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CASE STUDY

DUCHENNE MUSCULAR DYSTROPHY (DMD)

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ABSTRACT

Muscular dystrophies are a group of diseases that make muscles weaker and less flexible over time. Duchenne muscular dystrophy (DMD) is the most common type. It's caused by flaws in the gene that controls how the body keeps muscles healthy. The disease almost always affects boys, and symptoms usually begin early in childhood. Children with DMD have a hard time standing up, walking, and climbing stairs. Many eventually need wheelchairs to get around. They can also have heart and lung problems. Although there isn't a cure, the outlook for people with DMD is better than it has ever been. Years ago, children with the disease usually didn't live beyond their teens. Today, they live well into their 30s, and sometimes into their 40s and 50s. There are therapies that can ease symptoms, and researchers are looking for new ones, as well.[1,3]

INTRODUCTION

Incidence And Etioloy[3,4]

DMD affects about one in 5,000 males at birth. Estimated prevalence of duchnnebecker muscular dystrophy (DBMD)was 1 in every 7,250 males aged 5-24 years. It is the most common type of muscular dystrophy. The average life expectancy is 26; however, with excellent care, some may live into their 30yrs or 40yrs.

Etiology [6,7]

DMD is caused by a problem in one of the geneslocus Xp21. Genes contain the information in our body needs to make proteins, which carry out many different body functions.If have DMD, the gene that makes a protein called dystrophin is broken. This protein normally keeps muscles strong and protects them from injury.

Pathophysiology [8]

The distrophin gene is located on the short arm of the chromosome X that, when flawed (mutated), causes both duchnne muscular dystrophy.Genes contain codes, or recipes, for proteins, which are important biological components in all forms of life.DMD occurs because the mutated gene fails to produce virtually any functional dystrophin. (Individuals with Becker MD genetic mutations make dystrophin that is partially functional, which protects their muscles from degenerating as badly. Dystrophin is responsible for connecting the cytoskeleton of each muscle fiber to the underlying basal lamina (extracellular matrix), through a protein complex containing many subunits. The absence of dystrophin permits excess calcium to penetrate the sarcolemma (the cell membrane). Alterations in calcium and signalling pathways cause water to enter into the mitochondria, which then burst. In skeletal muscle dystrophy, mitochondrial dysfunction gives rise to an amplification of stress-induced cytosolic calcium signals and an amplification of stress-induced reactive-oxygen species production. In a complex cascading process that involves several pathways and is not clearly understood, increased

Signs And Symptoms [1,6]

The main symptom of DMD, a progressive neuromuscular disorder, is muscle weakness associated with muscle wasting with the being first affected, especially those of the hips, pelvic area, thighs, shoulders, and calves. Muscle weakness also occurs later, in the arms, neck, and other areas. Calves are often enlarged. Symptoms usually appear before age six and may appear in early infancy. Other physical symptoms are:

- Awkward manner of walking, stepping, or running – (patients tend to walk on their forefeet, because of an increased calf muscle tone. Also, toe walking is a compensatory adaptation to knee extensor weakness.)
- Frequent falls
- Fatigue
- Difficulty with motor skills (running,hopping, jumping)

- Lumbar hyperlordosis, possibly leading to shortening of the hip-flexor muscles.
- This has an effect on overall posture and a manner of walking, stepping, or running.
- Muscle contractures of Achilles tendon and hamstrings impair functionality because the muscle fibers shorten and fibrosis in connective tissue
- Progressive difficulty walking
- Muscle fiber deformities
- Pseudo hypertrophy (enlarging) of tongue and calf muscles. The muscle tissue is eventually replaced by fat and connective tissue, hence the term pseudo hypertrophy.
- Higher risk of neurobehavioral disorders (e.g., ADHD), learning disorders (dyslexia), and non-progressive weaknesses in specific cognitive skills (in particular short-term verbal memory), which are believed to be the result of absent or dysfunctional dystrophy in the brain.
- Eventual loss of ability to walk (usually by the age of 12)
- Skeletal deformities (including scoliosis in some cases)
- Trouble getting up from lying or sitting position[4]

Diagnostic Findings

Pre Natal Test

- Ultrasound scan at 16 weeks or more recently by free fetal DNA testing.
- Chorion villus sampling (CVS) can be done at 11–14 weeks, and has a 1% risk of miscarriage. Amniocentesis can be Prenatal tests

Physical Examination[8,9,10]

Rule out other conditions that can cause muscle weakness.

Blood tests

- Creatinekinase(CK)- A high CK level is a sign that your child could have DMD. an enzyme that your muscles release when they are damaged.
- Genetest:Change in the dystrophin gene that causes DMD. Girls in the family can get the test to see if they carry this gene.
- Muscle biopsy- Low levels of dystrophin, the protein that is missing in people with DMD.

management

medical management:[10,11,12]

1. There's no cure for DMD, but there are medicines and other therapies that can ease your child's symptoms, protecct his muscles, and keep his heart and lungs healthy.
2. eteplirsen (exondys 51) - It is an injection medication that helps treat individuals with a specific mutation of the gene that leads to DMD. The most common side effects are balance problems and vomiting. Although the drug increases dystrophin production, which would predict improvement in muscle function.
3. Steroids- prednisone slow muscle damage. Children who take this medicine are able to walk for 2 to 5 years longer than they would without it. The drugs also can help the child's heart and lungs work better.
4. β_2 agonists increase muscle strength, but do not modify disease progression.
5. Mild, nonjarring physical activity such as swimming is encouraged. Inactivity (such as bed rest) can worsen the muscle disease.
6. Physical therapy is helpful to maintain muscle strength, flexibility, and function.
7. Orthopedic appliances (such as braces and wheelchairs) may improve mobility and the ability for self-care.

Nursing Management

It's overwhelming to learn that your child has DMD. Remember that the disease doesn't mean he can't go to school, play sports, and have fun with friends. If you stick with his treatment plan and know what works for your child, you can help him live an active life.

Stand And Walk As Much As Possible

Being upright will keep your child's bones strong and his spine straight. Braces or standing walkers can make it easier for him to stand and get around.

Diet

- There's no special diet for children with DMD, but healthy foods can prevent weight problems or help with constipation.
- Low-calorie, high-protein diet to avoid excessive weight gain (ambulation becomes more difficult if the child is overweight)
- Increase fiber and fluids in diet (to prevent constipation.)
- Stool softeners if needed.
- Work with a dietitian to make sure the child eats the right balance of nutrients and calories each day. Advice for need to see a specialist if child has trouble swallowing.

Stay active

- Encourage to remain ambulatory for as long as possible.
- Active and passive daily ROM exercise program.
- Splinting and bracing (to maintain lower extremity stability and avoid contractures)
- Exercise and stretches can keep your child's muscles and joints limber and help him feel better.
- A physical therapist can teach him how to exercise safely without overworking.

Family support

- Advice the how well the family adapts to the disorders.
- Give Other families living with DMD can be great resources for advice and understanding about life with the disease. Find a local support group or explore online discussion boards. It may also help you to talk about your feelings with a psychologist or counselor.

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