Standards of care for children with Duchenne Muscular Dystrophy in India

Brought out by

Muscular Dystrophy Association India

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Reviving Spirits

Muscular Dystrophy Association India

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First Edition: August 2009

Second Edition: Jan 2014

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Foreword

The book is in simple language and easily understandable and will be very useful to parents and the people caring for these children.

The simple clues on day to day care will be of great use to everyone.

This book will be a ready referral book and wish that it is made available free of cost to every affected family.

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Muscular Dystrophy Association India

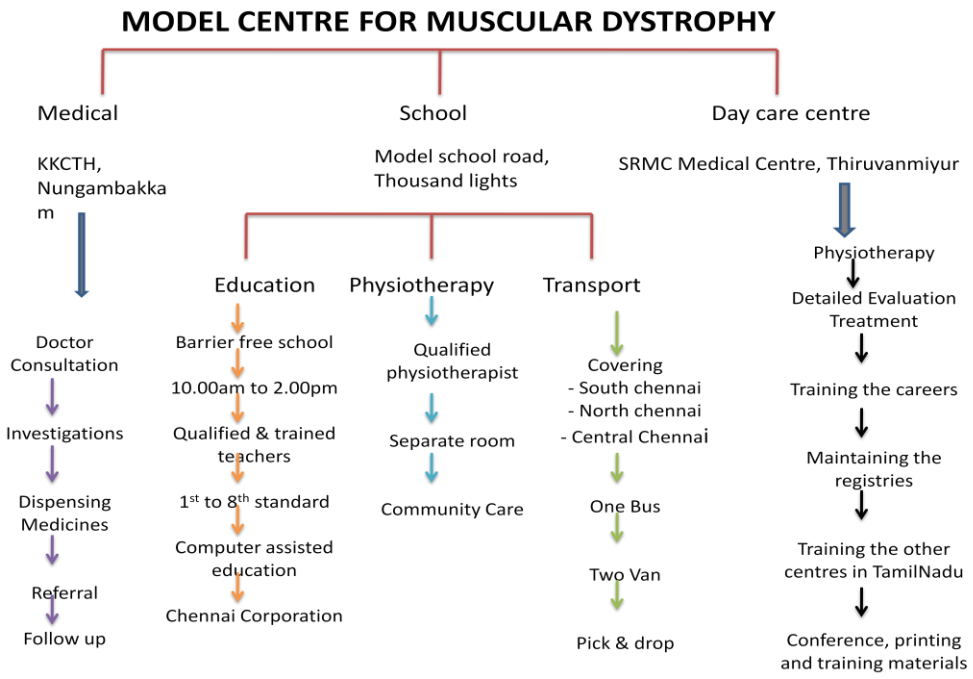
Our **NGO** was started in the year **2000**. It is running successfully for more than a decade with help and support from a huge number of philanthropists to make the lives of children with Muscular Dystrophy better.

We try our best to give these children as much comfort and care. **We assist** them in getting a **clear diagnosis, rehabilitation and care** for these children.

- We provide them appropriate equipments necessary –**splints, spinal braces, wheel chairs** – specifically designed and made for each of these children**, cough assist devices, incentive Spirometers** etc.
- We also **keep the families updated** with all the efforts that are ongoing in the world in trying to find a **cure for these genetic disorders** and take care of their medical needs with periodic evaluations.
- We are also **involved in international multi-center research in to muscular dystrophy** – It is the only center in India at present to be part of the Cooperative International Neuromuscular Research Group (CINRG) and the Treat NMD Group (European group) – and have been part of the group for many years now.
- Propagation of information on the disease
- To raise awareness about the disease
- Early Identification of Disability and Early Intervention.
- Help access at schools, place of employment, residence and public area for wheelchair bound children
- To advice the family on role of physiotherapy and provision of the same.
- Counseling sessions for the parents and close relatives Prevention of Disability.
- Rehabilitation services including counseling, strengthening capacities of persons with disabilities and their families, Physiotherapy, Occupational therapy, surgical correction and intervention.
- Distribution of supportive aids and appliances.

Model Centre for Muscular Dystrophy

Muscular Dystrophy Association India will be the model centre functioning from Chennai. Through the state planning commission direction Department of Differently Abled is guiding us in implementing the services for people afflicted with Muscle disease.



Role of the centre

Diagnosis
Investigation
Classification of Different Muscle disease
Clinical & Physical evaluation
Management-Medical & Rehabilitative
Dispensing of Medicines
Provision of Orthotics & Wheelchairs
Follow-up
Guiding in their Education
Possibility of having National registry for Muscle disease

Services Provided

Periodic evaluation

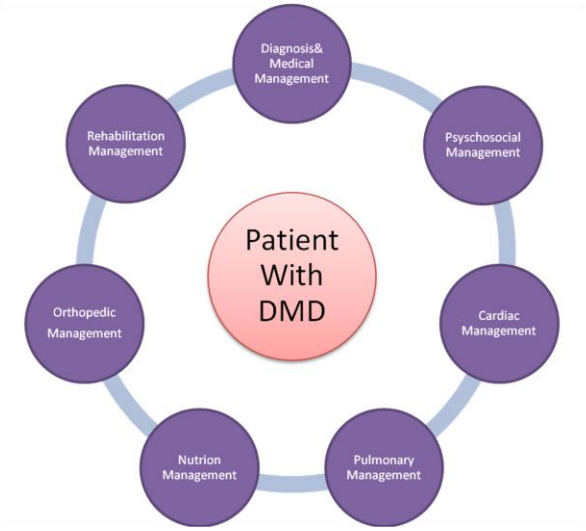
- Evaluation includes
  - Doctor evaluation
  - Physiotherapist evaluation
  - ECG
  - ECHO
  - Pulmonary function tests
  - Xray
  - Blood tests

- Administering Vaccines during the winter season.
- Transport facility to pickup and drop the child from home to the school and back to home.
- Provision of Physiotherapy in the centre as well as in the home.
- Training programme for the other day care centres.
- Conducting camps.
- Holding conference involving national & international people once in two years.

Muscle Disease team

Medical Team	**Allied Health Professionals**
Pediatrician	**Physiotherapist**
Neurologist	**Staff Nurse**
Orthopaedician	**Orthotist**
Pulmonologist	**Dietician**
Cardiologist	**Psychologist**
Medical Genetist	

APPROACH TO MANAGEMENT



Physiotherapy forms sheet anchor in the management of Children with Muscle Disease. It helps in

- Slow Disease Progression
- Control Secondary Conditions
- Improve Quality of Life of the Patients and Families with the disease.

Physiotherapist role in the day care centre

- Collection of patient contact information.
- Quality of Life Assessment
- This therapist will be in the day care centre between 09 and 02 pm.
- Detailed evaluation will be done
- The evaluation includes
    - Manual Muscle Test
    - Quantitative Muscle Test
    - Pulmonary Function Test
    - Timed Function Tests
    - Goniometry
    - Anthropometrics
    - Functional Grading Tests
    - 6 min Walk Test
    - EK Scale
    - North Star Ambulatory Scale
    - Assessing the needs of wheelchair and orthotics

- Maintaining the database of the patients
- Coordinating between the doctors and other day-care centres in helping to form registry
- Registry will help us to formulate standard of care for our population
- Also awareness to the community regarding the carrier Analysis and prevention

Educational / training to the differently abled persons:

The characteristic feature of this disease is loss of ambulation by the age nine and difficulty in climbing stairs by seven. Most children with disability are not in a position to join inclusive education system due to the nature and degree of disabilities.

As majority of the schools are not accepting these children, We started a special school for these children with the help of Chennai Corporation so that they can continue to get their formal education. The special features of the school are

- Barrier free classrooms, toilet, doorway & corridor
- Free education for all
- Computer assisted education
- Syllabus framed by eminent educationalists
- Special bus to bring the children from home for selected areas – with wheel chair hoists
- Regular /free Physiotherapy services at school
- Qualified teachers with the knowledge about the Muscular Dystrophy

Education options learning aids and tools, mobility assistance, support services, etc. are available for children with disabilities. Aids and appliances that can promote their physical, social and psychological rehabilitation by reducing the effects of disabilities are made available to them.

School for MD Children

First of its kind in India. The school has five large class rooms made uniquely child friendly with paintings of cartoon characters on the walls. The school is barrier free with free access on wheel chairs from the gate to the class rooms.

There is enough space for a bus to come in to the school campus and drop the children and pick up the children from the school. The school has very carefully chosen "Matt finish" flooring tiles to avoid skidding for these children as they are very unsteady on their feet and can fall easily.

The bathrooms have been made wheel chair friendly with wide spaces on the corridors as well. The wash basins have been placed at such a level that wheel chairs can go underneath that and the child can himself wash his hands. There are stainless steel hand railings provided all along the sides on the corridor and the bathrooms so that the children can go to the toilets themselves easily. There are three bathrooms provided with fitting and railings so the child can easily get up from the toilet seats by holding on to the bars without looking for any support from anyone else.

For children in the higher classes like the 9th, 10th, 11th and 12th it is proposed to integrate them with the other regular school here so and bring the class rooms to the ground floor and provide ramps. This way they will also feel that they are like any other normal child although they are wheel chair bound.

Role of Physiotherapists

- Attending to the Physiotherapy needs of the children at school
- Community care.
- Making home visits as needed
- Training the carer / attendants
- Liasioning between the doctor and patients

Standards of care for children with Duchenne Muscular Dystrophy

There are various guidelines published from different parts of the world on setting standards of care for children with Duchenne Muscular Dystrophy (DMD). We have tried to evolve some guidelines based on considered experts opinion to enable families to decide on the how best to take care of the children with DMD.

A broad overview of the various aspects of the care of these children including diagnosis, neurology, diet - nutrition, respiratory Care, cardiac Care, orthopedics, psychosocial, physiotherapy and rehabilitation have been presented here.

The primary goal of the recommendation is to help families deal with their children based on the experiences of the experts opinion in India as some times many of the International guidelines may not be relevant for a child who lives in India. This is only a beginning and we hope in future to evolve broad consensus guidelines from all the experts working in this field in India.

Clinical examination:

The common presenting symptoms for these children are toe walking around 2 years of age, h/o frequent falls, difficulty in climbing stairs, difficulty with getting from squatting position on the floor, big calves and an abnormal walking pattern. Although the age of presentation appears to vary – most of these symptoms are obvious by about 4 to 5 years of age. The clinical examination of the children should include observation of the gait pattern, difficulty with climbing stairs and “climbing up with support on the knees” – gowers sign on getting up from the floor. Marked weakness involving the proximal muscles of the arms and legs may be seen. In some children winging of the scapula when asked to push against the wall may be visible clearly. Many of these children appear to have “big tongue”. Neck flexor weakness is also common.

