

Indian Academy of Pediatrics (IAP)



## GUIDELINES FOR PARENTS

# Care of a Child with Duchenne Muscular Dystrophy

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### 10 FAQs on CARE OF A CHILD WITH DUCHENNE MUSCULAR DYSTROPHY

1. What is DMD and why does it occur?
2. In my family no one has any medical problem, then why my child has this problem?
3. What are the other problems, my child can have with time?
4. What are the treatment options available for treating my child? Is there any treatment that can cure my child's disease? Is there any role of stem cell therapy for the treatment of DMD?
5. What about my child's education and future? How much time he had got?
6. You mentioned that steroid therapy is a treatment option. What are the side effects of steroid therapy and what precautions should be taken?
7. How can we make the life of my child comfortable and productive?
8. What kind of diet should be given to my child?
9. You had mentioned that my child will require regular physiotherapy exercises. Should I take my child for formal physiotherapy sessions everyday, and what are the exercises I can do at home?
10. What are the chances of my next child having DMD? Do other kids in my family have a risk of developing DMD? Can we diagnose the disease before a baby is born?

**Under the Auspices of the IAP Action Plan 2021–2022**

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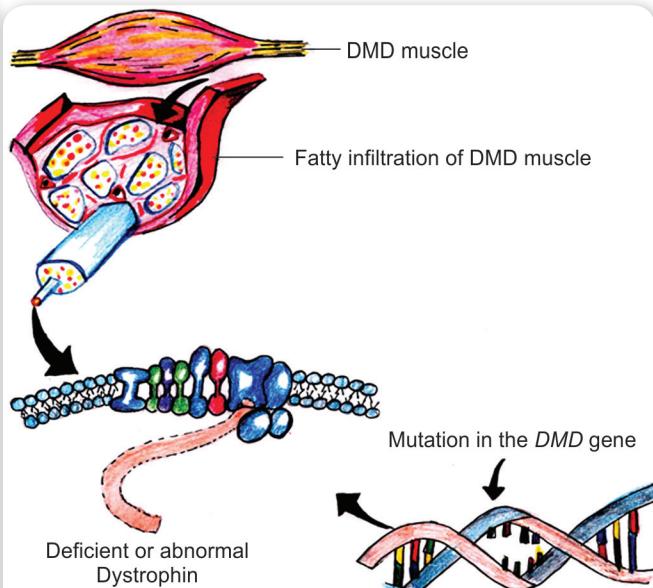
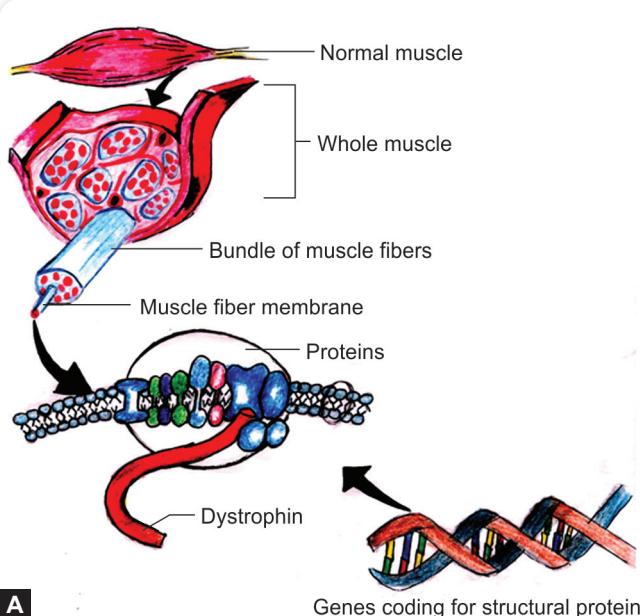
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# Care of a Child with Duchenne Muscular Dystrophy

