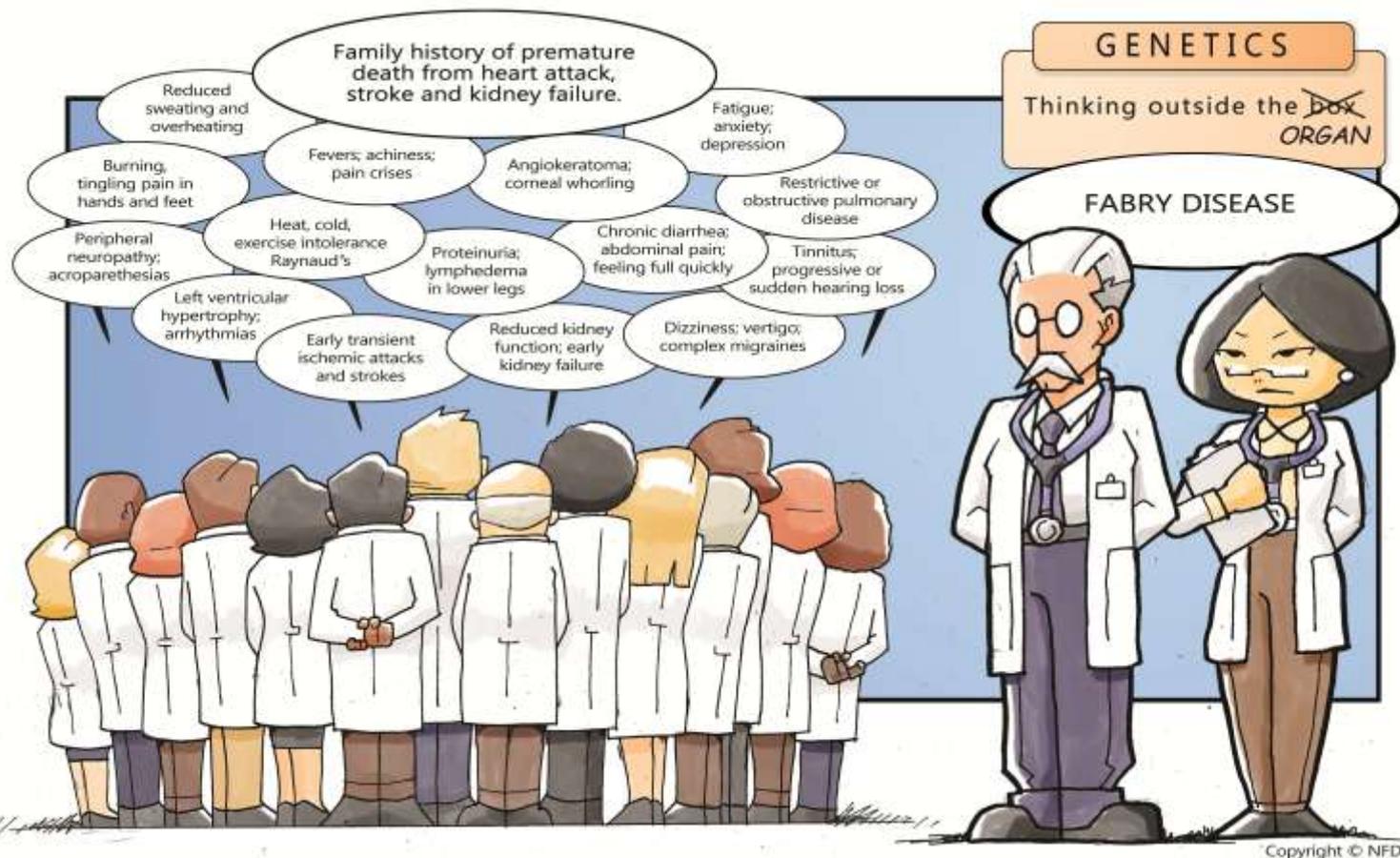




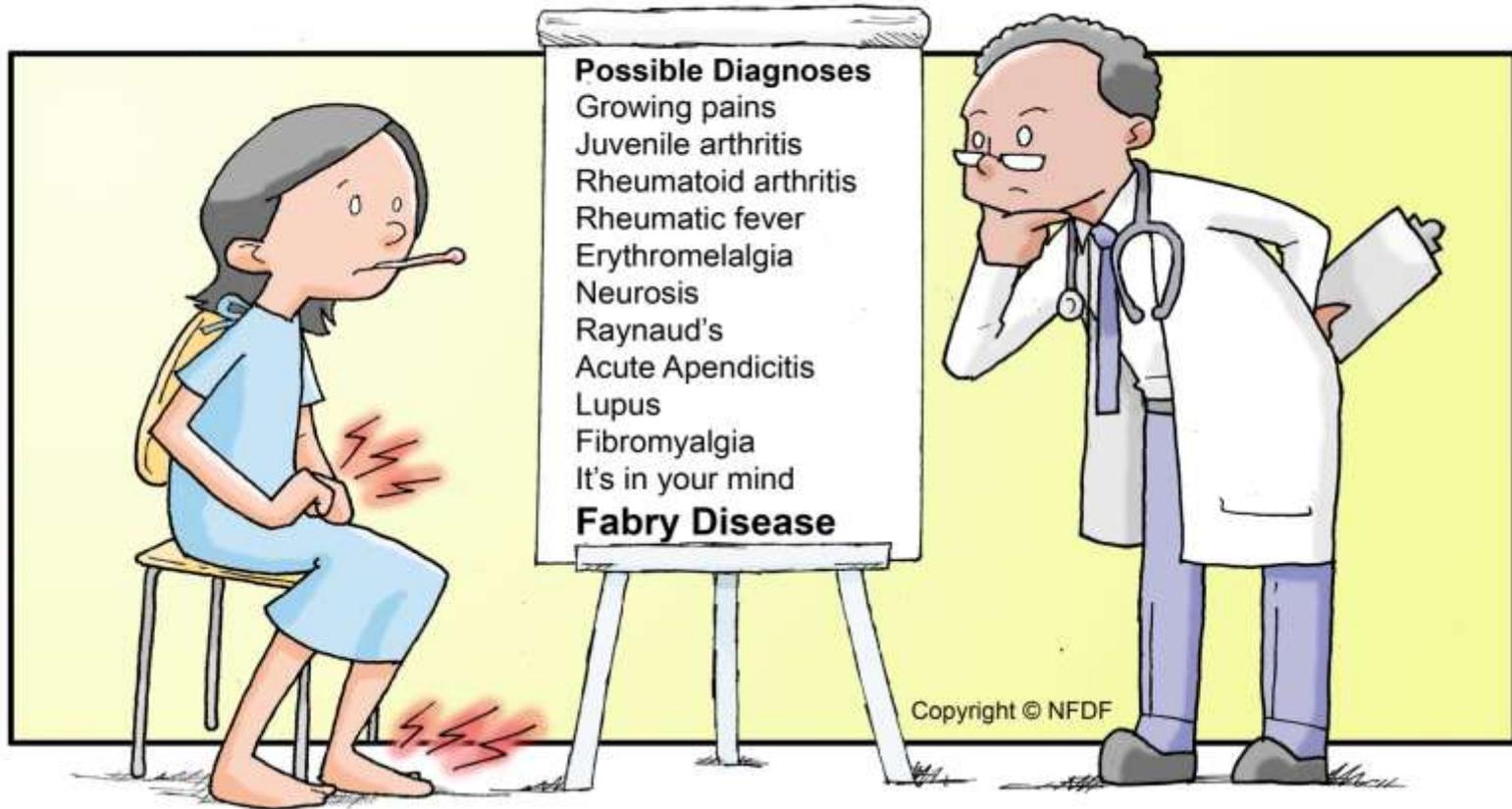
National  
**FABRY DISEASE** presents  
Foundation

