“WHAT’S MY DISEASE?” CONTEST VIDEO TRANSCRIPT

Hello everyone,

Welcome to our contest. It’s called What’s My Disease?

Here are some clues to help you with the answer, and to help you determine whether my disease is hiding in your family. Please watch this entire video. The best clues to the answer are at the end.

My rare devastating disease is hiding undiagnosed in thousands of families around the world.

There are about 7,000 known rare genetic disorders. My disease is one of about 6% of them with treatment available.

My disease is usually passed from parents to children but it can be a new occurrence in a family.

My disease is invisible to most people. It is not obvious by looking at us.

There are many common symptoms of my disease but they can vary a great deal among individuals.

When symptoms begin to appear, a child may only have one or two symptoms. Some people only have one or two symptoms their entire lives. But, most people have many symptoms.

Many of the symptoms are common in other conditions which makes it hard to diagnose but a few of the symptoms are somewhat unique.

Common childhood symptoms include:

- pain or burning pain mostly in the hands and feet
- a skin rash of small reddish/purple spots called angiokeratoma that usually occur from your thighs to your belly button but can be found on other parts of the body
- a pattern in the eyes commonly referred to as corneal whorling and medically referred to as cornea verticillata. They can be seen in an eye exam but are usually overlooked.
- gastrointestinal issues such as frequent abdominal pain, diarrhea and constipation as well as nausea, vomiting, and bloating.
- reduced or inability to perspire and getting overheated easily. Many children and adults cannot tolerate heat or strenuous activity. School gym class and sports can be very difficult for some kids.

As kids get older through adolescence into adulthood, these symptoms usually progress and new symptoms appear. Other common symptoms include:

- unexplained protein in the urine known as proteinuria sometimes at a very young age
- periodic unexplained fevers

- frequent or chronic fatigue
- ringing in the ears known as tinnitus and progressive or sudden hearing loss
- lung disease sometimes diagnosed as asthma or COPD
- transient ischemic attacks or TIAs commonly referred to as mini-strokes at an early age
- unexplained left ventricular hypertrophy and cardiac conduction abnormalities. Pacemakers and defibrillators are fairly common at an early age.
- unexplained congestive heart failure, heart attacks, strokes, kidney failure and often premature death from those events at a relatively early age
- many people die from symptoms of my disease under the age of 50.

There is a laboratory test to confirm my disease called the GLA gene test.

My disease is misdiagnosed as many other conditions such lupus, multiple sclerosis, arthritis, irritable bowel syndrome, growing pains, various other disorders that cause chronic pain, and other conditions.

If you can guess the name of my disease you may win the prize. You may also recognize the symptoms of my disease in family members or friends. If you do, you may help to extend or save their lives.

Thousands of people like me need to be recognized and diagnosed to give them a chance at a better life and a better future.

Please review these symptoms carefully. This may be the most important contest you will ever enter. Please share this contest as widely as possible. Everyone should have a chance to learn about my rare but treatable disease.

Key words that may help you search for the correct answer are cornea veticillata, angiokeratoma and the GLA gene laboratory test.

You may enter this contest as many times as you like through March 31st, 2015. After March 31st 2015, we will publish the name of my disease, the winner of the contest and how to get help on the www.thousandsLikeMe.org. Thank you and good luck!