Investigation:

Serum creatine phospho kinase (CPK): Massive elevation of the serum CPK (at least 10–20 x normal) is present in most cases. The finding of a high CPK level should prompt urgent specialist referral for confirmation of the diagnosis. The clinician should be made aware of the association of non-hepatic elevation of SGOT and SGPT in DMD. Unexpected elevation of transaminases should alert the physician to the possibility of DMD and so should check the CPK.

Genetic testing : This is the best first step available for the confirmation of the diagnosis of DMD and should be performed. Multiplex PCR testing for the full 79 exons of the DMD gene is available at several laboratories in India at a very nominal cost and this can be performed on a small sample of blood drawn from the child. A deletion of the dystrophin gene will be found in around 70% of cases. This will then confirm the diagnosis and no further testing may be necessary. At present in india we do not have laboratories capable of looking for a duplication or a point mutation on a routine basis. If the deletions are negative it would be important to go for the next step of muscle biopsy.

Muscle biopsy: This is usually performed on a sample of muscle taken from the child under anaesthesia. Most hospitals still use the quadriceps/ biceps muscle for sampling. Should be performed by an expert and hospital who are capable of handling any undue emergencies during anaesthesia in these children.

Although needle biopsy can be considered for getting the muscle tissue sine the yield of muscle tissue is so small and many times inadequate unless performed by an expert an open biopsy method is preferable. The biopsy specimen should be handed over quickly to a pathology laboratory which is capable of performing routine histopathology, enzyme histochemistry and immuno-histochemistry as these steps are important to quantify the amount of dystrophin present in the muscle tissue sample provided and help a definitive diagnosis.

Carrier analysis: of the mother by molecular genetic testing is an important next step along with genetic counseling. Unfortunately this is again not freely available and some times rather expensive. However it is an important step as it helps to counsel the family about the chance of recurrence during the next pregnancy in the family. Even if the condition has arisen as a result of a new mutation, there is an average 10% risk of recurrence due to germline mosaicism. Genetic counselling should also be offered to sisters and aunts (mother's side) in reproductive age if the mother carries the mutation.

Neurology

The role of neurologist in India is not only to make a definitive diagnosis and counsel the family but also to help the family through appropriate guidance regarding the various aspects of the problems faced by the child and family with DMD. This can of course be done by a physician who has reasonable knowledge of the availability of various services in the local area.

The use of corticosteroids in DMD:

Timing: Many centres in the world use steroids typically around the age of 4-6 as they feel that the benefits of this may be better. Less functional gain may be seen if initiation of steroids is delayed until close to the loss of ambulation. In our personal practice we find families whose children are around 4 to 6 years of age are reluctant to give steroids to their children in India due to side effects but are more favourable if prescribed the same around 7 to 9 years. The main concern for the families is the immuno-suppression and exposure of the child to infectious diseases like chicken pox and tuberculosis.

Regimes: the most common daily dosage regimes are 0.5 to 0.75 mg/kg/day prednisolone . Some centers use Deflazocort at a dose of 0.8mg /kg/day but it is a lot more expensive than prednisolone. They are likely to be equally effective, but have slightly different side-effect profiles. Deflazacort may produce less weight gain but has a higher risk of asymptomatic cataracts. The response to steroids and the side effects like weight gain appear to be variable in different children and some gain a lot of weight and others tolerate higher doses very well without any side effects. So the exact dosage needs to be titrated depending on the child's response and side effects seen.

Monitoring: of the child periodically for improvement in the clinical symptoms like time for getting up from the floor, ability to climb stairs, not falling as frequently as before, muscle strength testing, Forced Vital Capacity evaluation by lung function testing and parent and child perception of the value of the treatment are all important.

Side effects: Major side effects of steroids to consider are behavioural changes, failure to gain height, excessive weight gain, osteoporosis, impaired glucose tolerance, immune/adrenal suppression, dyspepsia/peptic ulceration, cataract, and skin changes. It is for this reason important to monitor weight, height, blood pressure, urinary dipstix (glucose), cushingoid features, mood/behaviour/ personality /GI skin changes, red reflex of eyes, bone fractures, and recurrent infections. Many of the side effects can be dealt with easily if closely monitored.

Tapering: If corticosteroids need to be stopped, they should be tapered/stopped slowly over weeks and not suddenly. Suggested tapering of drug dosage is to take ½ the regular corticosteroid dose the first week, ¼ the dose during the second week, ⅛ the dose during the third week and thereafter stop corticosteroid medication.

How long to continue steroid treatment: Most families in India quite reluctant to continue steroids beyond the stage when the child becomes non ambulant. There is some evidence that steroids do improve hand function and lung function in wheel chair bound children and so is routinely

prescribed / continued in children who become non-ambulant. However, some patients may notice excess weight gain and osteopenia and so needs to be discussed with the families before it is prescribed to non ambulant children.

Role of a Pulmonologist in neuromuscular illness

For us to be effective and successful in implementing disease modifying or preventive strategies in any disease we have to know the basics and principles for these interventions. Here are a few know how of the basics and problems of respiratory aspects in patients with neuromuscular disease. They are;

What is breathing and why do we breathe?

Our body is made up of tiny cells, each of which has a special job to perform for our existence. These cells need a special gas called oxygen that allows the cell to get energy from the food we eat. Oxygen in the air is brought into our lungs by an act of breathing called inspiration i.e. bringing in air from environment in to the lungs. The heart pumps blood and takes oxygen from the lungs to the cells.

Another gas, called carbon dioxide is a waste product of our cells when they produce energy required from our food with help of oxygen. It is very dangerous if it builds up in your body. The heart beat keeps our blood moving in our body and carries this carbon dioxide from our cells to the lungs. The lungs, then breathes out all the bad gas by an act of breathing called expiration i.e. bringing out bad air from the lungs in to the environment.

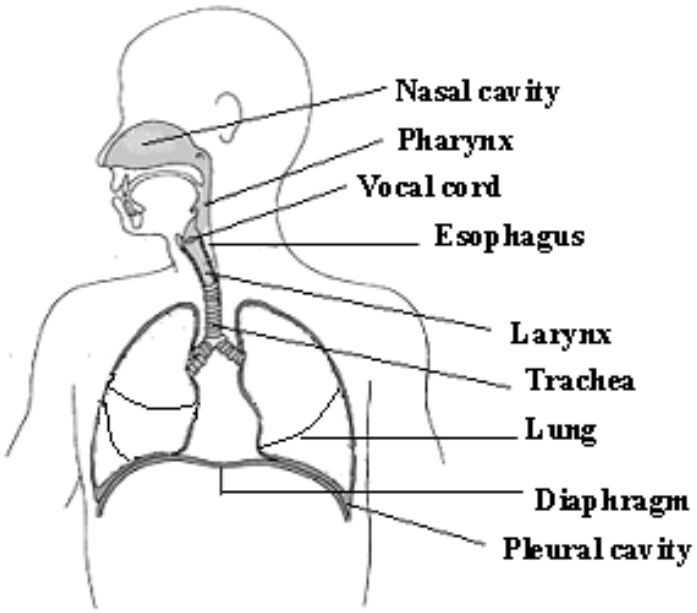
This act of both inspiration and expiration combined is called breathing. It is totally automatic and is controlled by our brain. The rate i.e. number of breathing per minute, varies with age. It is faster when young and reduces with age. The rate also increases on demand e.g. in fever or exercise, our cells are working harder, and they need more oxygen. They also produce more carbon dioxide. That's why we breathe faster and our heart beats more quickly.

In general our heart and lungs work together to make sure every cell in our body gets enough oxygen. The heart beat and breathing are the most integral part of life. For mere existence our body has to have the heart beating and the lungs breathing. If any of the both stops we are virtually dead.

What are the components of our respiratory system?

The respiratory tract (Fig 1) extends from the mouth and nose, through throat (pharynx) then airways or air tubes (larynx, trachea and bronchi) to the alveoli i.e sponge like areas in the end of the lung where oxygen and carbondioxide exchange takes place. The respiratory system also involves the paranasal sinuses (air pockets in the face along side of the nose and upper air passage), pleura (covering of the lung), ribcage (chest bones) and the muscles of respiration. The main function of the respiratory system is gas exchange and maintains adequate oxygenation to tissues i.e. cells.

Fig 1: Respiratory system



How do we breathe?

In normal breathing inspiration is an active process and expiration is a passive process. Diaphragm i.e a big thick sheet of muscle between the chest and abdomen is the principal muscle of inspiration. An active contraction of diaphragm elevates the rib cage i.e. chest bones and enlarges the chest and this sucks in air from environment in to the lungs. The other muscles of inspiration are; the *External Intercostal Muscles* i.e. outer muscles between the chest bone and the *Accessory Muscles* i.e. neck muscles and upper muscles of the back e.g. Scaleni, sternomastoid, trapezius and back muscles. These muscles help to raise the chest wall and are active only during increased work of breathing and are normally not active in normal regular i.e. tidal breathing.

In normal tidal breathing the expiration occurs by elastic recoil of the lung and chest wall. This is like a rubber band which on stretching when released gets back to its original position. However there are muscles of Expiration are; A*bdominal Muscles* and *Internal Intercostal Muscles* i.e. inner muscles between the chest bone. These muscles of expiration do not work regularly but work only during coughing, straining, vomiting and with airways obstruction.

Why are the lungs always at risk for infection and insult?

The lung is at a remarkably risk for infection and insult because of airways i.e passage that brings air from environment, to the lungs being in direct connection between the lung and the atmosphere which is not always clean or sterile. Every day an average adult inhales approximately 10,000 litres of air. The alveolar membrane covers a surface area of over 140 m^2 which is approximately three fourth the

size of a tennis court and such large area is exposed to external environment with each breath. Lungs in addition to its chief role of gas exchange have to have a strong line of defence to counter threat from the environment.

How do we protect our respiratory systems?

The respiratory tract and its mucosal surfaces i.e. layer covering the air passages has good and efficient defence mechanism that repulse the regular threat of infection and injury to our respiratory system. They are

N*asal vibrissa* or nasal hairs that acts as filter of large particles.

The *Epiglottis* i.e the check gate at the junction off air pipe and food pipe in the throat, this protects the food particles from entering the wrong way in to the respiratory system.

Further the *branching design of our airways* from bigger to smaller unit's results in the quick and effective filtration of the inhaled particles from entering the terminal alveoli.

Coughing and sneezing are protective mechanism where the mucus deposited with organism or the irritant substance is ejected outside the respiratory tract.

Certain irritant can cause bronchoconstriction i.e. shrinking of the lower air tubes as a protection reflex of the respiratory system.

Mucociliary escalator: a mucus carpet of the respiratory passage from the anterior third of the nose till the bronchiole. They contain ciliated cells i.e small actively cells with continuous beating movement of about 1000-1500 times a minute that pushes the irritants and organism from inside towards the mouth. These secretions are finally swallowed or expectorated (spitted out).

The lungs also contain various specialized antimicrobial secretions, to guard it effectively.