## Understanding the many symptoms of Fabry disease



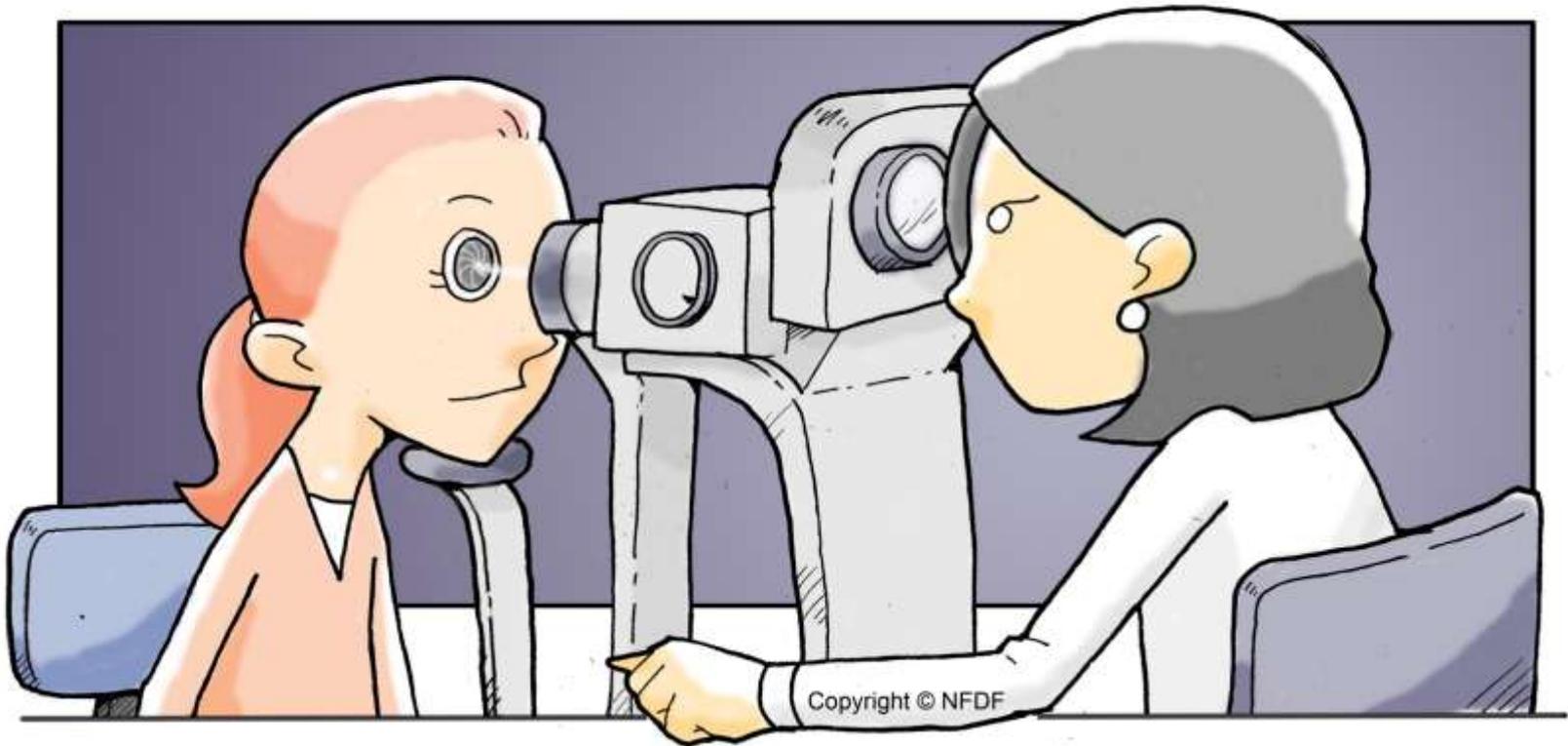
Fabry Disease is a rare, progressive, destructive and life-threatening multi-system genetic disorder that is severely unrecognized and underdiagnosed. While there are many very common symptoms of Fabry disease, the manifestation, presentation, severity, and age of onset can vary significantly among individuals even within the same family. An individual with Fabry disease may have only one or all of these symptoms. Unlike most rare disorders, an FDA approved treatment is available but it is not getting to those who need it!