**Q1**

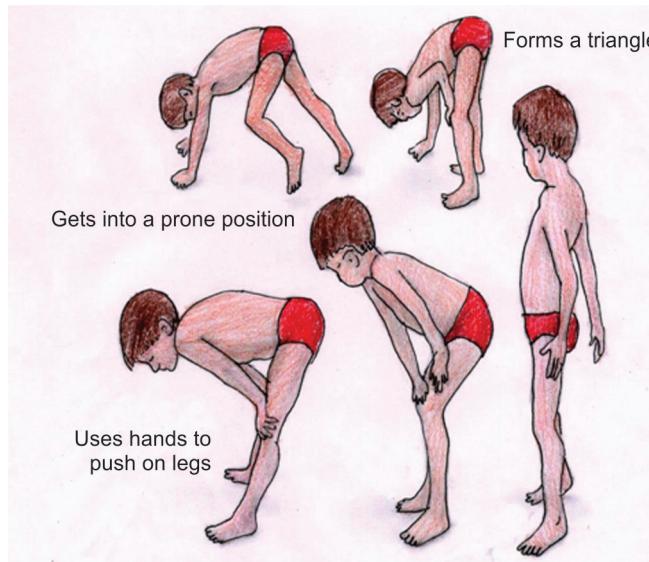
## What is DMD and why does it occur?

- Duchenne muscular dystrophy (DMD) is a genetically inherited neuromuscular disorder characterized by progressive muscle wasting and weakness.
- DMD occurs due to a mutation (harmful alteration) in the *DMD* gene, which is located on the X-chromosome. The gene is responsible for producing a protein called dystrophin which normally functions to protect muscle fibers (**Figs. 1A and B**). In absence of dystrophin protein, muscles are broken down by enzymes leading to progressive damage and dysfunction of muscles.

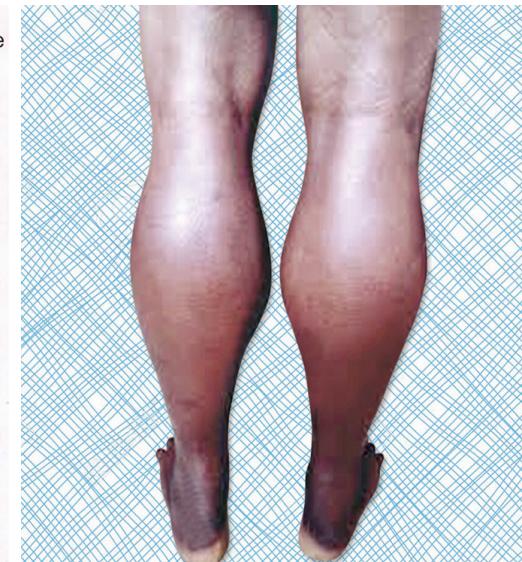


**Figs. 1A and B:** Pathogenesis of Duchenne muscular dystrophy (DMD). (A) Normal muscle; (B) DMD muscle.

- DMD is predominantly seen in boys and occurs in 1 of 3,500–5,000 newborn males. It affects girls at a much lower rate.
- Initially, DMD affects the muscles of legs, and children are noticed to have frequent falls while walking, difficulty in getting up from squatting position—Gower's sign (they try to get up from the floor by holding on to their knees) (**Fig. 2**), difficulty in climbing stairs and swelling of calf muscle (**Fig. 3**). With age, gradually children develop weakness of upper limb, neck, truncal, respiratory, and cardiac muscles.



**Fig. 2:** Gower's sign. The child assumes the hands and knees position and then climbs to stand by "walking" his hands progressively up his shins, knees, and thigh.



**Fig. 3:** Calf hypertrophy.

Q2

## In my family no one has any medical problem, then why my child has this problem?

A boy develops DMD, if he has the disease-causing mutation in his X-chromosome. A girl who has the same mutation becomes a "carrier" and usually does not manifest the symptoms of DMD, but can pass on the mutation to her children. About 50% of the boys born to a carrier mother may have DMD. Similarly, there is a 50% chance that a girl born to a carrier mother becomes a carrier herself. Hence, in a family, DMD may not be evident because there may be carriers and unaffected boys. Rarely, DMD may also occur in a family due to the mutation occurring for the first time called as "*de novo mutation*".