Problems in neuromuscular patients

Poor swallowing mechanism and cough reflex: Normally our mouth too contains disease causing germs that are over powered by our body defence and they stay in our body without causing disease i.e. commensals. However if they over power our body defence or if the body defence become weak they can cause disease i.e. pathogen. In neuromuscular disorders the muscles of swallowing can be weak and causes pooling of saliva. This makes the commensals to become pathogens, causing disease. In addition the cough reflex can also be weak, hence we are prone for aspiration i.e. what is to enter the food passage gets in to the air passage causing infections. In normal individuals food or our own saliva cannot enter our air passage as we are protected by normal swallowing mechanism and good cough reflex. Whereas in person with neuromuscular problems these protections are compromised resulting in infections. Frequent infections of the lungs destroy the normal air filled spongy looking lungs and convert them in to pus filled stiff lungs i.e. suppurative lung disease.

Shoulder weakness is the earliest sign towards the onset of respiratory muscle weakness. It is usually the expiratory muscles that are first affected followed by the inspiratory muscles. Even the surrounding muscles of neck and back are affected resulting in drooping or sagging of shoulders. This results in the patients being unable to expand their chest to maximum limit and also causes poor clearance of mucus, which is normally expected to be cleared of the airways. All these together contribute towards collapse infection and poor lung function.

In kyphoscoliosis (kyphosis is bending forward of spine bone, scoliosis is bending sideway of spine bone) the lung function is restricted by decreased chest wall expansion and can result in unequal lung expansion resulting in basal atelectasis (collapse of the lung) on the concave side and over expansion of the convex side, leading to defect in oxygen delivery to the cells. This causes increased work of breathing as compensation and may predispose to respiratory failure.

Obesity leading on to increased adipose tissue in muscles resulting in poor muscle function. The muscles appear big but are not strong and at times not even capable of doing the normal functions expected of it.

In normal individuals, physical exercise increases mucus (protective secretions in respiratory tract) elimination by as much as 40% compared to normal breathing and is an important component of normal airway or air tube clearance. Whereas in Non ambulant and/ or Bed ridden patient's they are unable to do physical exercise this results in retained secretions that becomes very thick leading on to collapse of the lung. This poor clearance of secretion affects normal mucus function of the pulmonary defence system. Excessive or retained secretions undergo qualitative changes to become thick, sticky and infectious, eventually injuring the healthy lung tissue and deranging oxygen- carbondioxide exchange.

Any person with impaired respiration is more worse during sleep as our voluntary compensatory mechanism which helps us to compensate to some extent during day or when awake are unavailable to us as we sleep. This in conjunction with the significant collapse of upper air passage i.e. laxity of back of nose and throat muscles, (some amount of minimal insignificant laxity happens in all individuals) results in obstructive sleep apnea syndrome (OSAS). OSAS is loss of oxygen to our brain and body during sleep because of obstruction of our upper air passage during sleep. OSAS may lead on to disturbed sleep, irritability, loss of concentration, excessive sleepiness during day, hypertension (Increased Blood pressure) and pulmonary (Lung) hypertension

Impaired oxygen delivery to the cells and poor carbondioxide removal from the cells calls for increased effort of respiration. This increased effort in a child or person with poor respiratory muscle strength leads on to respiratory failure i.e respiration not able to meet the body's needs and demands. Respiratory failure causes breathing difficulty and may lead on to death if not intervened appropriately.

Lungs and heart are like 2 rails of a railway tract. They coexist and any problem in one is reflected in the other. When there is more demand on the lungs the same is reflected in the heart. Long standing increased demand to perform on the lung can result in pulmonary (lung) hypertension and this causes more pressure on the right side of the heart causing the right heart to become bigger and bigger.

This later, at one point of time causes the heart failure i.e. Corpulmonale. In addition children with dystrophy are prone for cardiomyopathy (weakened heart muscle) where the heart may look big like the other muscles of the body but is not effective to carry out its function resulting in heart failure.

Common causes of death in neuromuscular problems?

The major and common causes of death are

Aspiration

Pneumonia

Respiratory failure

Heart failure

Strategies to counter these problems when & how?

General strategies:

Good oral and dental hygiene, to prevent colonization of pathogenic organisms and to maintain a normal oral flora.

Encourage proper positioning. Upright posture while sitting to prevent kyphoscoloasis. Propped up positioning during sleep like placing a average sized pillow while sleeping to prevent aspiration (said to help to some extent)

Breathing exercise of upper airways, like pranaya or deep and forceful breathing through nose to facilitate nasal breathing and to clear them of retained secretions.

Regular vaccinations against vaccine preventable respiratory illnesses like, BCG, DPT, Measles, Hemophilus influenza B (HIB), Varicella, Pneumococcal diseases and yearly influenza vaccine.

Treat comorbid (other diseases that may coexist) conditions if any. More specifically asthma, allergic rhinitis and GERD (gastro esophageal reflux disease) as these target the lungs and airways.

Age-appropriate dietary therapy to help them avoid obesity. Obesity is especially harmful because it places additional strain on their already weak muscles. Unfortunately, most patients are at high risk of obesity because of their limited physical activity. To chew well and swallow as it might decrease the risk of GERD and aspiration. Avoid lying down immediately after a meal as it might increase risk of aspiration.

Early aggressive treatment of respiratory illnesses is advisable as chances of having prolonged or severe infections are high in view of poor airway clearance, risk of aspiration and poor cough reflex.

Normal ambulant child:

In addition to general strategies, encourage normal activity as much as possible let them not over exert or under exert.

Non-ambulant child:

a) Proper positioning with straps, pillow and braces to prevent kyphoscoliosis as it might further impair respiration. As normal upright posture is essential for us to breath to our maximum capacity.

b) Physiotherapy of shoulders and back to prevent muscle contractures and to facilitate chest wall movement.

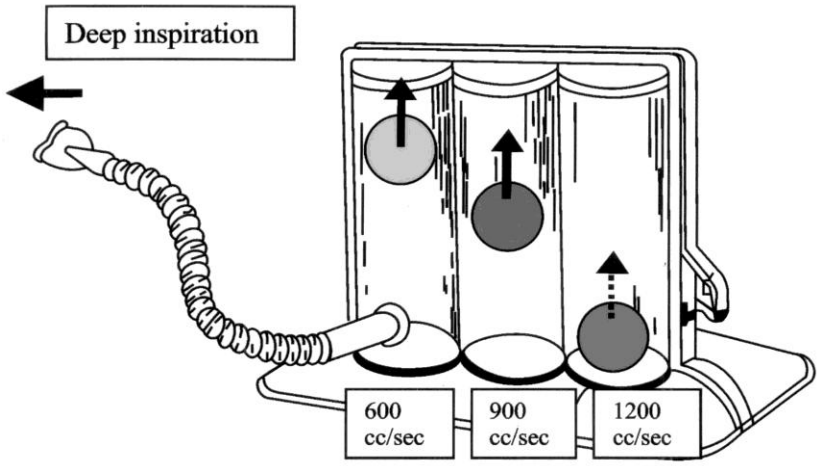
c) ***Breathing exercises*** have to be initiated in non ambulant patients and also in ambulant patients with weak shoulder muscles. Breathing exercises are meant to increase the breathing efficiency. It increases the diameter of airway above that to what happen in normal breathing thus helping to loosen and dislodge airway secretion. This prevents alveolar collapse, facilitates lung expansion and peripheral airway clearance. Breathing exercise encourages to make a larger than normal inspiratory effort to mimic natural sighing or yawning. In general, normal breathing without sigh results in alveolar collapse within one hour. *Incentive Devices* use the principle of sustained maximal inspiration (SMI) and have a positive reinforcement on the patients by helping them to visualise their progress through visual indicators. Though several such devices are available, the incentive deep breathing exerciser *(Fig 2)* is most suitable for pediatric age group. The patient is asked to sustain his inspiratory effort for a minimum of 3 seconds atleast or to the maximum possible.

d) All non ambulant child despite having good cough and normal breathing mechanics should have daily chest physiotherapy atleast 2-3 /day for 5- 10minutes and about 3-4 times/ day when they are not well more so with respiratory infections. As in the absence of normal physical activity the airway clearance of mucus is definitely impaired in the dependent regions (lower regions of the lung) and is at risk of retention, collapse and infection.

e) Cough stimulation: In all children after chest physiotherapy and breathing exercise the child is requested to cough as the mucus cleared or collected has to be either swallowed or expectorated. Cough enhances mucus clearance of upper and larger airways. In child with feeble or ineffective cough, cough reinforcement with cough exsufflator is of much help.

f) The first sign of respiratory failure in children is usually sleep disturbance or difficulty in sleeping during sleep, non invasive ventilation (BiPAP) with or with out oxygen with pressures being titrated to patients needs. With progressive detoriation they might require BiPAP even through the day.

Fig. 2: Incentive deep breathing exerciser



Cardiac evaluation and follow up of Duchenne Muscular Dystrophy children-guidelines

Duchenne muscular dystrophy, primarily a neuromuscular disorder involving the skeletal muscles, has a strong predilection to affect the myocardium leading on to features of dilated cardiomyopathy and rarely cardiac dysarrthythmias.

The major source of morbidity is the progressive motor deficits and contractures requiring orthopedic intervention in the form of leg braces and tendon release surgeries. With the advent of portable ventilatory assist devices,the life expectancy has increased drastically from 15 years in the early 1960s to 30-40 years of age.

Despite advances in the supportive care and life style modification, the mortality in children below 20 years of age is predominantly due to cardiac complications like progressive Left ventricular dysfunction and rarely cardiac dysarrthythmias. These cardiac morbidity are treatable but not completely reversible. In children below 14 years of age, the incidence of significant cardiac abnormality is around 15%. Children with LV dysfunction are usually asymptomatic as the neurological deficits limit their level of physical activity.

Hence, any undue physical activity precipitates subclinical cardiac involvement. DMD children often develop evidence of early myocardial dysfunction in ECHO prior to the onset of cardiac complains.

Cardiac evaluation of a child with Duchenne Muscular Dystrophy:

Investigations:

1. X ray-chest (PA view)

2.ECG to detect rhythm and conduction abnormality.

3.ECHO is the most useful tool to detect cardiac involvement in DMD (chamber dimensions, LV systolic and diastolic function, RV function and PA pressures).

4.Cardiac MRI if ECHO findings are not satisfactory due to poor echo window.

Protocol for Cardiac Evaluation:

1. A complete cardiac evaluation in the form of clinical examination, chest X ray, ECG, ECHO when DMD is first diagnosed.

2. Cardiac evaluation every 2 years till 10 years of age followed by annual check ups.

3. Detailed pre operative cardiac evaluation when any surgical intervention is planned.

4. Cardiac re evaluation when the child develops cardiac complaints like dysnea, PND, orthopnea, palpitation, dizziness or syncope.

5. Siblings of children with DMD should be screened.

6. In carriers if the initial work up is negative, regular follow up every five years (assessed yearly if cardiac involvement is detected in ECHO).

