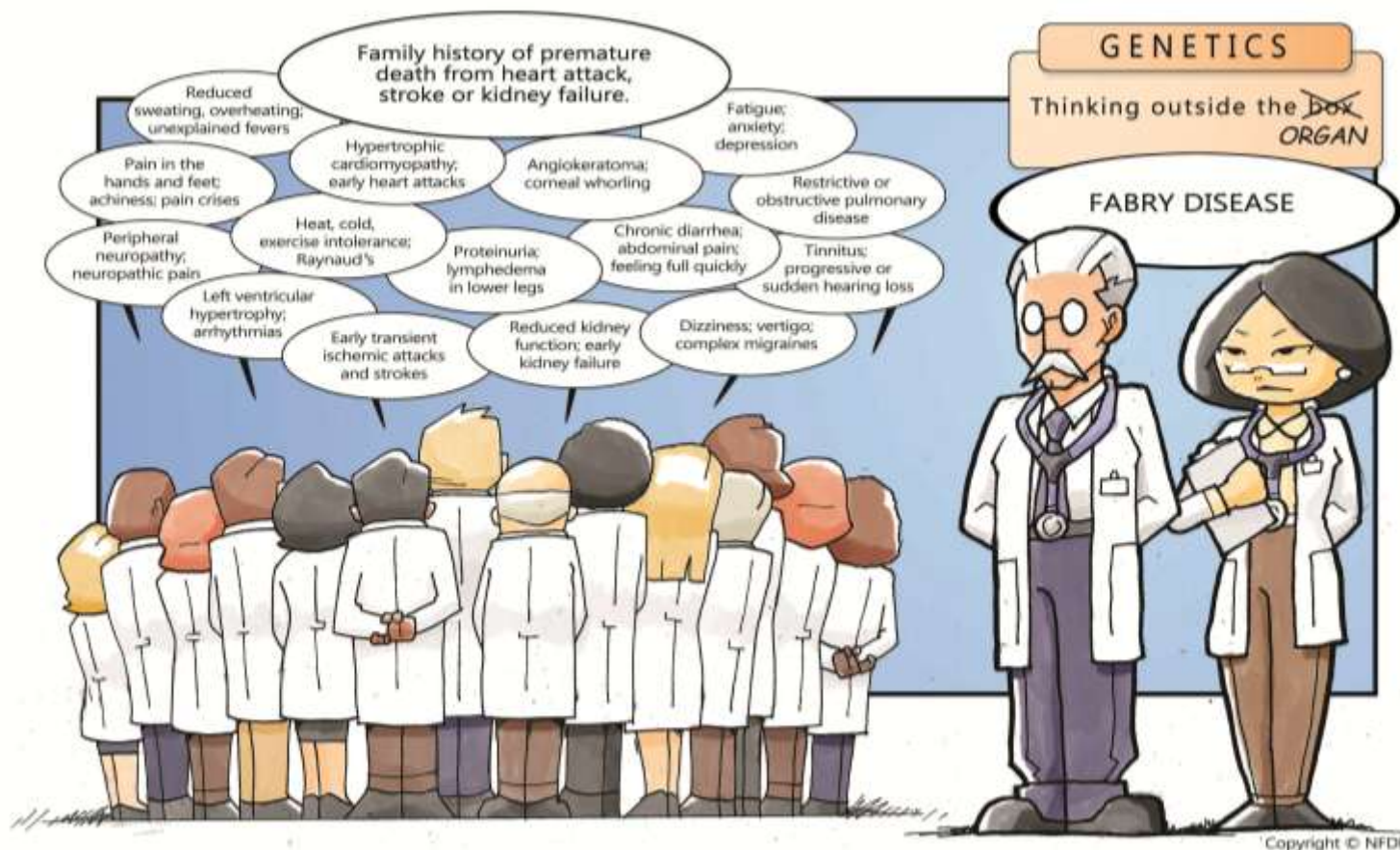




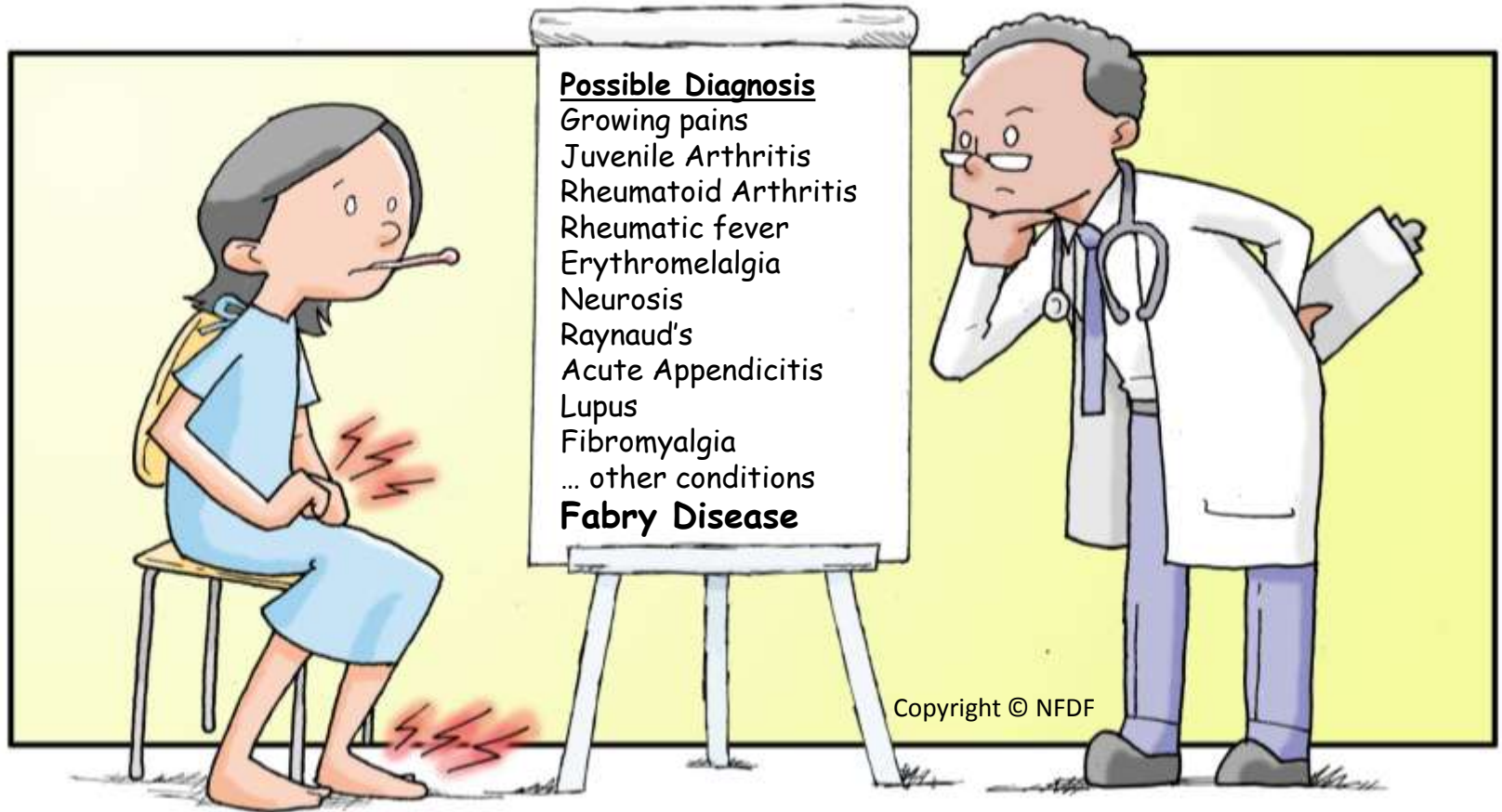
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Do you or does someone in your family have one or more of these symptoms?