**Q3**

### **What are the other problems, my child can have with time?**

- Children with DMD have a poor growth rate and can have short stature even in kids who have not been given steroid therapy.
- As the disease progresses, your kid can have swallowing difficulties, choking episodes, and nasal regurgitation of feeds while feeding. This is because of the weakness of oropharyngeal muscles.
- Cardiac dysfunction is seen in half to three-fourths of these children with DMD.
- Children with DMD frequently have mild cognitive impairment or global developmental delay.
- Children with DMD have increased rates of autism, attention-deficit hyperactivity syndrome, obsessive-compulsive disorder, and anxiety disorder. Also, family members of children with DMD have an increased risk of depression and anxiety.
- *Orthopedic complications:*
  - Children with DMD have an increased risk of ~20% of fractures of arms, legs, and spine fractures which are caused because of frequent falls and decreased bone density.
  - In all patients with DMD progressive deformity of the spine (scoliosis) develops which along with the respiratory muscle weakness causes difficulty in breathing.

**Q4**

### **What are the treatment options available for treating my child? Is there any treatment that can cure my child's disease? Is there any role of stem cell therapy for the treatment of DMD?**

Early identification of children affected with DMD is important to prevent complications and prolong the lifespan of the patients. The management of a child with DMD encompasses: (a) disease-modifying therapy; and (b) prevention and management of complications.

### Disease-modifying Therapy for DMD

- Presently, there is no curative treatment for DMD. This means that the therapies which are available, are to reduce the complications and speed of progression of the disease.
- Steroid therapy (prednisone or deflazacort) is the primary treatment available for DMD and has been shown to increase muscle strength and function, lung function, and decrease the progression of scoliosis.
- Upcoming treatment options such as eteplirsen, ataluren, golodirsen, and viltolarsen and gene transfer therapies for DMD are in various stages of research and are showing promising results. There are several drug trials going on across the world and your doctor will be able to guide you for those available in India and whether your child is fit for it.
- Presently, there is *no role of stem cell therapy for the treatment of DMD*.

### Prevention and Management of Complications

- *Physical therapy* to maintain muscle function and prevent joint stiffness and deformity of bone and skin. This part is very important, whether or not your child receives any drug treatment.
  - Regular active- and passive-stretching exercises (**See Tables 1 and 2**)
  - *Activity-based therapy:* Hydrotherapy, cycling, yoga, etc.

**TABLE 1: Active-stretching exercises for patients with Duchenne muscular dystrophy (DMD).**

	<i>Position</i>	<i>Stretch</i>
<b>Self-stretching for Calf Muscle</b> 	With his face and toes pointing toward the wall, the child stands with his front leg bent and the back leg and knee kept straight with the heel on the floor	The child is asked to lean toward the wall, keeping his bottom tucked in until he can feel the stretch in his calf in the back leg
<b>Self-stretching for Knee 1</b> 	<ul style="list-style-type: none"> <li>• The child sits against the wall with his hips and spine kept straight on a hard surface</li> <li>• He is asked to keep his one leg stretched out in front, slightly to one side with the knee kept straight and toes pointing upward</li> <li>• His other leg is bent so that the foot touches the inner thigh of the opposite leg</li> </ul>	<ul style="list-style-type: none"> <li>• Stretching of the hamstring muscles of the straight leg is achieved by sitting in this posture. The degree of stretch can be increased by leaning forward</li> <li>• Similarly, stretching of the hamstring muscles of the other leg should be done</li> </ul>

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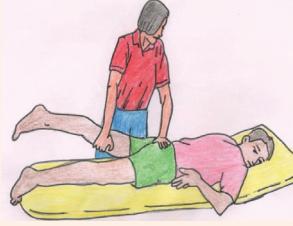
	<i>Position</i>	<i>Stretch</i>
<b>Self-stretching for Knee 2</b> 	<ul style="list-style-type: none"> <li>The child should lie on the floor with his face upward either in the doorway or beside a post</li> <li>The child places one of his legs to be stretched on the doorframe or post with his knee slightly bent and his bottom close to the wall</li> <li>The other leg is kept straight on the ground</li> </ul>	The child is then asked to straighten his knee