7. Holter evaluation if child complains of palpitations, giddiness or syncope with or without evidence of dysarrhythmia in resting ECG.

Management

1. ACE Inhibitors are started when there is echo evidence of LV dysfunction even when the child is asymptomatic.

2. Beta blockers need to be added along with ACEI in children with Chronic Heart Failure.

3. Diuretics can be added in children with evidence of fluid retention. [pedal oedema or sacral oedema in bedridden children] or with basal crepitations.

4. Low dose aspirin (3-5mg/Kg) in children with Dilated cardiomyopathy. Oral anticoagulants in children with severe LV dysfunction to prevent embolism.

PHYSIOTHERAPY MANAGEMENT FOR DUCHENNE MUSCULAR DYSTROPHY

INTRODUCTION

This book is intended to support the practice of physiotherapy at home for children and young people with Duchenne muscular dystrophy.

WHAT IS PHYSIOTHERAPY?

Physiotherapy is the physical treatment and management of a disease or condition which enables people to reach their maximum physical potential. Physiotherapists help to ensure that their patients lead as fulfilling a life as possible by advising children, families, carers and school staff about how the condition affects physical development.

WHY NEED PHYSIOTHERAPY?

Physiotherapy input is essential for the maintenance of function in DMD.

PHYSIOTHERAPY BENEFITS

- Muscle exercises help because being inactive can make the disease worse.
- Physiotherapy can help maintain muscle strength and flexibility.
- Physical aids such as braces or wheelchairs can help give more mobility.
- These measures aim to give people with MD a better quality of life.

WHAT WILL PHYSIOTHERAPIST HELP IN?

- Maintain good range of movement and symmetry at different joints.
- Maintain the best possible function.
- Prevent the development of fixed deformities
- Prevent pressure problems with the skin.
- Aid in standing and walking
- Monitor respiratory function and advice on techniques to assist with breathing exercises and methods of clearing secretions.

PHYSIOTHERAPY AT HOME

The exercises recommended by your physiotherapist need to be done regularly if they are to be effective. This means developing a routine at home.

Any physiotherapy regime should be based on:

- ❑ The needs of your child
- ❑ The advice of your physiotherapist
- ❑ The needs of the family (a practical routine to suit your family's lifestyle)

Although the exercises should never be painful, stretching exercises may cause the muscle to feel different and your child will need to become accustomed to this. Some children will be able to do self-stretching exercises, as well as exercises with a carer, and a physiotherapist can help with this.

There has been some concern that over activity, such as exercising with weight, may cause more harm than good to a child or young person with muscular dystrophy. Exercise should be at a moderate level and not cause extreme or severe fatigue. Any activity that a child does voluntarily and without becoming over tired, will have a positive effect.

CHANGES IN MD

MUSCLES

Muscles allow us to move, stand and perform that range of movements needed for daily living. Each muscle is made up of fibres, although the type and amount of fibre varies depending on the sort of work the muscle does. The muscle fibres we use for standing are, for example, different to those in the muscles used to make fine, quick finger movements.

Muscles are attached to bones via specially adapted parts of the muscle, called tendons. A muscle spans at least on joint and a movement occurs when it contracts or shortens. Muscles and their tendons are normally very flexible, allowing movement through lengthening and shortening. Usually when one muscle contracts or shortens the opposite muscle lengthens.

In Duchenne muscular dystrophy, muscle fibres break down and are replaced by fibrous and / or fatty tissue causing the muscle to gradually weaken. The rate at which this happens can vary between children with the same condition. Some muscles will be affected earlier than others and if one muscle weakens sooner than another, it can upset the normal balance of strength and cause contractures.

BONES

When bones are not subjected to the normal stresses of everyday living (such as occurs during walking, running and jumping), they lose calcium and become soft or brittle. Soft bones fracture more easily than normal bones. This is not often a problem until well into the wheelchair phase, when accidents such as falling from the chair or during transfers to or from the chair may cause a fracture. Sometimes it is not very obvious that a fracture has occurred, until an X-ray is taken because of persisting pain or increasing swelling.

If most of the weight of the body is put onto one buttock for long periods while sitting in a wheelchair there may be discomfort in that area.

CONTRACTURES

When muscles are not used or become weak, they lose their stretchiness along with the associated tendons and ligaments (the tissue around the joints which connects bones, and controls the extent or range of movement). The joint becomes stiff and tight, usually more in one direction than the other. When a joint becomes fixed in one position, this is known as a contracture, and a deformity may

occur. If not treated, these will become severe, causing discomfort and restricting mobility and flexibility.

These are caused by the walking position which the child adopts – on the toes with feet apart to maintain balance as the hip, knee and trunk muscles weaken. Children in the later stages of the condition spend more time sitting down which increases the tendency to develop hip, knee and ankle contractures.

The postures, which they habitually assume to improve their function, will inevitably lead to more tightness in some muscles and the eventual development of contractures.

Contractures can affect the knees, hips, feet, elbows, wrists and fingers.

As soon as possible after diagnosis, seek advice about physiotherapy and start treatment and management aimed at preventing contractures. These can be minimized significantly by keeping the body as flexible, upright and mobile as possible.

Ensuring that his joints are moved regularly through their full range, with gentle pressure at the end of the range, minimizes the risk of contractures. The pattern of weakness and tightness is very predictable in DMD, and your therapist will provide an appropriate stretching program to be performed either at home or school. Parents' commitment to a regular daily stretching program will bring their son the best results and maintain his flexibility. Stretching programs should begin as soon as the DMD diagnosis is made and be continued without fail (within reason) every day at the same time.

ADAPTING TO CHANGES

Early Developmental Stages

The child will enjoy acquiring gross motor skills (movements which use the large muscles of the body) such as crawling, rolling, walking, cycling on adapted trikes and may be running and jumping. All these activities provide good opportunities for learning and development but some will need to be adapted as the child loses strength and tires more easily over time.

During this stage of the child's development is important to encourage activity that does not cause extreme or sever fatigue. Parents or carers may want to consider introducing a physiotherapy and exercise programme that includes:

- Regular stretches – self and / or manual stretches as well as passive
- Swimming, hydrotherapy
- Wearing orthoses (splints) at night to slow down contractures in the ankles.

Later Developmental Stage

There will be a progressive loss of function. Although the child will be walking for periods of time, he or she will also require a wheelchair for mobility, especially over long distances. The upper limbs will also be weaker but daily function activities should be encouraged.

A physiotherapy programme at this stage may include:

- Regular passive stretches
- Stretches to the upper limb muscles
- Swimming
- Wearing orthoses (splints) at night
- Prone lying and other good positioning

Wheelchair Stage

Both good posture and manual stretching programme are important.
A physiotherapy programme at this stage may include:

- Regular stretches to minimise the development of contractures in hips, knees and ankles and ensure comfort in bed, ease in dressing and positioning in wheelchair.
- Stretches for the upper limbs to minimise contractures
- Prone lying and other good positioning
- Swimming
- Use of orthoses.

Physiotherapy support may need to be adjusted following any surgery for scoliosis. In particular, the wheelchair may need adapting to accomodate the child's improved posture.

HOW TO USE THIS BOOKLET

This booklet provides guidelines and instructions on how you can use physiotherapy to help your child with Duchenne muscular dystrophy.

The stretches, exercises and guidance included in this booklet are general, and additional ones may also be recommended.

EXERCISE

It is natural to worry about how much exercise your child should do and whether it's possible to do too much or too little. Finding a balance can be difficult and very often your child is in the best position to say how much is right for him or her. Exercise should never be done to the point of extreme or severe fatigue, although it is unlikely that you could persuade your child to do this.

Research from the British Heart Foundation indicates that all children should exercise at a moderate level for at least one hour a day. A child with Duchenne muscular dystrophy should be able to exercise on a daily basis.

Walking is good exercise and should be encouraged whenever possible, although this can be difficult if your child falls frequently. Children with muscular dystrophy may tire more quickly but can

walk for some distance and time if they are not rushed. They may struggle to walk uphill, downhill, and on uneven surfaces, such as sand or grass. Using a wheelchair for longer distances can alleviate tiredness, enabling your child to enjoy him or himself more on arrival.

It is important to not limit your child's play by being too overprotective. Physical and appropriate sporting activities can be beneficial to a child with Duchenne muscular dystrophy, helping to maintain strength and increase self-confidence.

Swimming is particularly good exercise at all ages; both for the muscles and the lungs, and children may be able to swim or take lessons at school.

Children need to be as active as possible and not spend too much time sitting in front of the television, computer or using electronic games. They could play computer game while standing at a table, or lie on their tummy to help stretch the hips while reading or watching television.

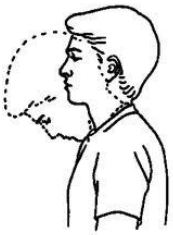
ACTIVE RANGE OF MOTION EXERCISES

Head and Neck exercises:

Starting position:

Sitting: Face forward. Your shoulders should be straight and relaxed.

Chin-to-chest. Gently bow your head and try to touch your chin to your chest. Raise your chin back to the starting position.



Head turns. Turn your head toward the right to look over your right shoulder. Tilt your chin down and try to touch your right shoulder. Do not raise your shoulder to your chin. Face forward again. Next, turn your head to look over your left shoulder. Tilt your chin down and try to touch your left shoulder.

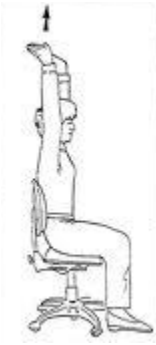
Head tilts. Tilt your head to the side, bringing your right ear toward your right shoulder. Then slowly tilt your head to bring your left ear toward your left shoulder.

Bend your head backward as far as possible so you are looking up toward the ceiling.

Shoulder and Elbow exercises:

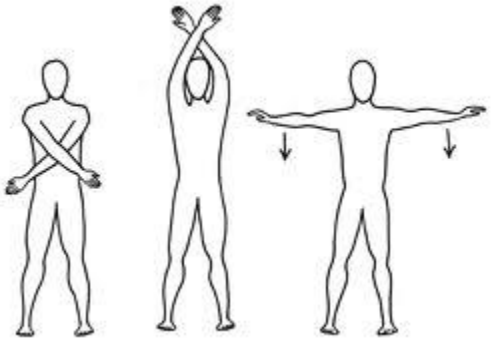
Starting position: Sitting: Hold your arm straight down at your side. Face palms in toward your body. It is best to use a chair without arms if you are in a sitting position.

Shoulder movement, up and down. Raise your right arm forward and upward over your head. Try to raise it so that your inner arm touches your ear. Bring your arm back down to your side.

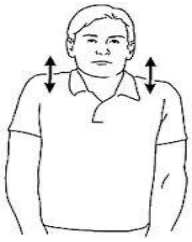


Shoulder movement, back. Bring it back as far as possible behind your body. Return your arm to the starting position.

