

Duchenne’s Muscular Dystrophy (DMD)

Often, children are not diagnosed with DMD until after age three. You may begin to see symptoms prior to age three, but onset can vary in each child. If your child is diagnosed at this early age, the following will be important for the **first phase of your child’s life (infancy to 3 years)**.

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| <p>What kind of professionals might my child need for his/her first few years of life?</p> | <p>Primary Care Physicians (Pediatricians) will help to manage your child’s care consistently. Refer to their expertise in times of transition or change as well.</p> <p>Cardiologists will monitor your child’s heart.</p> <p>Orthopedists will be important when addressing musculoskeletal issues.</p> <p>Pulmonologists may not be needed for the first few years of life, but often are needed later to provide your child with respiratory and lung care.</p> <p>Physical Therapist: see “therapies” for a complete description.</p> <p>Occupational Therapist: see “therapies” for a complete description.</p> <p>Respiratory Therapist: see “therapies” for a complete description.</p> |
| <p>What conditions may affect my child?</p> | <p>-Delayed Development of Motor Milestones (walking, crawling, etc.) will often be one of the first signs of Duchenne’s.¹</p> <p>-Pseudohypertrophy is the appearance of enlarged muscles. Your child’s body is laying down scar tissue instead of reforming muscle, so muscles such as the calves will appear large and often feel harder than normal. This should not be a painful condition for the child¹</p> <p>-Intellectual Disability is present in about a third of children with DMD.¹ There are most often deficits seen in short-term memory, visual/spacial long-term memory and verbal skills.²</p> |
| <p>What kind of surgeries will my child need? When?</p> | <p>Often, in this first phase of life no surgeries will be needed. If surgery is necessary, it will be in response to contractures (abnormal shortening of one’s muscles that can lead to faulty joint function and limited movement).</p> <p>When contractures have advanced, the following might be necessary:</p> <p>Tendon Release Procedure</p> <ul style="list-style-type: none"> -On the effected muscle, the surgeon will cut the tendon to allow for a relaxation of the muscle.³ -Often, there is a loss of strength associated with the procedure, but the release will allow for a more functional movement of the area involved.³ -This surgery is often done in hopes of maintaining the child’s ability to walk.¹ |

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| <p>What kind of assistive devices are available?</p> | <p>At this age, your child should not need many, if any, assistive devices. Your child may begin to display weakness and a lack of balance, but most will still be able to walk on their own for the first several years of life. The likely assistive devices needed at this age would be orthotics or splints.</p> <p>Night Splints: The use of night splints (also known as ankle and foot orthotics/AFOs) on the ankles of children with DMD has shown promising results. Data shows that they may contribute to extended time of independent walking (when used in conjunction with daily stretching).⁴</p> <ul style="list-style-type: none"> -Daytime use of AFOs is usually avoided because it places a greater stress on the leg and often makes walking even more difficult.⁶ |
| <p>What kinds of therapies are available?</p> | <p>Drug therapy:</p> <p><i>Corticosteroids:</i> This is the main class of medications that have been found to slow the course of DMD.¹ Some physicians begin drug therapy as soon as a diagnosis is made, while others wait until difficulty with walking occurs. Be sure to discuss these options with your physician.</p> <ul style="list-style-type: none"> -<i>Prednisone:</i> This is the drug usually chosen by doctors. It has been seen to “increase strength, timed muscle function, and pulmonary function.”¹ -Be sure to watch for side effects such as: weight gain (often associated with a rounded, puffy face), cataracts, fractures as the result of thinning of bones, behavioral problems.⁵ (A low-calorie, low-sodium diet is usually recommended to help decrease the weight gain and fluid retention often seen with this drug.)¹ If any of these symptoms are noted, follow up with your child’s doctor and/or pharmacist. -<i>Deflazacort:</i> A similar drug to prednisone that is not yet available in the United States, but has been used to treat DMD in other areas of the world. Advantages and disadvantages in comparison to prednisone have not been fully explored.¹ <p><i>Calcium and Vitamin D</i> are often prescribed to counteract prednisone’s effect on decreasing bone density.¹</p> <p>Physical therapy: In the first years of your child’s life, physical therapy should focus on keeping joints mobile, preventing contractures, avoiding the development of scoliosis, and assisting independent walking.⁵</p> <p>Occupational therapy teaches your child how to interact with their environment. The occupational therapist (OT) will help your child with eating, playing, dressing, and other everyday activities.⁵</p> |

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| | Aquatic Therapy gives your child a chance to enjoy movement in the water is a great way to decrease stress to his body while improving or maintaining strength, flexibility, and overall fitness. ¹ |
| What else can I do? | Make sure your child is eating well and getting the nutrients he or she needs. Seek financial aid for help with the extra medical costs, if needed. Seek counseling for yourself, your child, and your family. Enjoy your child! |

For the **next stage of life, ages 3-12**, continue to refer to the above information. This chart includes additional information for your growing child.