# Getting people to understand Fabry disease can be a very painful experience!



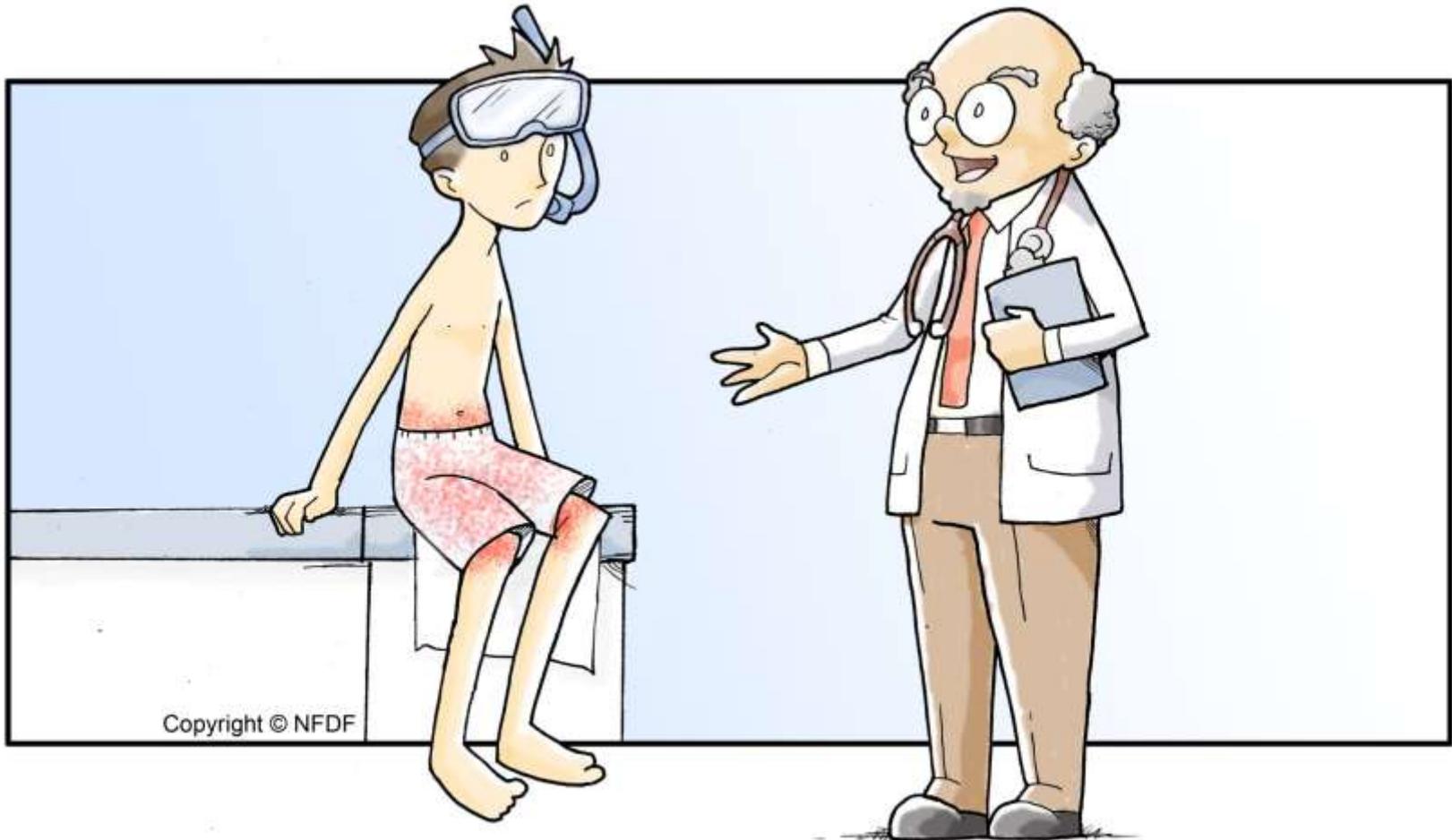
Usually beginning in childhood, burning or tingling pain or numbness in the hands and feet are common. Other pain symptoms include overall flu-like achiness, sharp shooting pains in the extremities, and/or extended pain episodes/crises. Symptoms are usually described as peripheral neuropathy and acroparesthesia. Unexplained fevers often occur. Fabry disease is frequently undiagnosed or misdiagnosed as other illnesses.