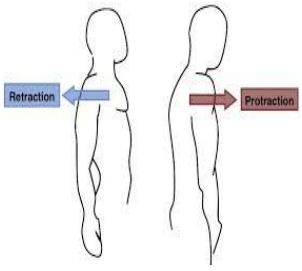
Shoulder movement, side to side. Raise your arm sideways and upward over your head as far as possible. Return your arm to your side.



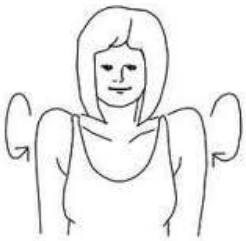
Shoulder Elevation and Depression Raise your shoulders up toward your ears, as if you were trying to shrug. Drop them down again and relax your shoulders.



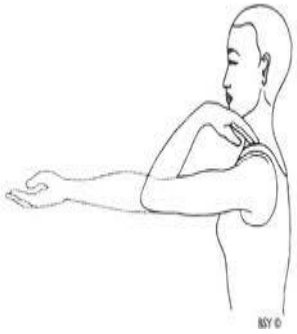
Shoulder Protraction and Retraction Pull your shoulders back, then relax them again.



Shoulder rotation Rotate (move) your shoulders forward, down, back, and then up in one smooth circle. Next, move your shoulders in reverse, moving them up, back, down, forward, and up again in a circle.



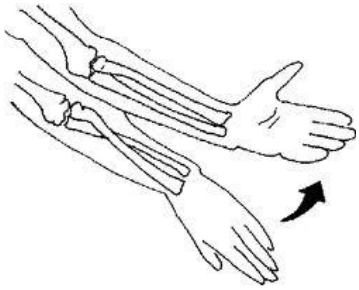
Elbow bends. With your palm facing forward, bend your elbow. Bring your fingertips forward and try to touch your shoulder. Return your arm to the starting position.



Forearm exercises:

Starting position: Your forearm is the part of your arm between your elbow and wrist. Sit down to do this exercise. Bend your elbow and keep it tucked against your side. Rest your right forearm on your right thigh (your lap) or on a flat surface like a table.

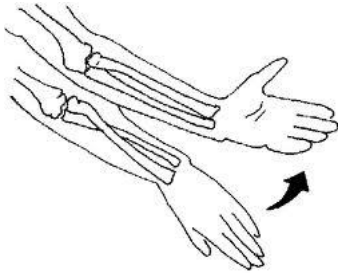
Palm up, palm down. Face your palm down. Rotate (turn) your palm so that it faces up toward the ceiling. Rotate your palm again so it faces down.



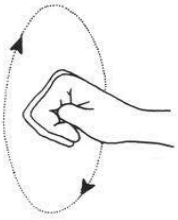
Wrist exercises:

Starting position: Sit down. Bend your right elbow and rest your forearm on a flat surface, like a table. Make sure your wrist hangs loosely over the side of the table.

Wrist bends. Bend your hand back toward your wrist so that your fingers point toward the ceiling. Bend your hand down so that your fingers point toward the floor.



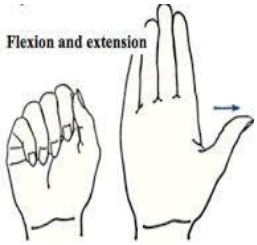
Wrist rotation. Rock your hand back and forth sideways. Gently rotate (turn) your hand in circles.



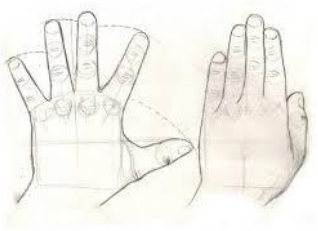
Hand and Finger exercises:

Starting position: Sit or stand. Place your hand out in front of you.

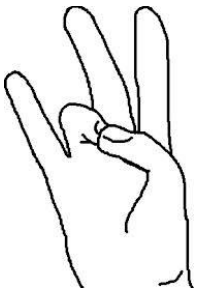
Finger bends. Make a tight fist. Then open and relax your hand.



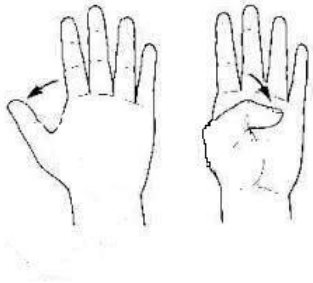
Finger spreads. Open your hand and stretch the fingers as far apart as possible. Bring your fingers together again.



Finger-to-thumb touches. One at a time, touch each fingertip to pad of the thumb.



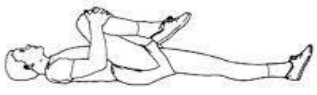
Thumb-to-palm. Move your thumb and rest it across your palm. Move it out to the side again.



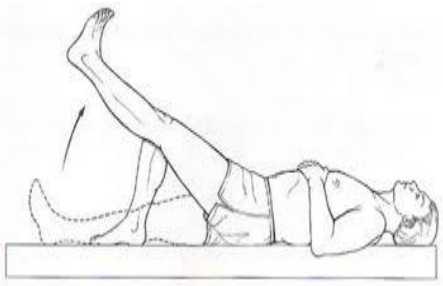
Hip and Knee exercises:

Starting position: Lie flat on the bed with your legs flat and straight.

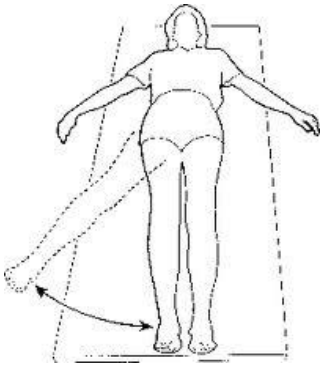
Hip and knee bends. Point your toes. Slowly bend your knee up as close to your chest as possible. Straighten your leg and return it to a flat position on the bed.



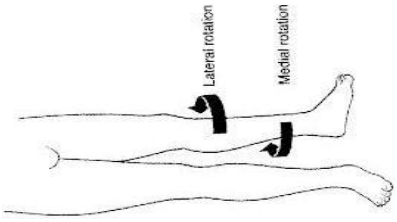
Leg lifts. Raise your leg upward so that your foot is 6 to 12 inches (15 to 31 Centimeters) off the bed. Hold it in the air. Return your leg back to the bed.



Leg movement, side to side. Flex your foot so your toes point up toward the ceiling. Move your leg out to the right as far as possible. Bring your leg back to the middle.



Leg rotation, in and out. With your leg flat on the bed, roll your leg toward the middle so the big toe touches the bed. Roll your leg outward. Try to make your smallest toe touch the bed.

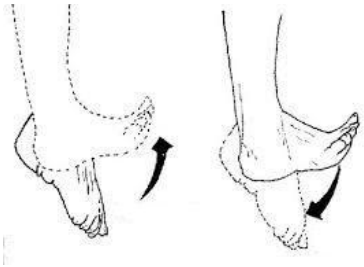


Knee in and out. Bend your knee so the bottom of your right foot is flat on the bed. Return to the starting position.

Ankle and Foot exercises:

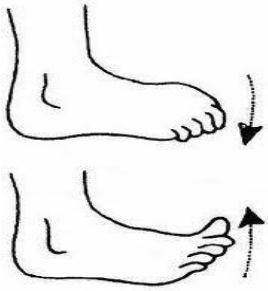
Starting position: Sit in a chair with both feet flat on the floor.

Ankle bends. Keep your toes on the floor and raise your heel as high as you can. Lower your heel and relax. Keep your heel on the floor and try to raise your toes as high as you can.

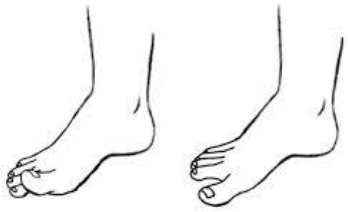


Ankle rotation. Raise your foot slightly off the floor. Rotate (turn) it in circles.

Toe bends. Curl your toes down toward the sole (bottom) of your foot. Straighten them. Curl them up toward the ceiling, and then straighten them again.



Toe spreads. Spread your toes apart. Bring them together again.



BREATHING EXERCISES

Breathing exercises become important as the child becomes less able to actively exercise.

When we breathe in (inspiration), muscles lift the rib cage up and out, making the chest larger. Air then rushes into the lungs to fill the extra space created. When we breathe out (expiration) the muscles relax and the air is pushed out but the elasticity of the lungs. We only use muscles of expiration when air is forced out, as in coughing.

As the respiratory muscles weaken in children and young people with Duchenne muscular dystrophy, this reduces the ability to inhale and exhale air forcefully. Shoulder weakness is the earliest sign towards the onset of **respiratory muscle weakness**. It is usually the expiratory muscles that are first affected followed by the inspiratory muscles. Even the surrounding muscles of neck and back are affected resulting in drooping or sagging of shoulders. This results in the patients being unable to expand their chest to maximum limit and also causes poor clearance of mucus, which is normally expected to be cleared of the airways. All these together contribute towards collapse infection and poor lung function.

Breathing exercises have to be initiated in non ambulant patients and also in ambulant patients with weak shoulder muscles. Breathing exercises are meant to increase the breathing efficiency. It increases the diameter of airway above that to what happen in normal breathing thus helping to loosen and dislodge airway secretion. This prevents alveolar collapse, facilitates lung expansion and peripheral airway clearance.

Incentive Devices have a positive reinforcement on the patients by helping them to visualise their progress through visual indicators.

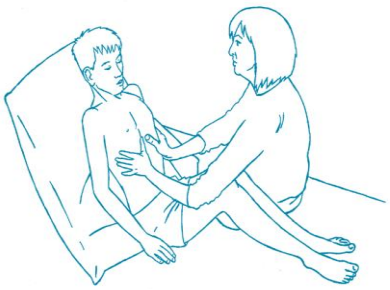
It may also be helpful to encourage your child to play a wind instrument, the recorder for example, or join a singing group. Younger children could blow party blowers or bubbles.

DEEP BREATHING

Position

The child should be reclining comfortably, with the knees bent up and two or three pillows behind the head and shoulders for support. Breathing exercises can also be done in a sitting position with the child's arms supported.

Place your hand over the lower part of the ribcage, with your fingers pointing towards the back, and apply firm but gentle pressure. Ask the child to breathe in as much as possible and try to move the ribs outwards against your hands.



POSTURAL DRAINAGE

Some children with Duchenne muscular dystrophy have difficulty clearing phlegm from their chest,particularly when they have a cold. Postural drainage can often be helpful.

Make a wedge using a very firm cushion or pile of towels, covered with a pillow or blanket. The child lies face down over these so that the hips are across the wedge and the chest is about 45 degrees from horizontal.

The child stays in this position for 10 -20 minutes and is encouraged to take deep breaths.

Pause between every two or three breaths otherwise the child may become dizzy.

POSTURAL DRAINAGE FOR OLDER CHILDREN

Older children with Duchenne muscular dystrophy who have difficulty clearing secretions on the chest may find a prone position unsuitable for postural drainage, as it can push the diaphragm up and make it hard to breathe. In this case, the child may benefit from lying on his or her side in a high supported position.

