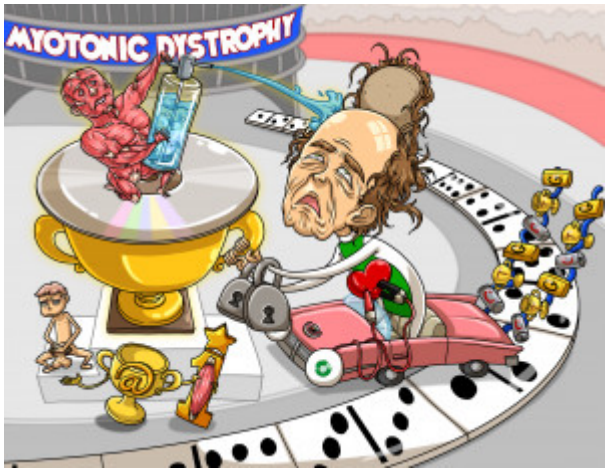
# Myotonic Dystrophy

Myotonic dystrophy is a chronically progressive multisystem disease, which is inherited in an autosomal dominant fashion. The genetic origin results from trinucleotide repeats of CTG. Myotonia, which is the continued involuntary contraction of a group of muscles, is the characteristic symptom in this disease. Patients often complain of muscle stiffness and have difficulty releasing their grip. Patients also commonly have cataracts, testicular atrophy, frontal balding and facial weakness. Lab findings can show conduction defects and muscle breakdown.




PLAY PICMONIC

## Pathophysiology

**Autosomal Dominant**
Domino
This disease is inherited in an autosomal dominant fashion.

**Trinucleotide Repeat**
DNA-strands with Repeats
Trinucleotide repeat inheritance results in the accumulation of CTG repeats, and worsening of the disease over generations.

**CTG**
Cans and Trophies with Gold
CTG is the trinucleotide repeat sequence. It is found on the DMPK gene, on the long arm of chromosome 19.

## Signs and Symptoms

**Facial Muscle Weakness**
Weak and Drooping Facial Muscles
Atrophy of the muscles of the face is characteristic of myotonic dystrophy, often leading to ptosis and a typical facial appearance.

**Frontal Balding**
Frontal Baldness
Frontal balding is an additional finding in many of these patients.

**Sustained Grip**
Locked Grip
Sustained grip is one of the classic findings in myotonic dystrophy, and is caused by conduction defects and subsequent inability to inhibit the increased tone of grip.

**Conduction Defects**
Conduction-cables to Heart
Conduction defects can be found in patients with the disease due to nerve damage.

**Cataracts**
Cadillac-cataracts
Cataracts are the clouding of the lens in the eye that obstruct the passage of light. They are present in almost every patient and may be detected early in the course by slit-lamp examination.

Sign up for FREE at picmonic.com

**Selective Atrophy of Type 1 Fibers**

@-trophy with (1) Wand

Histochemical stains have demonstrated selective atrophy of type 1 fibers in patients. Type 1 fibers are slow-twitch and oxidative fibers that contain large amounts of myoglobin and mitochondria with many blood capillaries, giving the muscle a red appearance.

**Testicular Atrophy**

Testicle @-trophy

Testicular atrophy is a characteristic finding in myotonic dystrophy.

Sign up for FREE at picmonic.com