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| What will my child do for school? | <p>Going to school is important for emotional and social development of your child. Your child has a right to attend public school and receive the same education as every other child. If he/she does have a disability that will affect his or her ability to learn, they will most likely qualify for special education assistance.</p> <p>A few important terms should be understood before entering your local school system.</p> <p>Least Restrictive Environment (LRE): It is required by law that every student be placed in his or her least restrictive environment. The general education classroom is defined as the LRE, and from there a child's team (teachers, therapists, etc.) will determine the level at which this child will function optimally. This may be in a general education classroom with an aide, a separate classroom for some or all classes, and everything in between.</p> <p>Inclusion: The public schools have begun a trend known as inclusion, in which those with disabilities are no longer always separated from other children. Whether this is done in all classes or in select electives, it is meant to provide both a social and educational advantage for your child.</p> <p>Individualized Education Plan (IEP) The IEP will be specially designed to fit the needs of your child. It is through this forum that teachers, professionals, and parents can communicate with one another in order to create functional and meaningful goals for your child.⁶ As a parent, it is important that you are involved in these IEP meetings. Be sure to voice your concerns and hopes for your child. And be certain that you communicate with your child's team throughout the year, not only at the annual IEP meetings. You also have the right to disagree with the IEP team, so be sure to be your child's advocate throughout the process. ⁶</p> <p>More information about transitioning to school can be found on the following websites: National Down Syndrome Society US Department of Education</p> |
| Any new assistive devices to consider? | During this phase of life, your child will likely lose the ability to walk independently. This can be a difficult transition, but the following information can help you to |

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| | <p>make an informed decision.</p> <p>Knee-Ankle-Foot Orthoses (KAFOs) have been shown to prolong assisted walking and delay contractures, but often involve surgical intervention (Achilles tendon release) and physical therapy in addition to the brace. Often, a walker is needed to “relearn” to walk with this new brace.⁷</p> <p>Standing Frame: Should be used to increase the time your child spends upright. When your child begins to lose the ability to walk, it is still important that he is able to stand. Spending time in supported stance helps avoid excessive bone loss, increases circulation, decreases contractures, and decrease the risk of scoliosis.⁷</p> <p>Walker: A walker could potentially be utilized after independent walking is no longer possible, and before a wheelchair is necessary. This decision should be discussed with both your physician and physical therapist.</p> <p>Wheelchair: Most children will be using a wheelchair by age 12.¹ There are many features to consider before purchasing a wheelchair. Discuss this transition with your primary care physician as well as your physical therapist. A few things to consider are as follows:</p> <ul style="list-style-type: none"> -manual or power wheelchair? -seat comfort/adjustability -postural positioning -leg rests ⁵ <p>Transferring Aides:</p> <p><i>Transfer Board:</i> A device that, when used properly, will help ease the process of helping your child move from wheelchair to bed, bed to chair, etc.¹</p> <p><i>Hoist/Lift:</i> If it becomes too difficult to transfer your child, there are mechanical devices that make the process less demanding. Discuss this option with a therapist or doctor if you think it may be appropriate.⁵</p> |
| Any new related conditions? | <p>Cardiomyopathy:</p> <p>As your child’s muscle weakness continues, his heart (also a muscle) will continue to weaken as well. This should be monitored closely by a cardiologist. It is recommended that children get a cardiac evaluation every 2 years until age 10, after which point it should be every year.</p> <p>Be sure to look for signs of heart problems:</p> <ul style="list-style-type: none"> -fluid retention (swelling of limbs, abdomen, etc.) -Shortness of breath -Chest Pain -Bluing of nail beds or lips ¹ <p>Pulmonary Conditions:</p> <p>-It requires muscle function to inhale and exhale. With each breath we are</p> |