ASSISTED COUGHING

This technique can be used to clear secretions from the chest and should be done while postural drainage is taking place or when the child is sitting and leaning forward.

Encourage the child to take several deep breaths and then ask him or her to 'huff' as this helps move the sticky secretions from the furthest part of the lungs up towards the throat.

To 'huff' the child takes a deep breath and then forces the air out as hard as possible with the mouth open. Pause after two or three 'huffs' and breathe gently for a moment.

Repeat this several times with periods of deep breathing in between, and when the phlegm is near the throat, a cough will clear it.

A parent or carer can help the child to cough by placing their hands firmly round the lower ribs.

Note: **All the exercise has to be done with the guidance of a therapist initially then shall be done at home with the advice of the therapist who worked with children with Muscular Dystrophy. Too much of exercise also will weaken the muscle.**

GOOD POSTURE

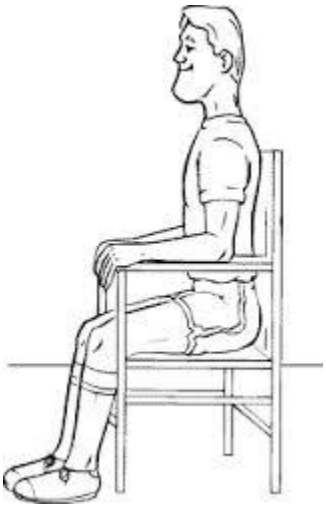
Muscle weakness in key areas such as the spine and hips can affect the posture of a child with Duchenne muscular dystrophy.

Weakness of the spine muscles can cause scoliosis (curvature of the spine), and weakened hip extensor muscles cause lordosis (a pronounced inward dip of the lower back). When one side of the child's body is stronger than the other, this can create an uneven or non-symmetrical posture.

Your child may adopt unusual postures- in sitting, standing and lying - to compensate for muscle weakness, limited mobility and contractures. It is important to correct these postures because, if left, they can cause further problems, particularly in the spine. Good seating at all times helps to maintain good posture.

SITTING

The feet should be at a 90° angle to the legs when the child is sitting down. The seat of the chair of should be firm and, ideally, not too wide. The back of the chair also needs to be firm and either upright or slightly slanting backwards (10°). The seat should be as deep as the thigh is long, so that the child is encouraged to use the back of the chair and not slump. The armrests need to be at the right height and not too far apart so that the elbows can be supported without causing hunched shoulders or leaning.



POSITIONING

The way a child moves and the positions adopted - to write, eat or rest, for example - are a direct response to losing muscle strength and having contractures. The child will naturally find the easiest and least tiring option, without thinking about it. Sometimes muscle strength and/or the stiffness of a contracture may be different on each side of the body. When this happens, an asymmetry or imbalance occurs which can cause scoliosis.

Passive stretching and night splints can delay the onset of contractures but it is important to know which positions to encourage and which to discourage, without nagging.

PRONE LYING

The prone lying position (face downwards) is good for resting. It can also help prevent contractures developing in the hips and knees. Prone lying can be combined with activities such as reading or watching television.

The child lies face down on a floor, couch or similar firm surface. Place a small pillow or wedge just below the hips (which should be level and the pelvis down) to encourage hip extension. The weight of the lower leg will straighten out the knees but it is important that the feet are free.

STANDING

Standing should be encouraged, during the day, for short periods. Standing for a few hours each day, even with minimal weight bearing, promotes better circulation, healthier bones and a straight spine. It helps to increase bone density and improve posture thereby assisting in the management of contractures.

WHY ARE ORTHOSIS USED?

To provide stability for the limb
To providing safety, stability & comfort
To prevent or reduces contractures
To prevent or correct deformity

WHAT ORTHOTIC OPTIONS ARE BEING USED?

Ankle Foot Orthosis (AFO)
KAFO
Spinal Brace
Wheel Chair

NIGHT SPLINTS

They are worn at night and are usually only for the ankles. Night splints (ankle-foot orthoses or AFOs) can be used to help control contractures in the ankle. These need to be custom-made and not provided 'off the shelf'. After the loss of ambulation, daytime splints may be preferred, but daytime splints are not recommended for boys who are still walking.

Night splints in conjunction with passive stretching are most effective way of delaying the development of contractures. Night splints are not a substitute for passive stretching. They should only be used in combination with a stretching once there is an obvious feeling of tightness.

Daytime AFOs are not recommended for ambulant children with Duchenne muscular dystrophy as these compromise their ability to walk by preventing the characteristic equinus gait. Children with Duchenne muscular dystrophy need to walk on their toes once the quadriceps muscle is weak.

Daytime AFOs should be supplied once ambulation is lost to prevent painful contractures and foot deformity.

CALLIPERS (KAFOS)

KAFO – knee-ankle-foot-orthosis, usually used to help walking or to stand. May

also be called a caliper. Long leg splints may be useful around the stage when walking is becoming very difficult or impossible. KAFOs can be useful to help control joint tightness and to prolong ambulation and delay the onset of scoliosis.

WHEELCHAIRS

The majority of children with Duchenne muscular dystrophy will need a wheelchair for transportation and independent mobility, long before they have lost the ability to walk. Choosing the right wheelchair is essential for the child's well being.

It is essential that any wheelchair, once in use, is routinely checked and monitored so that it continues to meet the child's requirements. As the child grows and their physical condition changes, the wheelchair will need to be modified to offer additional support and maintain independence.

STRETCHING

Contractures can make some movements and activities more difficult. Regular daily stretches help maintain muscle length and keep joints mobile.

There are three different types of stretches: passive, active assisted and self-stretches.

PASSIVE STRETCHES

As the name suggests, the child does not actively take part in the stretching process. Passive stretches are done by a parent, carer or therapist. Slow and firm passive stretching will not harm the joint or muscle and can be done everyday.

Tight and/or shortened muscle tissue is stretched by moving the joint as far as possible and maintaining the position for at least ten seconds (your physiotherapist may recommend longer, depending on your child's needs).

Position the child so that he or she is well supported and comfortable, and the joints not being moved are stabilised. The child must relax completely and not make any active movement or resist the stretch. If the stretching is done too quickly, the child is more likely to resist and become frightened.

ACTIVE ASSITED STRETCHES

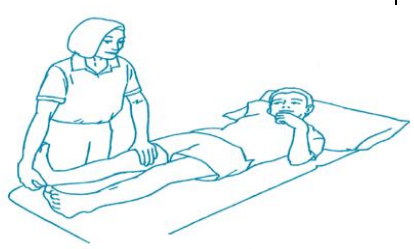
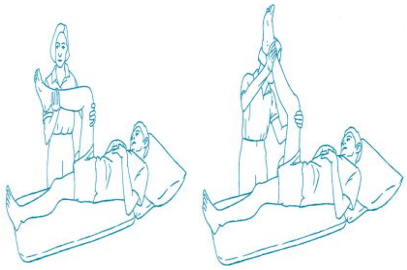
Active assisted stretches are done by the parents with the child assisting the movement. When a joint becomes contracted, the tight tissue prevents the opposite muscle group from working properly. Active assisted stretches can stretch the tightened muscle and work the opposing muscle group at the same time.

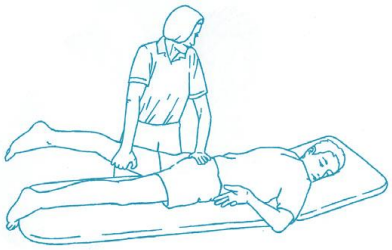
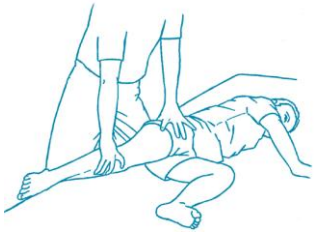
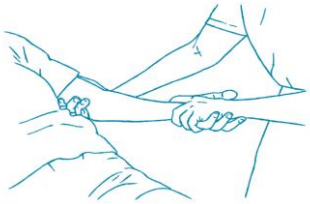
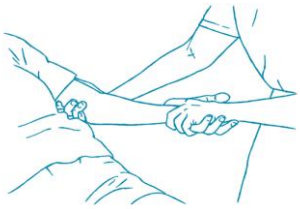
Active assisted stretches are particularly useful for the ankle. While you stretch the Achilles tendon, for example, your child pulls up his or her toes. The harder you work together, the more effective the stretch will be. This form of stretching helps the time pass more quickly and makes the stretches less boring for your child.

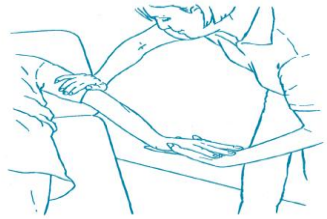
SELF-STRETCHES

Self –stretches the child is taught to do stretch by himself. These are most effective in children who are still walking and are particularly useful for the ankles, knees and hips.

HOW TO PERFORM STRETCHES?

Type of Stretch	Position	Stretch
Ankle Stretch 	• The child lies on his or her back. • Place one hand on the sole of the foot with the fingers pointing towards the heel. • Grasp the heel, firmly but gently, between the fingers and thumb. • Hold the knee straight with the other hand but do not push down on it.	• Gently, but firmly, pull down on the heel, as if trying to make the leg longer,and push the front of the foot up to a right angle (dorsiflexion) or as far as possible. • Do not let the knee bend. • If you encounter some tightness or resistance to the movement, maintain the pressure for a little and then gradually increase it again to move the foot a few more degrees. It may be helpful to ask the child to try to pull up his or her foot as you stretch.
Knee Stretch 	• The child must lie on his or her back. • Bend one leg so the hip and knee are at right angles (90 degrees).	• Gradually straighten the knee keeping the thigh still. The other leg should be kept down flat.
Hip Stretch 1 	• The child lies on his or her back and the leg not being stretched is bent up towards the chest and held in that position by the carer or, if possible, by the child.	• Place your hand just above the knee of the leg to be stretched and exert a downward pressure.

Hip Stretch 2 	• The child lies face down. • Place the hand nearest the child's head firmly on the child's bottom and push downwards.	• Use your other hand to grasp underneath the thigh of the leg nearest to you and lift the leg up as far as possible.
IlioTibial Stretch 	• The child lies on his or her side with the leg to be stretched uppermost and the knee straight. • The lower leg is bent. • Use your hand and knee to stabilise the pelvis.	• Gently move the uppermost leg as far back as possible. • Apply firm downward pressure at the knee.
Elbow Stretch 	• Stand on the same side as the elbow to be stretched. • Hold the upper arm firmly in one hand while keeping the child's palm facing up.	• Hold the wrist with your other hand and exert a gentle downward pressure to straighten the elbow.
Elbow and Wrist Stretch 	• Hold the upper arm firmly in one hand while keeping the child's palm facing up. • Move your other hand down to hold the child's hand. The grip should be as if you were shaking hands but with the fingers extended over the wrist.	• Keep the shoulder still and with the elbow bent at 90 degrees, simply turn the forearm so that the child's hand faces up.
Elbow, Wrist and Finger Stretch 1 	• Use one hand to support the forearm near the wrist joint. Keep the child's elbow as straight as possible. • Place the palm of your other hand on your child's palm.	• Move the wrist backwards, trying to keep the fingers straight.