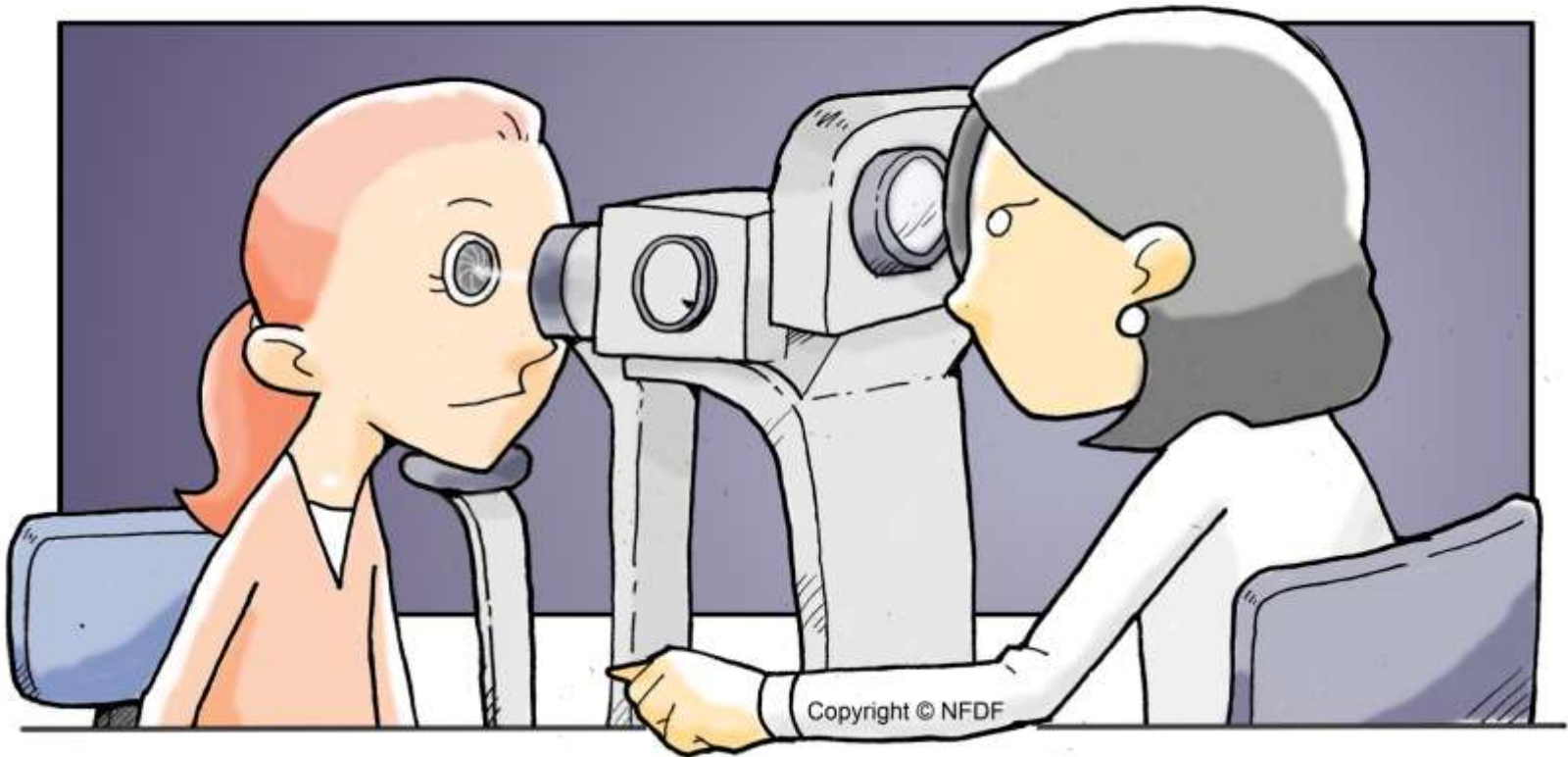
Undiagnosed Fabry Disease is hiding in thousands of families across America and around the world and it's treatable!