**TABLE 2:** Passive-stretching exercises for a child with Duchenne muscular dystrophy (DMD).

	<i>Position</i>	<i>Stretch</i>
<b>Ankle Joint Stretching</b> 	<ul style="list-style-type: none"> <li>Child lying on back with knee straight</li> <li>Place your one hand on the sole holding the heel and one hand on the knee</li> </ul>	Gently pull the heel downward and try to push the front of the foot upward keeping the knee straight
<b>Knee Joint Stretching</b>  	Make your child lie on his back and then bend his leg so that the hip and knee joints are at 90°	Slowly straighten the knee keeping the thigh still meanwhile keeping the other leg flat
<b>Hip Joint Stretching 1</b> 	Make your child lie on his back with the one leg not being stretched is folded up toward the chest and held in that position by you or your child	Keep your hand above the knee of the leg to be stretched and press downward

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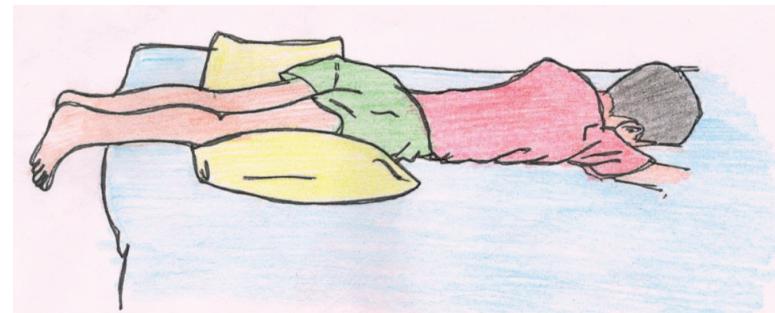
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	<i>Position</i>	<i>Stretch</i>
<b>Hip Joint Stretching 2</b> 	Make your child lies face down on his tummy and put one hand firmly on his bottom and push downward	Hold from below the same thigh with your other hand and try to lift the leg as far as it is possible
<b>Iliotibial Stretching</b> 	Make the child lie on his side with the leg to be stretched pointing upward and keeping the knee straight. The lower leg is folded. Stabilize the pelvis of your kid with your hand and knee	Slowly try to move the uppermost leg as far back as possible simultaneously applying firm downward pressure on the knee
<b>Elbow, Wrist, and Finger Stretching</b> 	<ul style="list-style-type: none"> <li>• Hold your child's upper arm firmly in one hand while keeping the palm facing upward</li> <li>• With your other hand support his forearm just above the wrist joint with your palm touching the palm of your child</li> </ul>	Move the wrists backward trying to keep the fingers straight

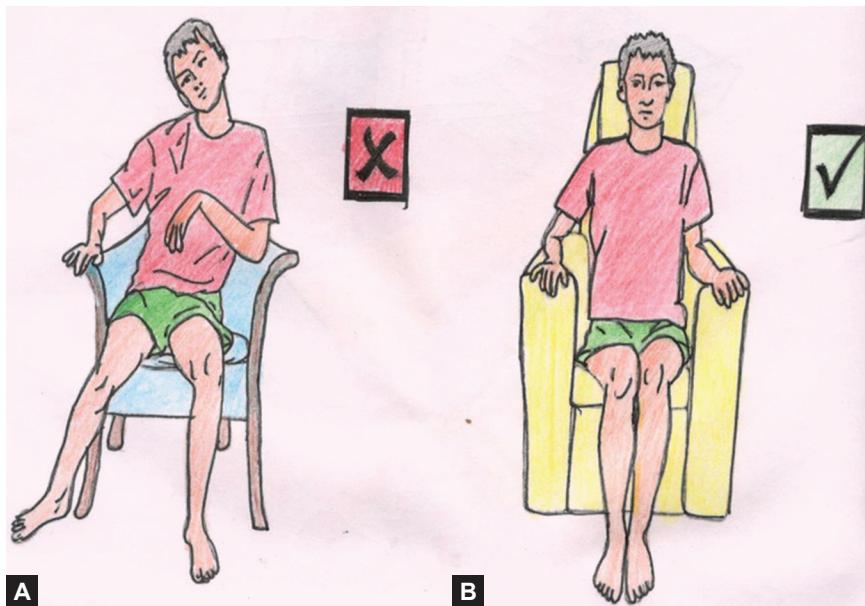
- **Proper positioning:** Because of muscle weakness and spine deformity, your child can adopt an unusual posture while sitting, standing, or lying as a compensatory mechanism. It is important to correct these postures because if left untreated can lead to further deformity and disability. Prone-lying position (face downward) is good for resting and helps in preventing development contractures of hip and knee (**Fig. 4**). Sitting should also be done in a well-supported not too wide chair and the feet should be at 90° to the legs when the child is sitting down (**Figs. 5A and B**). Standing should be encouraged for short periods during the day.