Elbow, Wrist and Finger Stretch 2 	• The child lies on his or her front with the arm over the edge of a couch. • Grasp the upper arm with one hand.	• Place your other hand under the palm and fingers of the child's hand and straighten the elbow, keeping the wrist bent backwards and the fingers extended.
Self Stretch For Calf 	• The child stands facing the wall. • The back leg and knee should be kept straight with the heel on the floor, toes pointing to the wall.	• The child leans towards the wall, keeping the bottom tucked in, until he or she can feel the stretch in the calf of the back leg.
Self Stretch For Knee 1 	• The child sits on a floor or hard surface with hips against the wall and the spine as straight as possible. • One leg is stretched out in front, slightly to one side, with the knee as straight as possible, toes should point upwards. • The other leg is bent in so the foot touches the inner thigh of the straight leg.	• Sitting in this position will stretch the hamstring muscle of the straight leg, but the stretch can be increased by leaning forwards. • Repeat with other leg.
Self Stretch For Knee 2 	• The child lies on his or her back in the doorway or beside a post. • The child places the leg to be stretched on a doorframe or post. The knee is slightly bent and the bottom is close to the wall. • The other leg is straight on the floor.	• Straighten the knee.

CHILDREN'S QUESTIONS

Q. What is the point of stretching exercises?

A. Stretching exercises are perhaps the most important exercises for all ages and keep your joints comfortable and moving.

Q. If my muscles are weak why can't I use weight to make them stronger?

A. Using weights won't make your muscles stronger because your muscles are different from other peoples. Overworking the muscle by lifting heavy weights could damage it. But there are other active exercises which you can do that are good for you.

Q. What about my hands and arms?

A. Hands and arms need to be stretched too. Playing computer games is fun and your fingers need to be flexible in order to be an expert! Ask your physio what you can do (yes- it's more stretches!).

A. Looking good is important too. If contractures of the feet and ankle develop, this might make it difficult to wear to the soft of trainers of shoes you prefer. Wearing splints will keep your feet in the right position so that you can look good.

Q. What if it gets hard for me to walk?

A. There is nothing wrong with using a wheelchair for some of the day to save energy for playtime. Keeping your legs stretched with the help of your physio or carer will help keep you walking for as long as you can. Even when it gets really hard, there are other ways to those important steps, such as using callipers (KAFOs).

Q. Can I do any harm when stretching?

A. You will need to be very well instructed by your therapist to ensure that you understand the best holds and positioning.

There are some basic important guidelines for effective manual stretching. They are:

- Position the child to ensure that he is well-supported and comfortable
- Stabilize the joints that are not being moved by following the recommended holds
- The intensity of the stretch should be sub-maximal and should never cause pain; however,the stretch should be felt. Applying a little pressure at the end of the range should be sufficient.
- If the child resists strongly, then you should employ another method.
- Hold the stretch for up to 5 seconds and repeat the same stretch 5-10 times.
- Take care not to overstretch. Your physical therapist will explain normal ranges and expectations for your child. A stretch can be managed better if it begins gently and increases slowly to the maximum intensity without causing pain. If you meet with active resistance, use some distractions, as the lengthening contractions due to resistance can cause damage to the muscle fibers.

Q. Should my son wear night splints?

A. Night splints or ankle foot orthoses hold the feet in the most comfortable desired position for the duration of sleep. It is recommended to use these splints on a regular basis.

Q. What is incentive Spirometry?

A. Your son may be given an incentive spirometer to encourage him to expand his lungs fully at least once a day. The aim is not to train respiratory muscles, but simply to achieve as full aeration of as much of the lungs as possible and to encourage chest-wall mobility. Boys in the middle years are not running around and raising their heart rate, nor are they ever required to breathe deeply, so regular incentive spirometry may be the only way that air reaches the base of the lungs.

Q. Our son gets tired and find difficult to walk distance? Does he require a wheelchair at this stage?

A. Wheelchair can be used on a part-time basis at first, mainly for long-distance travel, such as for school excursions and lengthy walking trips with friends and family. It allows him to save his energy for the activities he enjoys rather than expending it by simply getting there.

Q. What features should I look while selecting a wheelchair?

A. Choose a chair with a firm seat. The chair should have full-length armrests, which can be positioned at the correct height. Good support for the feet should be provided at the correct height so that feet rest flat on the footplates and the thighs are fully supported. A seat belt and working brakes should be provided. The chair should be lightweight and fold for ease of transport.

GLOSSARY

GLOSSARY OF PHYSIOTHERAPY TERMS

Abduction - movement of a limb away from the body

Adduction - movement of a limb towards or across the body

AFO - ankle foot orthosis. In Duchenne muscular dystrophy this usually means night splints worn in bed to prevent the foot from pointing down

Assisted coughing - a technique used to help clear secretions on the chest

Asymmetry - when one side of the body is different to the other

Atrophy - decrease in muscle size and strength

Callipers - see KAFO

Cardiac - affecting the heart

Contracture - when a joint cannot move through full range because the muscles are tight

Distal - part of the limb the furthest away from the body (e.g.the hand or foot)

Dorsiflexion - pulling the foot up to a right angle

Extension - a straightening movement

Flexion - a bending movement

Hydrotherapy - water based exercise, usually done in a warm pool under the supervision of the physiotherapist

Hypertrophy - increase in muscle size, usually in the calf muscles (in Duchenne muscular dystrophy this is usually because the muscle contains a lot of fat)

Gastrocnemius - the calf muscle

Gluteal muscles - the buttock muscles, used to stand upright and climb stairs

Gower's sign/Gower's manoeuvre - a way of getting up from the floor into a standing position by pushing the hands on the legs

Gross motor skills - movements such as crawling, running or jumping which use the large muscles of the body

Hamstrings - the muscles at the back of the knee which help it to bend and also stabilise the pelvis

Ilio-tibial band (ITB) - the fibre on the outside of the thigh running from the hip to knee, which has muscles attached to it

KAFO - a shortened name for a **k**nee **a**nkle **f**oot **o**rthosis which extends from the toes to the hip

Lordosis - the extended position of the lower back, visible as an inward dip at the base of the spine, typical in children with Duchenne muscular dystrophy

Muscle biopsy - removing a small piece of muscle for examination

Night splints - made of polypropylene and worn at night to prevent contractures; usually start at the toe and finish just below the knee

Orthoses - another term for splints, callipers or anything worn externally to support the limb

Passive stretching - a technique used to stretch tight muscles by moving the joint as far as possible and maintaining that position

Patella - kneecap

Physiotherapy - the physical treatment or management of a disease or condition through a specially

designed programme of exercising, stretching, positioning etc

Plantarflexion (of the foot) - pointing the foot down

Postural drainage - using physiotherapy to clear phlegm from the chest

Prognosis - the expected course and outcome of a condition or disorder

Pronation (of the forearm) - turned palm down

Prone - lying face down

Proximal - part of a limb nearest to the body (e.g.the shoulders or pelvis)

Quadriceps - the muscles at the front of the thigh that straighten the knee

Resistance - using weight or manual pressure to strengthen the muscles

Rotation - a movement which turns a part of the body

Scapula - shoulder blade

Scoliosis - sideways curvature of the spine

Spinal jacket - a corset or brace made of polypropylene or leather worn to keep the spine straight

Standing frame - equipment that holds the child in a standing position with minimal effort from the child

Steroids - drugs used primarily to treat muscle inflammation but also able to slow down deterioration of the muscle

Supination(of the forearm) - turned palm up

Supine - lying face up

Tendon - the fibrous part of the muscle which is fixed to the bone

Tendo-achilles(**TA)** - the large tendon at the back of the heel

Tilt tables - equipment that can take a child from a standing position to a lying position without any effort from the child

Walking frame - equipment, usually on wheels, designed to give the child stability while walking

PSYCHOSOCIAL ASPECTS IN DUCHENNE MUSCULAR DYSTROPHY (DMD)

As we have already understood, the physical impact of DMD to the individual is tremendous. Medical care for DMD is incomplete without support for psychosocial wellbeing. Parents often find stress due to psychosocial problems (and getting them recognised) exceeds that caused by physical aspects of DMD. In India, the situation faced by persons with DMD and their families is defined by the social, economic and traditional factors that characterize Indian culture and psyche. Most psychosocial issues not unique to DMD, but children with DMD are at increased risk of problems. It is recommended that psychosocial difficulties should be treated with same effective, evidence-based interventions used in general population. The psychosocial issues faced by the children and the families can be influenced by various factors that can include:

- Biological factors : lack of dystrophin and the effect of this on brain development/function
- Social/emotional impact of the illness
- Treatment factors e.g. steroids

Difficulties faced by children with DMD

- Psychological issues:
    a. Issues with learning potential and cognitive development: There might be difficulties with learning e.g. impaired intelligence, specific learning disorders. Often there is a mild, non-progressive cognitive deficit that the child shows, that can be evident in the poor scholastic performance. Difficulty with working and short-term memory, limitations in language development and comprehension may be present
    b. Anxiety over physical decline can lead to problems with emotional Adjustment and Depression
    c. Increased risk of neurobehavioural/neurodevelopmental disorders, including autism spectrum disorders, ADHD, and OCD
    d. Oppositional/argumentative behaviour and explosive temper problems

- **Social issues:**
  Difficulties in social functioning may be due to specific challenges in particular skills, such as getting on with others, judging social situations, and perspectives, while the consequences of DMD (such as physical limitations) may result in social isolation, social withdrawal, and reduced access to social activities. The child may perceive himself as not being able to catch up with others of the same age group. This may result in his withdrawal from social activities and at times bullying from peer group.

Parents of Children with DMD

Parents also go through significant distress along with their sons, and this need to be recognized. DMD is an illness that by its progressive course, incapacitation involved and fatal outcome along with the genetic component upsets the usual parental expectations.

- Psychological issues that parents go through can involve,
    a. Coping with the day to day crisis posed by the progression of the disease
    b. Uncertainty about reversibility of these crisis episodes
    c. Communication about the disease to the child and others inside and outside the family.
    d. Experiencing and expressing emotions in the context of the child's illness
    e. Dealing with the practical care taking responsibilities, and exhaustion
    f. Fear of separation and loss – of hopes and expectations, 'normalcy' in the child
    g. Maintaining close couple relationships which tend to derail as a result of the illness in the child and the care giving responsibilities
    h. Dealing with future and end of life issues
    i. Guilt and shame that they may have been the unsuspecting cause of the illness in the child
- In the social sphere, parents tend to experience:
    a. Changing values, expectations, roles and responsibilities
    b. Difficulties in obtaining healthcare services
    c. Lack of support groups
    d. Restriction on family's freedom and social activities
    e. Loss of social network and extended family members
    f. Loss of dreams and future grandchildren $\rightarrow$loss of normal life
- Economic
    a. Limited finances
    b. Cost of medical care and special equipments
    c. Diminishing family resources

Intervention Strategies with Children with DMD

Psychosocial wellbeing is equally important as physical wellbeing in DMD. In planning management of psychosocial issues, strong emphasis should be placed on prevention/early intervention, which will maximise potential outcome. If there are any limitations in psychosocial functioning, it is important to seek help early so that effective interventions can also be instituted early.