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



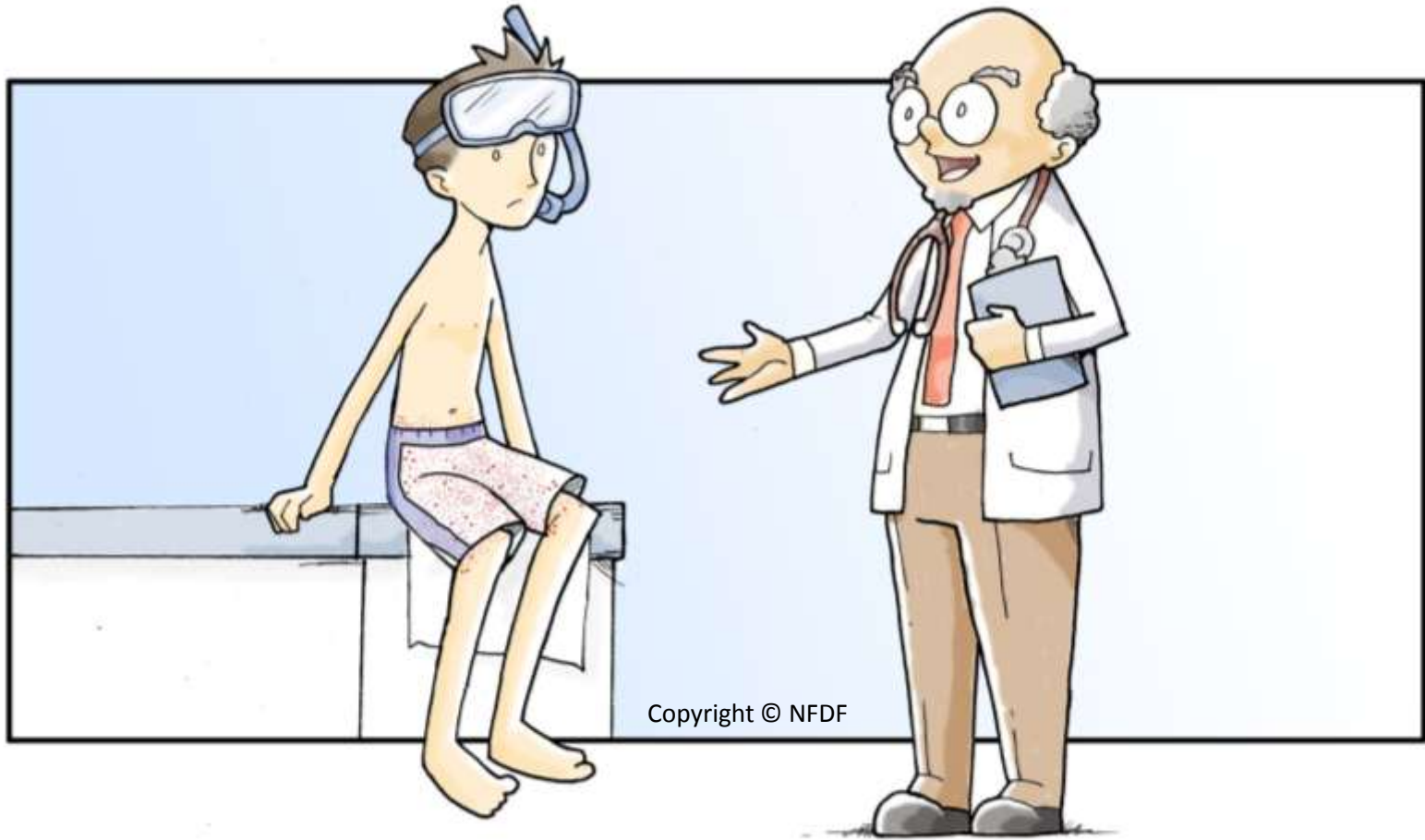
Usually beginning in childhood, burning or tingling pain or numbness in the hands and feet are common. Other pain symptoms include overall flu-like achiness, sharp shooting pains in the extremities, and/or extended pain episodes/crises. Symptoms are usually described as peripheral neuropathy and chronic neuropathic pain. 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 (commonly referred to as corneal opacities or whorls and technically named corneal verticillata) are very common. They occur in nearly all males and about 75% of females. They rarely cause impaired vision. Corneal opacities can be seen during a routine slit lamp examination by an eye doctor. Tortuous vessels and “Fabry cataracts” are also common. The medications Amiodarone and Chloroquine 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 increase in number and size with age. They are reported to be present in about 30% of children under 16 years old, about two-thirds of adult males, and more than one-third of adult females.

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



People with Fabry disease often have a reduced ability to perspire (hypohidrosis) and overheat easily. Hypohidrosis has been reported in about half of the men and about one quarter of the women with Fabry disease. Intolerance to heat, cold and strenuous physical activity are common and often result in increased pain and chronic fatigue.

Some people with Fabry disease are always in a rush!



