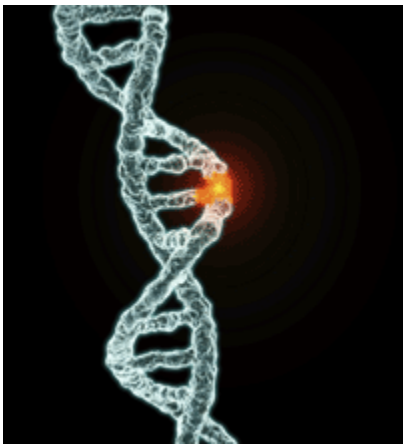
# Genetics in Special Education Series

## August 2011



## Genetic disorders presented in this issue:



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# Dercum Disease

### What is Dercum disease?

Dercum disease - also known as Adiposis Dolorosa, Anders' syndrome and Dercum-Vitaut syndrome - is a rare condition that is characterized by multiple, painful fatty lipomas (benign, fatty tumors) that occur chiefly in post-menopausal, obese women of middle age. However, although it is 20 times more common in women, 16 percent of the reported cases are males and it can also occur in people who are not obese

The lipomas are located primarily on the trunk region and on the extremities close to the trunk. Unlike ordinary lipomas, there is also pain that can be severe and sometimes debilitating. Dercum disease is a chronic condition, meaning that it is a long lasting condition. In addition, it tends to be progressive.

## What are the symptoms of Dercum disease?

This syndrome consists of four cardinal symptoms: (1) multiple, painful, fatty masses; (2) generalized obesity, usually in menopausal age; (3) weakness and fatigability; and (4) mental disturbances, including emotional instability, depression, epilepsy, confusion and dementia.

The pain can last for hours, can be paroxysmal (occurring only at certain times) or continuous, and worsens with movement. Dercum disease is often associated with generalized weakness, depression, and irritability. The condition can also be associated with early congestive heart failure, myxedema (a condition associated with severe hypothyroidism), joint pain, paroxysmal flushing episodes, tremors, cyanosis (bluish discoloration of the skin), hypertension (high blood pressure), headaches, and epistaxis (nosebleeds).

## What causes Dercum disease?

The understanding of the cause and mechanism of Dercum disease remains unknown. The origin of the pain is obscure, and the disease is better known as a description of its symptoms rather than as a physiologic or metabolic process. The fatty deposits (lipomas) cause nerve compression and result in weakness and pain.

Some cases reported in the literature have suggested possible causes for Dercum disease, such as the use of corticosteroids, a disturbance of endocrine function, or a genetic cause since it seems to run in some families. Some researchers have suggested that Dercum disease is an autoimmune disorder (a condition that occurs when the body's immune system attacks normal, healthy body tissue). However, no single cause has been pinpointed.

## What do we know about heredity and Dercum disease?

Dercum disease has been reported to occur in families, and in some of these cases it seems to be inherited as an autosomal dominant trait (this means that one out of a person's two copies of the gene, inherited from one parent, is altered). However, most reported cases of this condition are sporadic (the only case to occur in the family).

## What is the treatment for Dercum disease?

There is no "cure" for Dercum disease. At the present time, treatment for this condition is symptomatic, meaning that it focuses on one symptom at a time rather than the whole condition. Depending on the person, options can include weight reduction, surgery for the most painful lipomas, and medications to control pain. Liposuction has been used in some cases.

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# Hemochromatosis

## What do we know about hereditary hemochromatosis?

Myotonic dystrophy is an inherited type of muscular dystrophy that affects the muscles and other body systems. People who have myotonic dystrophy have muscle wasting and weakness in their lower legs, hands, neck and face that get worse over time. Signs and symptoms of myotonic dystrophy usually develop when a person is in his or her twenties or thirties. The severity of myotonic dystrophy varies widely among those who have it, even among family members.

The weakness and muscle wasting that occurs slowly progress to the point of disability. Usually, disability does not become severe until fifteen to twenty years after the symptoms appear. The progression of muscle weakness is slower and is less serious in people who are older when the muscle weakness is first noticed.