### Respiratory Care

- Children with DMD should be vaccinated for the pneumococcal vaccine and yearly influenza vaccine to prevent pneumonia.

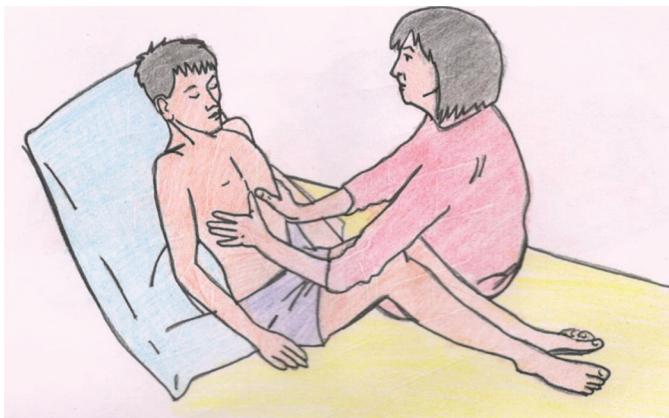


**Fig. 4:** Prone-lying position.



**Figs. 5A and B:** (A) Bad-sitting posture; (B) Good-sitting posture.

- Lung function testing should start when the child is 9–10 years old and should be repeated yearly before the child becomes wheelchair dependent. After the child becomes nonambulatory, it should be done at 6-month intervals.
- Chest physiotherapy (**Fig. 6**) and postural drainage (**Table 3**) in case of respiratory infections or poor respiratory function.



**Fig. 6:** Breathing exercises.

#### Position

- The child sits comfortably reclining on two or three pillows kept behind the head and shoulder with his knees folded up.
- Or, the child sits in a chair comfortably with his arms supported.

#### Exercise

- Hold your child's lower part of the ribcage with your hands with your fingers pointing to his back simultaneously applying gentle yet firm pressure.
- The child is asked to breathe in as deep as possible moving his ribs outward toward your palm.

**TABLE 3:** Postures to improve respiratory secretions drainage in children with Duchenne muscular dystrophy (DMD).

	<i>Posture</i>	<i>Technique</i>
<b>Young Children</b>	 <ul style="list-style-type: none"> <li>The child is made to lie face down on a pile of very firm cushions or a pile of towels, which have been covered with a pillow or blanket</li> <li>Ensure that the child's hips are across the wedge and his chest is around 45° from the bed</li> </ul>	<ul style="list-style-type: none"> <li>Ask the child to lie in this posture for 10–20 minutes and take deep breaths</li> <li>To prevent the child from becoming dizzy pause after every 2–3 breaths</li> </ul>
<b>Older Children</b>	 <p>The child lies comfortably in a highly supported position on his side</p>	<ul style="list-style-type: none"> <li>Ask the child to lie in this posture for 10–20 minutes and take deep breaths</li> <li>To prevent the child from becoming dizzy pause after every 2–3 breaths</li> </ul>

- Assistive devices such as night-time and day-time nasal bi-level or two-level positive airway pressure (BiPAP) device in case of poor respiratory efforts.

#### Cardiac Care

- Screening to look for cardiac dysfunction is done by electrocardiogram (ECG), echocardiography (ECHO), cardiac MRI (in kids with severe scoliosis and chest deformity), and cardiologist opinion. Your doctor will be able to guide you regarding these tests.
- Screening should be done annually in children with DMD and more frequently if symptoms are present. Carrier females should also be screened beginning from late adolescence and early adulthood.
- Oral medications will be prescribed for the prevention and management of the cardiac disease.

