PAEDIATRIC MUSCLE & NERVE CLINIC

Bangalore Baptist Hospital
QUALITY WITH COMPASSION SINCE 1973

DUCHENNE MUSCULAR DYSTROPHY- DMD:
Information for Families

What is Duchenne Muscular Dystrophy?
Duchenne Muscular Dystrophy or DMD is the most common type of muscular dystrophy, affecting approximately 1 in 3500 newborn boys. It causes muscle weakness in affected children along with calf muscle enlargement. Children with the disease, early on, have a tendency to walk on their toes initially and tend to fall frequently. They have trouble running, climbing stairs, getting up from the floor and jumping. Some boys with the disease also have mild learning and behavioural problems. Affected boys need a wheelchair (the age of wheel chair use is dependant on the proper usage of steroids and MDT to slow down disease progression) as they grow older and develop weakness of breathing muscles; severe enough to shorten their life span. With time, the disease also affects the muscles of the heart.

What causes DMD?
DMD is caused by the lack of production of an important protein called dystrophin. Dystrophin acts as a shock absorber in the skeletal muscle, protecting the muscle fibres from getting damaged when they contract. Defects in the dystrophin gene stop the body from making dystrophin protein, causing weakness in all the muscles of the body that worsen with time as muscles breakdown. The absence of dystrophin means that the body has no way of replacing damaged muscle or growing new muscle cells.

How is DMD inherited?
Because the dystrophin gene is on the X chromosome, boys are affected as they have only one X chromosome and one Y chromosome. Girls meanwhile have two X chromosomes and so even if they have one affected or faulty dystrophin gene, they can compensate with the other normal copy and hence don't show the disease and just remain "carriers" of the disease. This means that the disease can be passed on in families, although occasionally the mutation can arise spontaneously.

Is there any treatment for DMD?
Currently there is no available cure for DMD. For some mutations or specific types of dystrophin defects, the FDA has approved a genetic treatment, which is unavailable in India as of now. This doesn't mean that nothing can be done. A range of options are available for slowing progressive weakness, managing symptoms and pro actively preventing complications and thus improving quality of life. Internationally agreed Standards of Care in DMD outlines these options and emphasises the multidisciplinary nature of the management of affected children.

The first step is often starting corticosteroids at the right age and continuing a good dose as long as needed by pre-empting the expected side effects of steroids. Along with this, the other measures are maintaining bone health, preventing fractures, watching for early signs of involvement of the heart muscle and preventing worsening along with preventing and managing scoliosis (a condition which causes the spine of a growing child with a weak trunk to grow in a deformed way and thus end up compromising the heart and the lungs of the child) and also managing the gradually worsening breathing muscle weakness and supporting breathing. For all this, a protocol based multidisciplinary approach is crucial.

Steroids and DMD:
Till definitive cures are found, steroids are the mainstay in managing DMD. There is a huge disparity in the life expectancy and quality of care enjoyed by children with DMD in India in comparison to those in other developed nations. This difference is mostly attributable to inconsistent administration of steroids to affected children. These internationally established guidelines have made a big difference in the quality of life.

We live in an exciting world full of possibilities. There are many potential cures around the corner for many of these diseases, which were previously thought to be incurable. With the exception of a few small clinical trials, the vast majority of clinical trials are open only to those affected boys who have been treated as per international guidelines and have been on steroids.

Possibilities for cure:
DMD is one of the conditions with a lot of active research and hence it has a substantial number of newer promising therapies that are being tested in clinical trials around the world. New genetic and molecular therapies are becoming a possibility. Some of the areas of promising research can be divided into the following groups:

i. Mutation specific approaches- personalised medicine, exon skipping.
ii. Improving muscle mass
iii. Drug therapy
iv. Gene therapy
v. Cell therapy

Some resources:

1. TREAT-NMD website: http://www.treat-nmd.eu/dmd/overview

2. TREAT-NMD website:
   http://www.treat-nmd.eu/dmd/research-overview/introduction

3. NINDS: The National Institute of Neurological Disorders and Stroke:
   https://www.ninds.nih.gov/disorders/All-Disorders/Muscular-Dystrophy-Information-Page

4. NINDS: The National Institute of Neurological Disorders and Stroke:
   https:// clinicaltrials.gov/search/term=Muscular%20Dystrophy
   This website gives an update of clinical trials all over the world.

5. Some support organisations:
   i. The Muscular Dystrophy Association (MDA): https://www.mda.org
   ii. Parent Project Muscular dystrophy (PPMD):
   https://www.parentprojectmd.org
   iii. TREAT-NMD: http://www.treat-nmd.eu
   iv. World Duchenne Organisation (UPPMD): https://worldduchenne.org

The Paediatric Muscle and Nerve clinic

Bangalore Baptist Hospital

A service provided in collaboration with the ORDI: Organization for Rare Diseases India.

Diseases Treated:

1. Duchenne Muscular Dystrophy
2. Spinal Muscular Atrophy
3. Congenital Myopathies
4. Congenital Neuropathies
5. Congenital and Acquired Myasthenias
6. Limb Girdle Muscular Dystrophy

Specialists Involved:

1. Paediatric Neurologist and Neuromuscular Specialist: Dr. Ann Agnes Mathew & Dr. Rachel
2. Geneticist: Dr. Madhuri
3. Paediatric Endocrinologist: Dr. Suman Rath
4. Paediatric Orthopaedics
5. Paediatric Pulmonology
6. Paediatric Cardiology
7. Ophthalmology- For cataract surveillance
8. Paediatric and Neuromuscular Physiotherapy services: Ms. Ranjeetha & Ms. Sangeetha
9. Orthotists
10. Dietician: Ms. Daphne Balan
11. Patient Liaison personnel: From ORDI
12. Paediatric Surgery: Dr. Naveen and Dr. Anthony Robert
13. Mano Chethana (Liaison Child Psychiatry): Dr. Sowmyashree

For emergencies contact: 080 22024700,

For appointments contact:
8892555000 (ORDI helpline), Baptist OPD: 080 22024322

Clinic Timings: Thursdays of every week from 02:00 to 04:00 pm

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Bangalore Baptist Hospital
Bellary Road, Hebbal
Bangalore 560024
Phone : +91- 80- 22024700, 23437971
Mobile : +91- 9448496602
Fax : +91-80-23437970/ 23337817
E-mail : talk2us@bbh.org.in

Initiative supported by :

ORDI
Organization for Rare Diseases India

Organization for Rare Diseases India
#16/2, Sri Krishna Krupa, 19th Cross,
8th Main, kashi mutt Road, Malleshwaram,
Bengaluru – 560055, Karnataka, India.
Contact No : +91 9980133300
Email : contactus@ordindia.org
Website : www.ordindia.org
Rare Disease Helpline no : +91 8892 555 000