You can tell a lot about a person with Fabry disease by looking deep into their eyes!



Pale gray, brownish or yellowish streaks in the cornea of the eye (commonly referred to as corneal opacities or corneal whorls and technically referred to as corneal verticillata) are very common. They rarely, if ever, cause impaired vision. Corneal opacities can be seen during a routine slit lamp examination by an eye doctor. The medication Amiodarone can cause an eye finding similar to Fabry disease corneal whorling.

Connecting the rights dots can lead to a rare discovery!



Fabry angiokeratoma are small, sometimes clustered, red or reddish-purple skin lesions (dots) that often occur in the bathing trunk area but can be seen on most parts of the body. They usually start about ages 5 to 13, and normally progress in number and size with age.

People with Fabry disease can easily get hot, bothered and intolerant!



People with Fabry disease often have a reduced ability to perspire and overheat easily. Because of the resulting increased pain and fatigue, intolerance to heat, cold and strenuous physical activity are common.

Some people with Fabry disease are always in a rush!



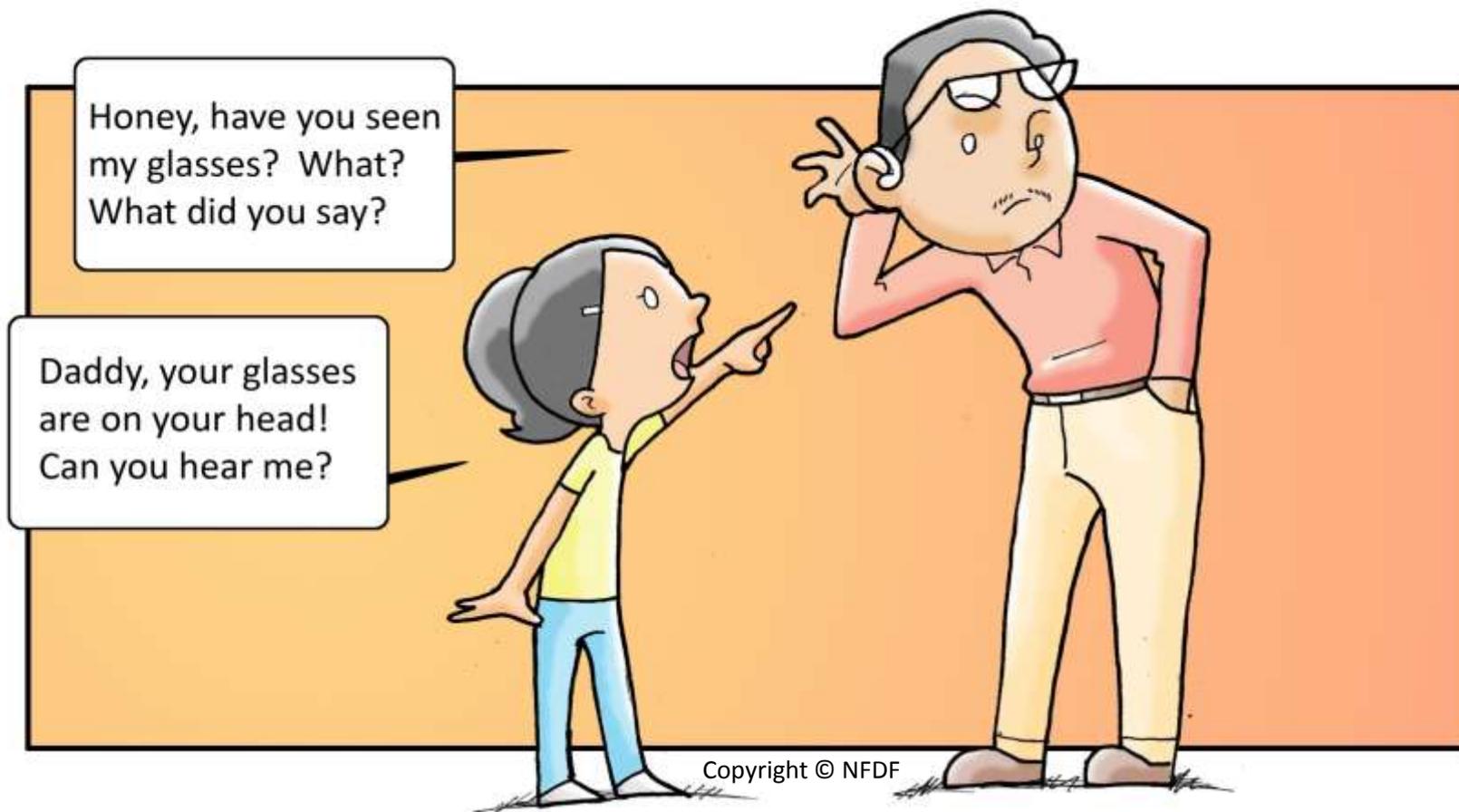
Chronic gastrointestinal (GI) upset is common. Chronic diarrhea, abdominal pain, and excessive gas are the most common symptoms but chronic constipation and vomiting also occur. GI upset is often misdiagnosed as irritable bowel syndrome or another GI disorder. Early satiety (feeling full quickly when eating) is also very common.