There are two types of myotonic dystrophy: Type 1 and Type 2. The two types are caused by alterations (mutations) in two different genes. The symptoms of Type 2 myotonic dystrophy are usually milder than those of Type 1. A severe type of Type 1 myotonic dystrophy can be seen at birth. This form of Type 1 is called congenital myotonic dystrophy. Congenital myotonic dystrophy has only been seen in Type 1 myotonic dystrophy and not in Type 2.

Myotonic dystrophy is the most common form of muscular dystrophy that begins in adulthood. It affects about 1 in 8,000 people worldwide. Type 1 myotonic dystrophy is the most common form in most countries. The commonness of the two types depends upon a person's ethnic background. For example, Type 2 myotonic dystrophy is as common as Type 1 in people who have German ancestry.

## What are the symptoms of myotonic dystrophy?

People who have myotonic dystrophy have progressive muscle wasting and weakness beginning in their 20's or 30's. The muscle wasting and weakness develop in their lower legs, hands, neck and face. They also have stiffness and tightness of their muscles (called myotonia), so they are slow to relax certain muscles after using them. Not being able to release their grip on in a handshake or a doorknob is one example of this problem.

In addition to muscle weakness and wasting, people who have myotonic dystrophy have clouding of the lens in their eyes (cataracts), and irregularities in the electrical control of their heartbeat (cardiac conduction defects).

Men who have myotonic dystrophy have changes in their hormones that can cause balding and sometimes the inability to father a child (infertility).

Babies who are born with signs and symptoms of myotonic dystrophy have congenital myotonic dystrophy. They have weakness of all their muscles, breathing problems, and developmental delays including mental retardation. Sometimes these medical conditions are so severe they may cause death.

Myotonic dystrophy is diagnosed by doing a physical exam. A physical exam can identify the typical pattern of muscle wasting and weakness and the presence of myotonia. A person with myotonic dystrophy may have a characteristic facial appearance of wasting and weakness of the jaw and neck muscles. Men may have frontal balding.

There are several laboratory tests that can be used to clarify the clinical diagnosis of myotonic dystrophy. One test, called electromyography (EMG), involves inserting a small needle into the muscle. The electrical activity of the muscle is studied and usually shows characteristic patterns of muscle electrical discharge.

The definitive test for myotonic dystrophy is a genetic test. For this test, a blood sample is taken to identify the altered gene (mutation) within the chromosomes which are contained within the white blood cells. Gene alterations in two genes - *CNBP* and *DMPK* - cause myotonic dystrophy. Myotonic dystrophy Type 1 is caused by a mutation in the *DMPK* gene. Type 2 myotonic dystrophy is caused by a mutation in the *CNBP* gene. Mutations in each of these genes involve a short segment of DNA that is abnormally repeated many times. This abnormal repetition forms an unstable region of the gene. These changes keep cells in the muscles and other body tissues from functioning normally, leading to signs and symptoms of myotonic dystrophy.

## What is the treatment for myotonic dystrophy?

There is currently no cure or specific treatment for myotonic dystrophy. Ankle supports and leg braces can help when muscle weakness gets worse. There are also medications that can lessen the myotonia. Other symptoms of myotonic dystrophy such as the heart problems, and eye problems (cataracts) can also be treated.

## Is myotonic dystrophy inherited?

Both Type 1 and Type 2 myotonic dystrophy are inherited in families in an autosomal dominant pattern. In autosomal dominant inheritance, having one copy of the altered (mutated) gene in each cell will cause the disorder. Usually a person who has myotonic dystrophy also has a one parent who has myotonic dystrophy.

In families that have myotonic dystrophy, the altered gene is passed down from one generation to the next. The disorder may begin earlier in life and signs and symptoms become more severe. This is called anticipation. In Type 1 myotonic dystrophy, anticipation happens because there is an increase in the length of the unstable region in the *DMPK* gene (expansion). The cause of anticipation seen in families who have Type 2 myotonic dystrophy is not yet known.

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