Muscular Dystrophy Canada

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Congenital Muscular Dystrophy

What is Congenital Muscular Dystrophy?

Congenital Muscular Dystrophy (CMD) is the name for a group of muscular dystrophies that are united by the fact that muscle weakness begins in infancy or in early childhood (typically before age 2). Sometimes, the condition is not detected until a child is found to have trouble with certain developmental milestones – such as learning to walk. Both boys and girls can develop CMD. There are several different types of CMD, which have different symptoms, degrees of severity, and rates of progression.

What are the symptoms of CMD?

With babies, the first symptoms include poor head control, slow movement, apparent lack of coordination, weak or floppy muscles (hypotonia), and possible hip dislocation. Other symptoms of CMD can include breathing problems or trouble swallowing.

Often, babies with CMD have contractures (a chronic shortening of muscles or tendons around joints) in their ankles, hips, knees and/or elbows. Due to contractures, some children have scoliosis or curvature of the spine.

Certain forms of CMD, such as Fukuyama CMD (seen almost exclusively in those of Japanese descent), Walker-Warburg Syndrome, and Muscle-Eye-Brain Disease include impaired intellectual development and vision problems.

How is CMD inherited?

The CMD is caused by genetic defects that affect important muscle proteins. Most forms of CMD are inherited in an autosomal recessive pattern, but at least one form appears to follow a dominant pattern of inheritance. Autosmal recessive disorders are inherited through a defective gene from both parents. If both parents are carriers there is a 25% chance that each child will inherit both defective genes.

People who are carriers have a defective gene but do not develop symptoms of the disease. This is because along with their defective gene they also have a healthy version of the gene. People with a family history of the condition may choose to undergo genetic counseling to better understand the risks of passing the gene on to their children.

How is CMD diagnosed?

There are several tests that may be used to confirm a diagnosis of CMD. A blood test can determine whether a muscle enzyme, creatine kinase (CK), is at high levels. This indicates muscle damage. An ultrasound study of the muscle shows whether there are abnormalities in the muscle. Electromyography,

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a test that measures electrical activity in the muscle, determines muscle health. A muscle biopsy may be performed to help identify a specific type of CMD. Here, a small amount of muscle tissue is removed and studied under a microscope. Family history, physical symptoms, and genetic testing are also helpful in making a diagnosis.

How does CMD progress?

CMD progresses differently in different people.The degree and progression of muscle weakness and degeneration varies with the type of disorder. While some children with CMD die in infancy, others will live into adulthood with only minor disability.

What treatments are available for CMD?

While there is currently no cure or treatment for any form of CMD, there are a variety of ways that help manage symptoms. Physiotherapy teaches specific ways to stretch the muscles to help reduce contractures. Exercises (and in rare cases surgery), may help improve or reduce contractures and slow the progression of scoliosis. For those with greater disability, braces and wheelchairs enhance independence. People who suffer from seizures can benefit from anti-seizure medications. Breathing problems can be helped by use of mechanical ventilation devices. Some people develop difficulty swallowing, and may benefit from the placement of a feeding tube to ensure good nutrition.

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