

Myotonic Dystrophy Type I

Also known as *Dystrophia Myotonia (DMI)* and *Steinert's Disease*

A Beautiful Life Was Shortened By This Multi-System Disease. What Health Professionals Should Know:

Dentist: Beth's jaw would dislocate at every dental visit. This is a known symptom of myotonic dystrophy.

Otolaryngologist: Chewing and swallowing were both difficult. She also suffered from sleep apnea.

Ophthalmologist: Before she was 60, Beth had developed cataracts in both eyes that required surgery. She also had retinal disease and at the time of her death her vision was 20/300 in each eye. This led to vivid hallucinations.

Physical therapist: Physical therapists would try to tell her to walk upright. They never understood that her illness had caused damage to the muscles that would've allowed her to maintain proper posture. Her walking gait was always unusual. Her feet would land heel first and then the toe would slam down on the pavement bringing her motion to a nearly complete stop with every pace. This type of unusual gait is well known among those who study myotonic dystrophy.

Oncologist: Myotonic dystrophy increases the likelihood of cancer. Beth was diagnosed with invasive ductal carcinoma 2 years before her death. She was also mistakenly diagnosed with a very and cancer, which turned out to be several large cysts—another manifestation of myotonic dystrophy.

Pulmonologist: Many people with myotonic dystrophy ultimately die of respiratory failure. Beth had respiratory insufficiency for many years due to a weakened diaphragm and weaker intercostal muscles. Her cause of death was respiratory failure brought on by aspiration pneumonia (she couldn't cough well enough to keep stomach contents out of her lungs).

Anesthesiologist: Before Beth was diagnosed with myotonic dystrophy, she had considerable difficulty with surgery. They would need to give her oxygen afterward because she was having difficulty breathing. We now know that there are many precautions that an anesthesiologist must take, including their choice of anesthesia agent.

Genetic counselor: Myotonic dystrophy is an autosomal dominant disease. This means that a person who has the illness and pass it on to their child. There is a 50% chance of a child having the illness.

Orthopedist: Myotonic dystrophy can cause bone anomalies. Beth may have had some of these, although they were overshadowed by her numerous other symptoms.



In 1967, Beth was a contestant in the Imperial Valley Miss America pageant. (She won Miss Congeniality and 2nd runner-up.) Fortunately, she had no idea then what lay ahead. She was born with the genetic defect for myotonic dystrophy, an autosomal dominant hereditary illness that would eventually affect nearly every organ of her body.

Neurologist: Beth was initially diagnosed with ALS, she lived with that diagnosis for several months until a better qualified neurologist accurately diagnosed her with myotonic dystrophy. During the last year of her life, several neurologists were convinced that she had a form of progressive dementia. An autopsy showed this was not true - she had brain damage caused by hypoxia (inadequate oxygen reaching the brain).

Psychiatrist: Her psychiatrist believed she was suffering from schizophrenia because she was having vivid hallucinations and was unable to realize that they were not true. A more likely explanation is that the damage to her logic and thinking portions of the brain combined with almost total loss of vision probably resulted in the bizarre hallucinations and delusions

Internist: In addition to the obvious muscle weakness, especially hands, feet, diaphragm and face, myotonic dystrophy causes numerous problems internally, and Beth had them all. She was plagued with intestinal pains from an early age, she had a fatty liver, and polycystic kidney failure. The kidney failure may have been an isolated problem related to medication. Myotonic dystrophy also causes persistent daytime sleepiness that could be misdiagnosed as clinical depression.

Gynecologist/obstetrician: Despite going through Lamaze class to prepare for natural delivery, Beth suffered through many hours of nonproductive labor with our first daughter and eventually required a cesarean delivery. Myotonic dystrophy is known to weaken uterine contractions. In addition, had we known of Beth's condition, we would have needed to be alert for our daughter being born with congenital myotonic dystrophy—a condition that is frequently fatal to the newborn.

Nephrologist: Beth was being treated for chronic kidney disease for several years before her death. Her kidney function was only 1/5 the normal and she was soon going to need dialysis. An autopsy determined that she had polycystic kidney disease.

Cardiologist: Sudden death due to arrhythmia is common for myotonic dystrophy. Although Beth had annual EKGs which were nearly normal, her autopsy showed signs of hypoxia in the brain, probably brought on by irregular heart rhythms. Cardiologists who understand myotonic dystrophy now recommend a 24-hour Holter monitor since a standard EKG can miss a pattern of irregular heartbeats.

Endocrinologist: Beth's thyroid function was abnormal and she had a persistent goiter that at one point required aspiration. This is a known effect of myotonic dystrophy. It can also cause reduced fertility (both men and women) and early frontal balding.

Dermatologist: People with myotonic dystrophy can have numerous skin issues. Beth required minor surgery on 2 occasions for skin cancers.

My wife had myotonic dystrophy. It was the underlying cause of her early death. Although we knew of her condition for the past 7 years, we were unaware just how life-threatening it could be. Unfortunately, her doctors were also ill informed since myotonic dystrophy is not a common illness. I am presenting some information here in the hopes that it could be useful, and even life extending, for others who may have myotonic dystrophy.

Myotonic dystrophy is caused by a defect in a specific area of chromosome 19 called DMPK (dystrophia myotonica-protein kinase). It is normal for protein sequences to repeat a few times, however when a particular sequence in this region repeats more than 35 times, a person is said to be affected by myotonic dystrophy. This was discovered in the early 1990s and since then, suspected cases can be confirmed by a genetic test. Myotonic dystrophy is an inherited disease passed from parent to child with a 50% probability. It tends to become worse with each successive generation. Thus, if a parent had a mild form of the disease (35 - 100 repeats) his or her child could have a classic or even a congenital form with 100 - 1000 or more repeats. My wife had 212 repeats and was seriously affected.

For those with the classic form, there are many symptoms, including weakened muscles in the forearms and hands, calves and feet, shoulders back and face, and diaphragm. Those weaknesses can affect a person's ability to walk, keep their balance, perform fine motor skills with hands, and breathe. Men may experience infertility. Women with this form of myotonic dystrophy frequently have difficulty with childbirth.

Weakness in the facial muscles can cause a person's appearance to change rapidly. There can be loss of hair on the front of the scalp, drooping

eyelids and an open mouth. These weaknesses can also cause frequent jaw dislocation, and difficulty swallowing.

The most common causes of death for those with myotonic dystrophy are respiratory failure and cardiac arrest. Most articles you will read about myotonic dystrophy recommend yearly EKGs because there is the likelihood that cardiac arrhythmia may develop. What they don't say, is that there should also be a 24 hour Holter study done, as this is more likely to catch an arrhythmia that comes and goes. Thanks to having an autopsy performed following my wife's death, we now know that she had been having episodes of insufficient blood flow to her brain for some time that had not been detected by either the annual EKG or MRIs.

Important disclaimer: I am not a medical professional and this should not be considered medical advice. I have read a great deal about my wife's illness and have attempted to present the information in a more readable fashion here. I am providing you with the links to my sources. You should also know that I am only covering the mild and classic forms of type I myotonic dystrophy. There is also the congenital form which can affect infants at birth and type II myotonic dystrophy where a different gene is affected and the disease can be milder.

http://en.wikipedia.org/wiki/Myotonic_dystrophy
<http://medicine.yale.edu/neurology/divisions/neuromuscular/md.aspx>
<http://ghr.nlm.nih.gov/condition/myotonic-dystrophy>
http://mda.org/sites/default/files/In_Focus_MMD.pdf

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