#### Bone Health and Orthopedic Care

- Children with DMD at the time of diagnosis and annually should be evaluated for calcium and vitamin D deficiency. Bone mineral density should be checked annually by dual-energy X-ray absorptiometry (DEXA) scan.
- It is advised to give a diet rich in calcium and vitamin D (milk and milk products).
- If a deficiency or fractures are identified oral medications or intravenous (IV) drugs might be given to your kid.
- Wearing ankle-foot orthoses (splints) at night-time based on the requirement to prevent the development of ankle contractures.
- Standing or walking may be maintained by using long leg braces.
- Wrists or hand splints for stretching of wrists and fingers in nonambulatory stages.
- Assistive devices for nonambulatory patients:* Crutch, walker, wheelchair, etc. can help to maintain mobility.
- Corrective surgery to release fixed contractures or children having significant spinal deformity.

Q5

### What about my child's education and future? How much time he had got?

- Kids with DMD have difficulty in climbing stairs roughly at around 7–8 years of age and unable to walk by 12 years of age and require the use of wheelchair/motorized chairs.
- Children with DMD should get normal education with support depending upon their level of intellectual impairment. This ensures that they have a good quality of life.
- Proactive efforts to educate the teachers, school, and peers regarding various aspects of DMD should be done by the parents which help in reducing negative attitude and bullying from peers.
- Help and cooperation from the school should be sought for achieving barrier-free access to washrooms, toilets, and classrooms. Also, computer-assisted education should be encouraged for your kids.
- Life expectancy is shortened with DMD but with early treatment and initiation of noninvasive ventilation children with DMD are now surviving into adulthood. The average lifespan of kids with DMD is about 18–25 years. ~50% of kids with DMD attain the age of 25 years but still, survival beyond the 3rd decade of life is uncommon.

Q6

You mentioned that steroid therapy is a treatment option.

What are the side effects of steroid therapy, and what precautions should be taken?

- All drugs have their side effects, so does steroid therapy.
  - *Minor side effects:*
    - *Heartburn and regurgitation:* To treat/decrease symptoms give the drug with food, increase water intake, avoid tea/coffee/spicy food/cold drinks; use antacids.
    - *Increased appetite:* It can be handled by providing a healthy diet that is low in carbohydrates and fats, and high in proteins, fresh fruits, and vegetables.
  - *Major side effects:*
    - Increased blood sugar levels
    - High blood pressure
    - Overweight and obesity
    - Steroid-induced cataract and glaucoma
    - Steroid-induced growth retardation
    - Steroid-induced decreased bone density
- Hence, the following recommendations should be followed to prevent and early detect these side effects.
  - Sugar levels should be monitored regularly. Initially, every 6-month hemoglobin A1c (HbA1c) should be done. If a family history of diabetes is present, monthly monitoring of random blood sugar (RBS) should be done.
  - Weight should be monitored monthly and excess weight gain should be avoided as it will further deteriorate the symptoms.
  - Blood pressure monitoring should be done monthly. If present, antihypertension drugs should be started.
  - Regular eye check-up once in a year is a must, if the steroids are used for a long period.
  - Calcium and vitamin D should be supplemented under the guidance of the treating physician.
- These side effects can be minimized by the use of some other types of compound (deflazacort) or a different regimen (alternate day or pulse therapy). Whatever it may be “Never stop the drug suddenly”. Always consult your doctor first.

Q7

### How can we make the life of my child comfortable and productive?

- The child should be helped to adapt to the supportive therapy and should be gently and gradually told about the nature of his muscle weakness and physical limitations.
- Environment modifications should be done to avoid the risks of falls and injuries as follows:
  - Removal of obstacles such as rugs, toys, cords, and clutter, etc.
  - Handrails on both sides of stairways
  - Nonslip mats in bathroom and bathtub
  - Western toilet installation in the washroom
  - While using wheelchairs to always use seat belts and use antitippers
- The child should be encouraged to do the drawing, use the computer and smart devices to learn. Encourage the child to listen to stories, online book readings, and to participate in quizzes. As long as the child is mobile, he can go to school, later on, participation in online learning courses or classes can be done.
- The child should be involved in the day-to-day activity and decision making of home affairs, to make him realize that he is an active member of the family. Music especially instruments such as synthesizer or flute can be learned. If efficient, the child can be made a part of an online business. Big applause should be given to him for all of his achievements.
- Remember, it is neither the parents' nor the child's fault that the family has DMD. So stay positive and improve the quality of life with your child.