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| | <p>contracting our diaphragm, a large muscle. Decreasing function of this muscle can make it much more difficult to get the needed oxygen to our body. Some signs to look for are:</p> <ul style="list-style-type: none"> -headaches -mental dullness -difficulty staying awake -nightmares¹ <p>-This decreased function can also lead to an increased risk of lung infection. <i>Pneumonia and excessive coughing</i> can quickly become major problems. Be sure to address these issues with your medical team immediately.</p> <p>Conditions of the Spine:</p> <p><i>Scoliosis:</i> Scoliosis is a condition of increasing curvature of the spine (if looking at your child from behind, his spine will start to take on the shape of an “S”). The decreased muscle mass associated with DMD can often lead to this condition. It develops more often if the child is not on prednisone therapy.¹</p> <p><i>Kyphosis:</i> More commonly known as a “hunchback,” where part of the back is rounded, giving a “slouched” look. Commonly develops after consistent time spent in the wheelchair.⁵</p> <p><i>Lordosis:</i> Known as a “swayback,” this condition can appear as though the child is sticking out his stomach. The shoulders are back, with the belly more forward. This too, usually develops after consistent time spent in a wheelchair.⁵</p> <p>Fractures: Fractures become common in this age group. As they begin corticosteroids, which decrease bone density, there is an increased risk of fracture. This group is also at an increased fall risk, adding to the high levels of fractures seen in this age group.⁷</p> |
| <p>What therapies should we continue?</p> | <p>Your child should continue to receive therapies in school, if seen as appropriate and necessary by their teachers and related professionals. Continuing physical therapy can help your child continue to improve their motor skill and balance. Occupational therapists will be beneficial in developing skills for school such as writing, dressing, and eating in socially acceptable ways.</p> <p>It is important to continue aquatic therapy as well. This will give your child a chance to move and be active without the additional force of gravity.</p> <p>Drug therapy: You may need additional drugs to help treat the developing heart condition. The following help to decrease the work load on the heart:</p> <ul style="list-style-type: none"> -Angiotensin Converting Enzyme (ACE) Inhibitor -Beta Blockers¹ <p>**Be sure to talk to your cardiologists about all of the options available**</p> |

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| | <p>Respiratory Therapy: As breathing ability declines, the family can get a cough assist device or learn procedures to assist with coughing and keep the bronchial system free from secretions. A respiratory therapist or pulmonologist can be consulted for the needed information.</p> <p><i>Oxygen:</i> At some point it may be necessary to begin supplemental oxygen through a mask or nosepiece in order to increase the oxygen in the blood.¹</p> |
| <p>Are there any surgeries we need to consider?</p> | <p>Spine Surgery: If your child's scoliosis progresses to a point where the structure is interrupting the lungs or heart function, surgery might be necessary. This is an invasive surgery where rods are placed along the spine to help straighten the body. This is only done in severe cases, and an orthopedic surgeon should be consulted for further information.⁵</p> <p>Respiratory Concerns: As breathing declines, surgery may become necessary.</p> <p><i>Ventilator Support:</i> If supplemental oxygen and cough assistive devices are no longer enough to keep the body oxygenated, it may be necessary to switch to ventilation. This can require a tracheotomy (an opening in the trachea, which is the structure that connects your mouth to your lungs).¹</p> |

As my child reaches the **teenage years, ages 13-18**, is there anything new to consider?

-Refer to the above chart above for information regarding this age group. Because each child's symptoms progress at a varying rate, all information is included above. Some of these concerns will not be important to you and your family until the teenage years or beyond.

What about my child's **adult life (18 and beyond)**?

-Again, refer to the above information for the development of your son's disease.

1. Muscular Dystrophy Association. Facts about Duchenne's and Beckers Muscular Dystrophy. Available at: <http://www.mdausa.org/publications/fa-dmdbmd-what.html#diet>. Accessed on: April 5, 2011.
2. Kreis, R. Brain. Metabolite composition in relation to cognitive function and dystrophin mutations in boys with Duchenne muscular dystrophy. *NMR in Biomedicine*. 2011; 24: 253-262.
3. Tendon Release. MD Guidelines. Available at: <http://www.mdguidelines.com/tendon-release>. Accessed on: April 11, 2011.

4. Scott, O. Prevention of deformity in Duchenne muscular dystrophy: A prospective study of passive stretching and splintage. *Physiotherapy* 1981; 67: 177–180.
5. Ciafaloni, E., Moxley, R. Treatment Options for Duchenne Muscular Dystrophy. *Neurology*. 2008, 10: 86–93.
6. Thompson N, Robb SA. The orthotic management of gait pathology in Duchenne muscular dystrophy. *Physiotherapy*. 1995; 81: 459–460.
7. Stevens, P. Lower Limb Orthotic Management of Duchenne Muscular Dystrophy: A Literature Review. *Journal of Prosthetics and Orthotics*. 2006; 18: 111-119.