At the time of diagnosis, neuropsychological and speech pathology assessment will help in early identification of specific cognitive and language deficits. Wherever required, psychiatric assessment will help in early identification of neuropsychiatric disorders and appropriate interventions can be instituted.

At each follow up visit, screening of emotional status and adjustment need to be done.

Close monitoring with systematic, routine follow-up is suggested, including consultation with the appropriate specialist if concerns arise.

The intervention should focus on:

Developing social and learning skills: Social skills training as needed to address deficits.

Pain management strategies

Providing emotional support for the child: the emotional distress that the child goes through often gets overshadowed by the physical limitations of the condition. The child's anxiety in response to the physical decline, 'Why I am different from other children', negative attitudes to himself and the outside world need to be addressed through appropriate mental health interventions.

Enabling and supporting child to reach personal goals: The children with DMD need to be treated as individuals with their own choices and wishes. Promoting their independence and decision making skills in is also essential, as is giving them time and space to accomplish things on their own.

Intervention Strategies with Parents of Children with DMD

Proactive intervention is essential to help avoid social problems and social isolation. Interventions should support broad spectrum of needs, but will vary depending on individual child's and family's resources and limitations.

- **Information needs of the parents** have to be met and emotional support provided for coping with the diagnosis and its impact on the child. Gaining understanding about the condition helps to adopt a realistic view of the situation, and this, in turn, helps in the process of acceptance of the chronic illness in the child. It also helps them to educate others, including the child's siblings, extended family members, friends, neighbors, teachers and others with whom the child comes in contact, as well as the child about the disease. Address issues related to grief, loss and bereavement in the supportive sessions.
- **Manage the child's condition on a day-to-day basis and cope with periodic crisis:** Taking each day as it comes is a way to cope with the demands of the disease. Taking small steps, and prioritizing what to do help to have a clearer picture of the demands and tasks. Clear description of problems, listing out possible solutions, generating advantages and disadvantages of strategies used for these solutions, identifying possible obstacles that might be encountered and how these might be handled, alternative means of approaching these goals, choosing the best possible strategy and implementing it, helps in handling the crisis.
- **Facilitate communication between family members and between them and the medical team**: In this, a care co-ordinator can be of great help, who will be able to guide the family through the health care system.
- **Parental management training**: recommended for externalising behaviours (e.g. noncompliance/disruptive behaviour and parent-child conflict)

- **Establish a support system:** The parents need to establish a support system to cope with the demands of the disease. The manifest concern shown by the relatives, extended family, especially in the practical tasks will help to tide over the crisis situations.
- **Identify sources of support from governmental and non- governmental sectors**
  Many families do not have the money to pay for the fare to take the child for regular consultations and physiotherapy sessions. Also, the progression of the disease makes it necessary that one parent stay at home to look after the child. This affects the economic stability of the family also. Hence, whatever available support they can receive for the care. Networking with Parent Associations, NGOs and special schools is helpful in dealing with the demands of the disease

- **Enable parents to meet child's normal developmental needs and those of other family members**
  While suitable allowances must be made for the child's handicap, parents must none the less, stimulate the child to maximum achievement at a realistic level. For most parents, this balance between protecting him and helping him to grow is difficult to strike. As with the well children, a child with DMD must learn how to capitalize on his abilities and strengths and at the same time, accept his limitations. The parents' attitude and encouragement in this process is crucial. The need to strike a balance between the needs of the ill child and those of the other family members is also important.

- **Involving the school in child's care**: Proactive approach important in increasing DMD awareness/knowledge among school personnel. Peer education about DMD will help in reducing negative attitude and bullying from the peers.

Hence, caring for DMD moves beyond the realm of caring for the physical limitations alone, and equal attention need to be given to psychosocial wellbeing of the children affected with DMD and their families.

References: Bushby, K., Finkel, R., Birnkrant, D.J., Case, L.E., Clemens, P.R., Cripe, L., DMD Care Considerations Working Group (2009). Diagnosis and management of Duchenne muscular dystrophy Part 1: diagnosis, and pharmacological and psychosocial management, *Lancet Neurology*, 9(1), 77-93. Epub 2009 Nov 27.

Nutritional Recommendation in Muscular Dystrophy

What type of diet that I should follow for my Child?

Healthy diet is what you should be giving your child. Since the drug, which is prescribed to your child, can produce weight gain by means of fluid retention, increasing appetite and body fat distribution. The increased body weight becomes burden on the already weak muscles and also obscures all benefits of the drug. Maintaining **Ideal body weight** becomes the target. So a diet based on high protein contents, fiber and low in carbohydrates and fat would be ideal.

What do you mean by low carbohydrate diet?

Avoid taking Sweets, Chocolates, cookies, cakes, Candies, Refined cereals, Pizza, sweetened cereals, fruit juices with sugar.

Eat more of **whole grain cereals** and consume only the required quantity.

Can you tell me which are the foods, which are high in fat?

Fried foods [deep fried in oil] like Poori, Samosa, Chips, Appalam, Vadai, Butter, Cheese, Ghee, and fast foods. All non-veg foods are high in fat especially Red meat, organ meats, Shellfish. So it is better to take Lean meat, Chicken without skin, Small Fish all in grilled or gravy form and not fried in oil.

Milk you can give as **Skim milk** instead of whole milk.

What Vegetables and fruits I should give my Child.

You can give all vegetables and Fruits. These are high in fiber. It is always better to give as fruits than as fruit juices. You must give around 300gms of vegetables [4 katories] and 100 Gms of fruits [2 fruits] per day.

Root and tuber vegetable like Potato, Yam, in fruits Bananas and Grapes can be given once a week.

What is the meal pattern I should follow for my child?

It is better to have small and frequent meal consisting of 3 main meals and 2 small Nutritious snacks.

What does Nutritious Snack mean?

Low fat snacks like Fruits, Vegetable salad, Sprouts, Dhoklas, Sandwich with green vegetable, Buttermilk, Soups, Nuts. Avoid Salty foods.

What are high protein foods?

Milk, Egg, Pulses, Legumes, Nuts and Non-Veg are high in protein

For your child you must be giving non-veg foods as said before, skim milk, whole egg thrice in a week. You can give dhal, Channa, soybean, Karamani etc more in the diet.

What oil can I use for cooking?

Combination of MUFA and PUFA oil will be better. For example Refined Ground nut oil and Sun flower oil. This can be alternately used in cooking.

As my child have bone and muscle weakness, do I need to concentrate on calcium?

Meeting the calcium requirements is very important. The RDA for calcium is 600 –800 mg/day. For better absorption of calcium, vitamin-D has to be provided in its appropriate quantity. The RDA of vitamin-D is about 400 IU/day. Calcium rich foods like milk and milk products; green leafy vegetables, Ragi and egg have to be included in the diet.

In all, good dietary habits, right choice of food both in quality and quantity following regular meal pattern have a beneficial effect.

Parent's voice on DMD

It was 7 and 1/2 years back that my son was tested positive for Duchenne Muscular Dystrophy. It was quiet a sad moment for us and all in our family circle. To recover from that and adopt the way the boy lives is the way of life we are practicing now. There are lots of inputs based on DMD from all over the world. But to put him into good health parents have to do more home work. Like what he likes to eat, play and keeping them busy in their daily routine is the more important one which we have to monitor.

Regular check up with the concerned doctor help us a lot to take preventive measures needed for the boys, like providing flu vaccination, monitoring lung function, diet etc. It is true like all the other boys in their earlier childhood our boys would like have to all food items. But to maintain the boy's weight we have to change the food style in our family. Monitoring the weight is very important in order to have easy mobility.

Learning is another factor which worries us more. Since these boys are prone to tiredness during writing, we have to spend more time and help them out to complete their home work. At school we have to keep informing the teachers about his inability to climb staircase, standing from the sitting position and frequent falling. Every day constant monitoring of the school happenings with the boy helps us a lot to update the teachers and keep him comfortable in the learning. It is good to slowly inform the boy about his muscle problems so that he can understand his inability to do things like others do. Things become lighter when the boys understand that.

Routine physiotherapy reduces pain and increase flexibility of the muscles. In our experience sharing our thoughts with other parents helps us more to know about the child's growth and happenings which is going to occur in the later stage. On doing this our awareness increases and helps to take some precautionary measures.

It is quiet natural that all parents will like to keep their children in good health and adopt good practices. To do that the Duchenne families have to meet regularly and discus topics related to learning, growth, food, exercise and checkups etc. Conducting classes by good knowledgeable persons in the related subjects like diet, exercise etc. will increase the parents' awareness towards each topic and also keep the boys fit. At least once in three months news circular about research happenings, physical exercise, good healthy food recipe, tips like what to do and what not to do can be given to parents .

All children and young men with Duchenne Dystrophy everywhere need and deserve access to accurate and timely diagnosis, genetic testing, state-of-the-art care, the opportunity to participate in clinical trials and access to promising treatments.

Often we worry that our waiting may be fruitless but there is meaningful progress, and that not only research advances, but medical care for the boys and young men gets better and better. Quite importantly the public and the governments and their health agencies should realize that Duchenne is a worldwide problem and together we must *end Duchenne*.

Challenge before Us

Though our kids have less active movement, they have good mind to compete with the normal kids in various activities. We the parents have to identify the apt one and make them do wonders in that line. On doing this there may be lot of tough challenges for us which we have to be mentally prepared and positive to exploit their skills and convert into a useful one for them to be used in their future life.

Where we stand

Knowing the facts and the future of the kids it is not so easy to think about a curriculum and extracurricular activities which will help them to survive this beautiful world. We are so distributed geographically but there are a lot of ways to bring us close using modern technology. We are far far behind or underutilized.

How to achieve

It is high time that all duchenne family members should act together and develop new concepts and methodology for kids which will help them to lead a hassle free life. For this frequent gathering of the families have to take place to understand each members kids and bring a method which will help to keep them happy in all aspects.

One of the ways is to develop an Internet site which can make us and our kids to share each other's information and bring more useful and simple concepts for them to practice.

Areas to concentrate and work are like schooling, extra circular activities, how to keep their health good, apt method to handle the kids on a longer run.

Rather than thinking about facing the difficulties and obstacles for our children we have to think about new concepts and methodology to make our children happy and independent. This should be our vision in the longer run.