The burden of Fabry disease can be very stressful!



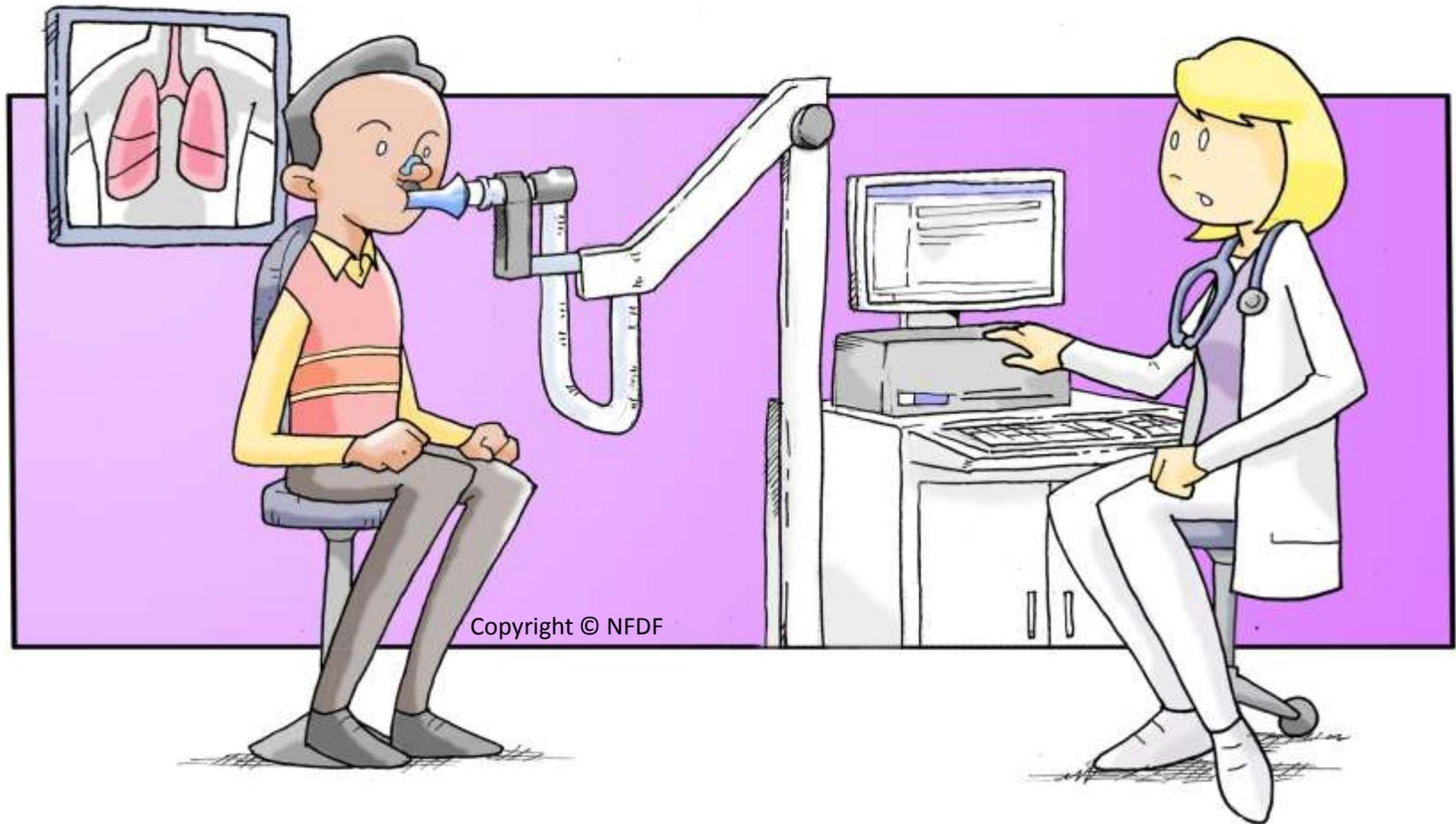
Anxiety, depression, antisocial behavior, and school avoidance are common symptoms of Fabry disease. Free, confidential counseling is provided to the Fabry community 7 days a week/24 hours a day through the NFDF's Family Assistance Program at 800-648-9557.

