children with DMD also have delay in their speech development, and many will not walk until after 18 months of age.

When to see a doctor

Many of the signs of DMD will be picked up in your regular Maternal and Child Health Nurse checks. However, if you are worried about your child's development, see your GP. If the doctor thinks your child may have DMD, you will be referred to a paediatrician or paediatric neurologist (a doctor who specialises in childhood brain and nerve conditions) for diagnosis.

What causes DMD

Problems with a gene called the dystrophin gene causes DMD. Children with DMD often have a family member who has the condition, but it can also happen with no family history.

While girls do not usually develop DMD, they can be carriers, which means they can pass the gene on to their future children.

Most carriers have no symptoms of DMD. However, some carriers will experience symptoms such as muscle pain or weakness. Some female carriers are at risk of heart problems.

Where there is a known family history of DMD, families are encouraged to have genetic testing to find out the risk of having a child with DMD. Your family doctor or neurologist can help organise for you to have this test.

Treatment

Treatment for DMD is aimed at reducing the symptoms of this disorder and keeping your child as healthy and active as possible, for as long as possible.

While there is no cure for this condition as yet, scientists around the world are working toward treatments that may make a significant difference to the outcomes of DMD.

Physiotherapy and occupational therapy

Your child will need to be under the care of an experienced occupational therapist and physiotherapist.

Regular physiotherapy is important to keep your child mobile, healthy and comfortable.

The physiotherapist may design a program including active and passive stretching, assisted stretching and muscle elongation. Splinting, orthotics and standing devices can also help your child stay more active for longer.

Regular medical care

A neurologist will monitor your child's strength and general physical wellbeing, check for scoliosis (curvature of the spine) and monitor your child's breathing (respiratory) and heart (cardiac) function. Surgery can help with spine and foot problems.

As DMD progresses slowly over time, your child's needs will also change. The amount of support and care your child needs will also change. Keep in regular contact and attend appointments with your child's neurologist and physiotherapist. They will help to make sure your child is getting appropriate care and will help you to plan for the future.

Medication

Many medications have been tried in the treatment of DMD. Corticosteroids, such as prednisolone, are the only proven effective medication for children with DMD.

Your neurologist may recommend steroid therapy to help with your child's movement and breathing.

Recent studies have found that a dietary supplement called creatine monohydrate may increase muscle strength in children with DMD.

Care at home

Modifications to your house may be needed over time to accommodate a wheelchair to help your child remain mobile and independent.

Items that can improve your child's comfort and independence include soft pressure care mattresses, ramps and modified taps.

Your child's physiotherapist or occupational therapist can advise you.

Excersise

Gentle exercise and participation in physical activity have both psychological and physical benefits for children with DMD.

Exercise can slow muscle degeneration and help to strengthen your child.

In addition to exercises recommended by your child's physiotherapist, activities like playing at the park, riding a bike, swimming or hydrotherapy help flexibility, muscle strength and confidence.

An increase in strength can improve performance of daily activities such as stair climbing and walking, and also help postural muscles needed to keep the spine straight.

Swimming can still be enjoyed after walking and riding are no longer possible. Regular play and incidental exercise will help keep your child participating in activities with their friends at school. As they become less comfortable walking, a wheelchair or electric scooter may help them to continue to participate in social and sporting activities and maintain independence.

Other benefits of exercise for children with DMD include maintaining range of motion in joints, and preventing spinal curvature (scoliosis) and obesity.
It is important to avoid your child becoming over-exerted or exhausted, however, as this can worsen their muscle damage.

The internet

While the internet can be a source of good information, it can often be a source of false hope, inaccuracies and misleading information.

Always read information on the internet with great scepticism. Rely on your doctor or neuromuscular team for expert and professional advice.

Support

You are not alone. Organisations such as the Save Our Sons Duchenne Foundation and Muscular Dystrophy Australia and your care team are there to provide support, practical advice and comfort.

Make sure you access and keep in regular contact with support people and groups. Parents, siblings and other family may also need regular emotional support.

Key points to remember

- DMD is a progressive disease-causing increasing weakness of the muscles of the arms and legs, the breathing muscles and the heart.
- DMD can be inherited or may occur in only one family member. Genetic testing is recommended, especially if you have a family history of neuromuscular disease.
- It is essential to keep regular appointments with a neurologist, physiotherapist and other health care professionals.
- DMD is a progressive disorder and your child's needs will increase as they get older.

Common questions RCH doctors are asked

- What is Gower's sign?

Gower's sign is an unusual way of standing up from the floor, where a child uses their upper limbs to compensate for weak lower limbs.

A child will push themselves up on their arms and knees, then use their hands to 'walk up' their legs before standing upright.

- Can my child attend a normal kindergarten and school?

Most children with DMD are able to attend mainstream kindergartens and schools, though appropriate adjustments need to be made for their physical and learning needs. Your child will have an Individualised Learning Plan and may qualify for additional support, in the form of an aide or school modifications.

- Are any learning problems associated with DMD?

Up to one third of boys with DMD have a learning problem, but these are unlikely to be significant. There are also increased risks of ADHD, dyslexia and cognitive skills.

For more information

- Save Our Sons Telehealth Nurse on 1300 798 328 or at nurse@saveoursons.org.au
- Your GP, neurologist or neuromuscular nurse coordinator
- Save Our Sons ACTT 2020 Conference Presentations at https://www.saveoursons.org.au/acttconference/
- Share4Rare at: https://www.share4rare.org/library/duchenne-muscular-dystrophy-dmd/what-duchenne-muscular-dystrophy
- Parent Project MD at: https://www.parentprojectmd.org/care/for-families/for-newly-diagnosed/learn-about-duchenne-and-becker/
- Treat NMD at: http://www.treat-nmd.eu/downloads/file/standardsofcare/dmd/uk_english/UK2018FamilyDMDGuide.pdf
- World Duchenne Organization at: https://www.worldduchenne.org/