Q8

**What kind of diet should be given to my child?**

- Children with DMD based upon their disease status and treatment provided tend to develop malnutrition either undernutrition or overnutrition. As the disease progresses because of decreased physical activity kids with DMD have lesser energy requirements. The use of steroids leads to increased appetite and food intake with increased water and salt retention. Some kids with DMD may develop feeding and swallowing difficulty which can lead to weight loss and undernutrition. Hence, the dietary plan for each child should be individualized by taking into consideration his weight, height, body mass index, level of physical activity, and any feeding difficulty.
- A balanced diet low in carbohydrates and sugars and rich in pulses, proteins, fruits, and vegetables should be given to your child.
- Avoid giving sweets, chocolates, cookies, refined cereals, and white bread to your kid. You should give whole-grain cereals and that too in the desired amount as recommended.
- A diet rich in calcium and vitamin D such as dairy products with adequate sunshine exposure should be provided.
- Kids with DMD should be encouraged to drink plenty of fluids—*<10 years: 5–8 cups, 10–20 years: 8–11 cups, >20 years: 13 cups per day* to prevent constipation.
- If your kid develops severe feeding difficulty, he may require nasogastric tube or gastrostomy tube feeding. If your kid has repeated episodes of regurgitation or complains of heartburn, he may be prescribed antireflux medications for the same.

Q9

**You had mentioned that my child will require regular physiotherapy exercises. Should I take my child for formal physiotherapy sessions everyday, and what are the exercises I can do at home?**

- Kids with DMD require a multidisciplinary assessment of motor function every 6 months or more frequently, if parents notice any deterioration or new complaints.
- It is not required to take all kids with DMD for formal physiotherapy sessions everyday, the frequency of formal physiotherapy sessions are decided based upon your kid's needs, stage of illness, response to therapy, tolerance, etc.
- Children with early stages of DMD should be encouraged to do regular gentle aerobic exercises such as swimming or cycling or yoga to prevent muscle wasting, excessive weight gain, and social isolation. Children with difficulty in mobility can do assisted exercises such as cycling and swimming, etc.
- It is important to note for any significant muscle pain or presence of reddish or cola-colored urine within 24 hours after a specific activity. These may indicate overexertion and that activity should be reduced or stopped completely.
- Overexertion, eccentric muscle exercises, yoga power poses, and high-resistance strength training or activities should be avoided as they can lead to further damage to the already fragile muscles.
- Preliminary studies have shown the benefit of 10 minutes standardized calf massages in reducing muscle stiffness and improving muscle length. Gentle calf massage for around 10 minutes can be given daily to children with DMD.
- Regular home-based daily stretching exercises are a must to prevent the development of contractures and deformities. Special attention should be given to stretching exercises involving the hips, knee, and ankle.
- When kids with DMD lose the ability to walk, it is important to do regular stretching exercises of the upper limb also.
- For the preservation of respiratory muscle function, children can be encouraged to do deep breathing exercises; blowing whistles, and balloons and making bubbles with soap and water.

Q10

**What are the chances of my next child having DMD? Do other kids in my family have a risk of developing DMD? Can we diagnose the disease before a baby is born?**

- There is a 50% chance of your other sons having DMD. Your daughters have a 50% chance to carry one of the abnormal copies of genetic material and are known as DMD carriers. Female carriers are usually asymptomatic but a minority ~20% can develop a variable degree of weakness and can have cardiac problems.
- In your family, your grandchildren have an increased risk of developing DMD and to be asymptomatic carriers depending upon the mutation status of their parents and adequate genetic counseling should be taken while planning for pregnancy.
- Prenatal diagnosis of DMD that is the diagnosis of DMD before the baby is born can be done by noninvasive tests such as preimplantation genetic study or invasively by chorionic villus sampling. In both tests, the fetal genetic material is analyzed to look for the abnormal DMD genetic mutation. It is advised to get these tests done during the early months up to 4–5 months of pregnancy.