## People with Fabry disease tend to lose things!



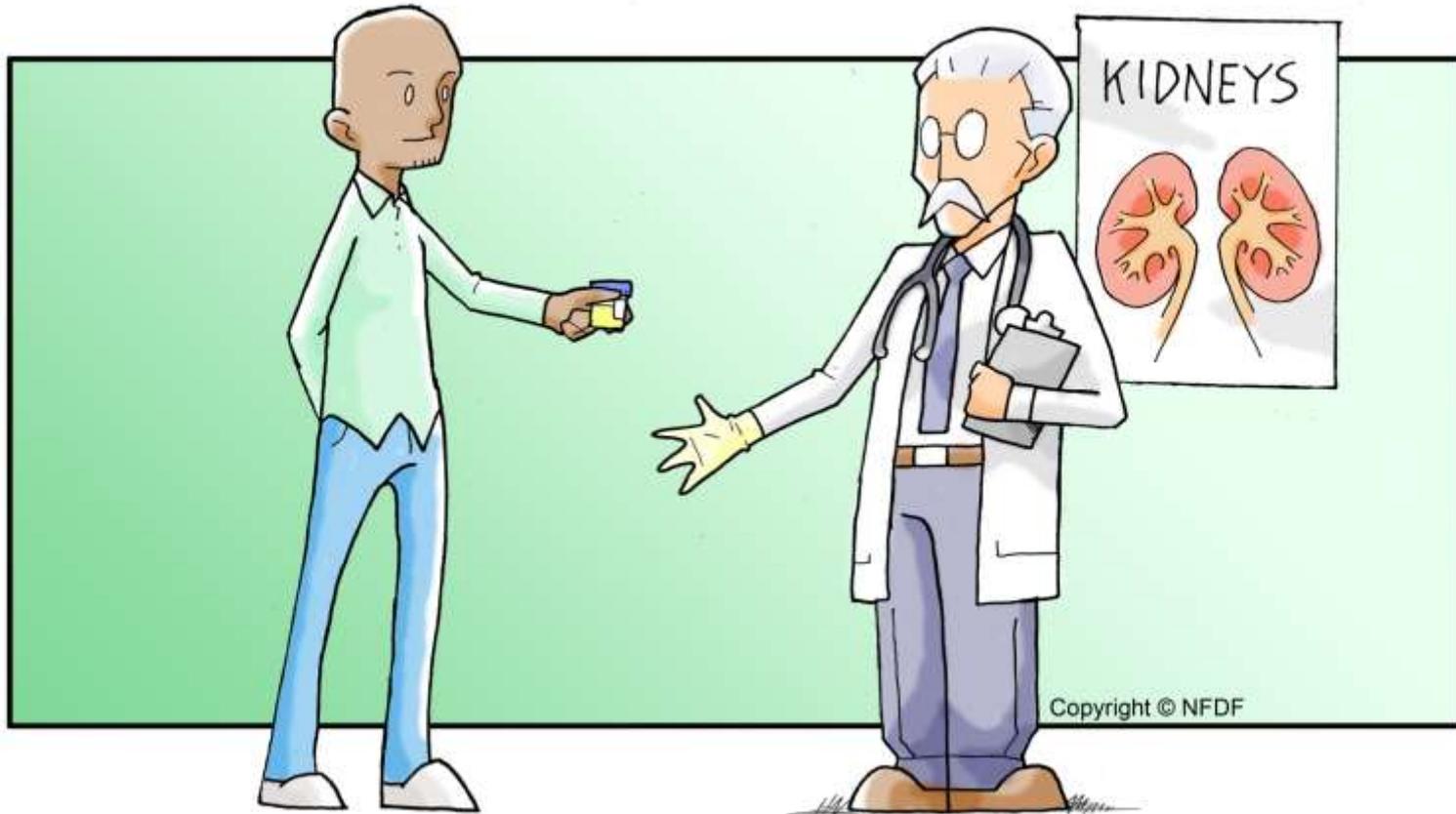
Common audiology symptoms include tinnitus and progressive or sudden hearing loss. Use of hearing aids is common and sometimes cochlear implants are necessary. Dizziness and vertigo (spinning dizziness) are also common symptoms of Fabry disease.

