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A resource on Physical Medicine and Rehabilitation (PM&R) topics developed by the American Academy of Physical Medicine and Rehabilitation (AAPM&R)

Duchenne and Becker Muscular Dystrophy

Condition: Duchenne muscular dystrophy (DMD) is a genetic disease that causes progressive muscle weakness and damage. Becker muscular dystrophy (BMD) is the less severe, and less common, form of the disease.

Background: Signs of DMD generally appear before age six; BMD usually appears after eight years of age. Both conditions affect skeletal muscle and heart muscle. With DMD, muscle weakness worsens more rapidly, and, by adolescence, patients usually require a wheelchair. BMD is milder and worsens more slowly. Currently, there is no cure for either DMD or BMD, but there are treatments for patients with certain genetic mutations.

Risk Factors: DMD and BMD occur almost exclusively in boys because the abnormal genes occur on the X chromosome. Boys have one X chromosome (from their mother) and one Y chromosome (from their father). Girls have one X chromosome from each parent, so the normal X chromosome silences the X chromosome that carries the mutation. These girls do have a 50% chance of passing the abnormal X chromosome to her children. However, mutations can occur spontaneously on the X chromosome.

History and Symptoms: The disease may first show up in toddlers with abnormal walking or delayed milestones. Weakness in the legs usually appears first, causing more problems with walking and getting up from the floor. Over time, weakness will also affect the arms. As the disease progresses, it can cause damage to the heart muscle (cardiomyopathy), which can lead to irregular heartbeat, fatigue, and shortness of breath. Decreased muscle range of motion (contracture) and scoliosis (abnormal curvature of the spine) are also commonly seen. Patients typically have weakening of the muscles that control the lungs while breathing, leading to a decline in lung function.

Physical Exam: Initially, a patient's weakness may be subtle, with weakness in the neck and hip muscles. Patients often have large calves and walk on their toes. The physical medicine and rehabilitation (PM&R) physician will check for the Gower sign, where patients rise from the floor to standing by "walking" their hands up their thighs to help make up for weakness in the thighs. The physician will also check strength in all the muscles and look for areas where the muscles are tight.

Diagnostic Process: The PM&R physician may order a blood test to check levels of a muscle enzyme called creatine phosphokinase (CPK). High levels of CPK raise concern for DMD or BMD. Next, the PM&R physician will order genetic testing to look for a mutation in the dystrophin gene, which causes DMD or BMD.

Rehab Management: The PM&R physician helps these patients maintain strength and range of motion. It is important for boys with DMD/BMD to remain physically active, but intense exercise can cause muscle damage. The PM&R physician can provide guidance as to appropriate exercise, such as swimming or biking. The PM&R physician is also specially trained in bracing and splinting to help with stretching and stability. Oral corticosteroids are used in DMD to slow disease progression, extend the time that kids are able to walk and protect the heart and lungs. As the children age, the PM&R

physician will help support the child's function. This can include evaluation for power wheelchairs and other devices to support mobility and make it easier for caregivers to assist the child. PM&R physicians will also help to evaluate for assistive technology to help with communication and community access. The PM&R physician can assist with discussions regarding advanced medical interventions (e.g. tracheostomy and ventilators) in the late stages of the disease.

Other Resources for Patients and Families: The National Institutes of Health offers a listing of resources for patients with Duchenne or Becker muscular dystrophy and their families.

Frequently Asked Questions

What is PM&R?
Physical medicine and rehabilitation (PM&R), also known as physiatry, is a primary medical specialty that aims to enhance and restore functional ability and improve quality of life to those with injuries, physical impairments or disabilities affecting the brain, spinal cord, nerves, bones, joints, ligaments, muscles and tendons. PM&R physicians, known as physiatrists, evaluate and treat the whole body, maximize patients' independence in their daily life and are experts in designing comprehensive, patient-centered treatment plans to empower patients to achieve their goals. By taking the whole body into account, they can accurately pin-point problems, decrease pain, assist in recovery from devastating injuries and maximize overall outcomes and performance with non-surgical and peri-surgical options. To learn more, visit www.aapmr.org/aboutpmr.

What makes PM&R physicians unique?
PM&R physicians' training focuses not just on treating medical conditions, but on enhancing the patient's performance and quality of life in the context of those medical conditions. They focus not only on one part of the body, but instead on the development of a comprehensive program for putting the pieces of a person's life back together – medically, socially, emotionally and vocationally – after injury or disease. PM&R physicians make and manage medical diagnoses, design a treatment plan and prescribe the therapies that physical therapists or other allied therapists perform or that are carried out by the patients themselves. By providing an appropriate treatment plan, PM&R physicians help patients stay as active as possible at any age. Their broad medical expertise allows them to treat disabling conditions throughout a person's lifetime.

Why see a PM&R physician?
A PM&R physician will thoroughly assess your condition, needs, and expectations and rule out any serious medical illnesses to develop a treatment plan. By understanding your condition and goals, you and your PM&R physician can develop a treatment plan suited to your unique needs.

How do I find a PM&R physician near me?
Visit www.aapmr.org/findapmrphysician or contact your primary care physician for a referral.