# Having Fabry disease can be a breathtaking experience!



Obstructive and constrictive lung diseases have both been documented in individuals with Fabry disease, often presenting as wheezing, dyspnea, or bronchitis. A diagnosis for Chronic Obstructive Pulmonary Disease (COPD) or Asthma is common.

People with Fabry disease can accumulate a lot of stuff  
in their lifetime!



Overt proteinuria (protein accumulating in the urine) and progressive Glomerular Filtration Rate (GFR) decline are important signs of Fabry neuropathy (nerve dysfunction). Kidney function may be reduced and progression often leads to dialysis and transplant. Parapelvic cysts and lymphedema especially in the lower legs are also common. Proteinuria, blood pressure and cholesterol should be aggressively managed

Many people with Fabry disease have a big heart and a courageous spirit!



Common cardiac symptoms include hypertrophic cardiomyopathy, left ventricular hypertrophy (LVH), conduction abnormalities, arrhythmias, and early heart attack/heart failure. Raynaud's syndrome secondary to Fabry disease is also common.

People with Fabry disease often have a lot on their mind!



Neurological symptoms such as complex migraines, white matter changes, transient ischemic attacks and strokes of unknown causes at a relatively early age are common with Fabry disease. Early neurological symptoms include but are not limited to chronic pain, gastrointestinal upset, and reduced sweating.

With hope and help, people with Fabry disease have a chance to live better and longer lives!



The statistical average age of death for an untreated male with Fabry disease is about 50 years old and about 10 years older for females. With treatment available, children should not have to live a diminished quality of life and adults should not have to die young from heart attacks, strokes and kidney failure. Widespread physician and family education is critical!



National  
**FABRY DISEASE**  
Foundation

National Fabry Disease Foundation  
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Even though a treatment for Fabry disease was approved by the FDA in 2003, 95% of the thousands of affected people in our country remained unrecognized and undiagnosed because of inadequate physician and family education programs to increase recognition of our disease. Our children should not have to suffer a diminished quality of life and our adults should not have to die young from premature heart attacks, strokes and kidney failure from Fabry disease.

Please support our education programs by making a donation online from the donation link in the upper right area of our homepage at [www.fabrydisease.org](http://www.fabrydisease.org) or send a check payable to "NFDF" to the above address.

**Thank you for giving people with Fabry disease an opportunity to live better and longer lives!**

For more information, please contact [jerry.walter@fabrydisease.org](mailto:jerry.walter@fabrydisease.org) or 1-800-651-9131.