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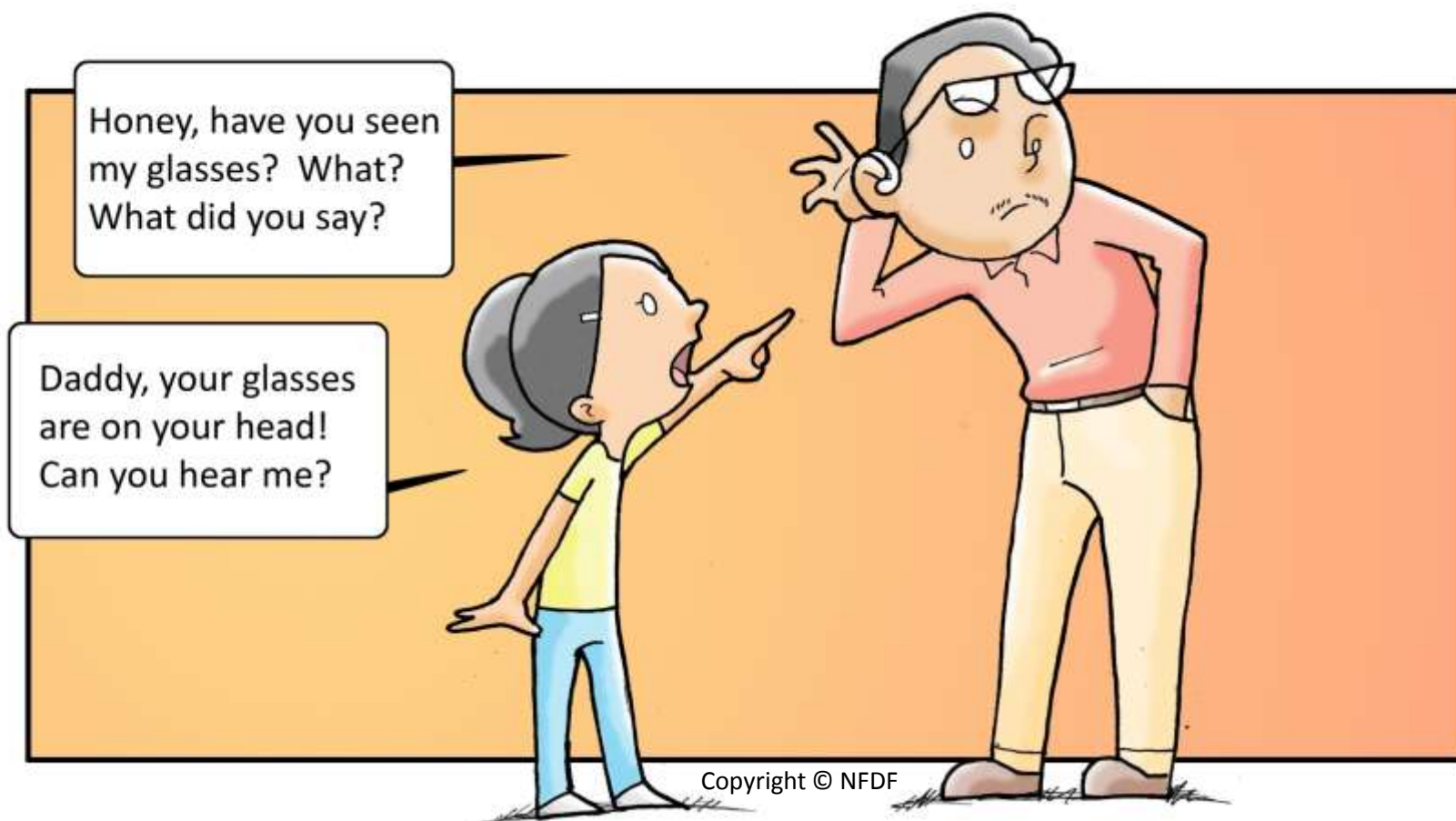
Chronic gastrointestinal (GI) upset is common in more than 50% of people with Fabry disease. Chronic diarrhea, abdominal pain, and excessive gas are the most common symptoms but constipation, bloating, nausea, and vomiting also occur. Early satiety (feeling full quickly) is also very common. GI upset is often misdiagnosed as irritable bowel syndrome or other GI disorders.

The burden of Fabry disease can be very stressful!



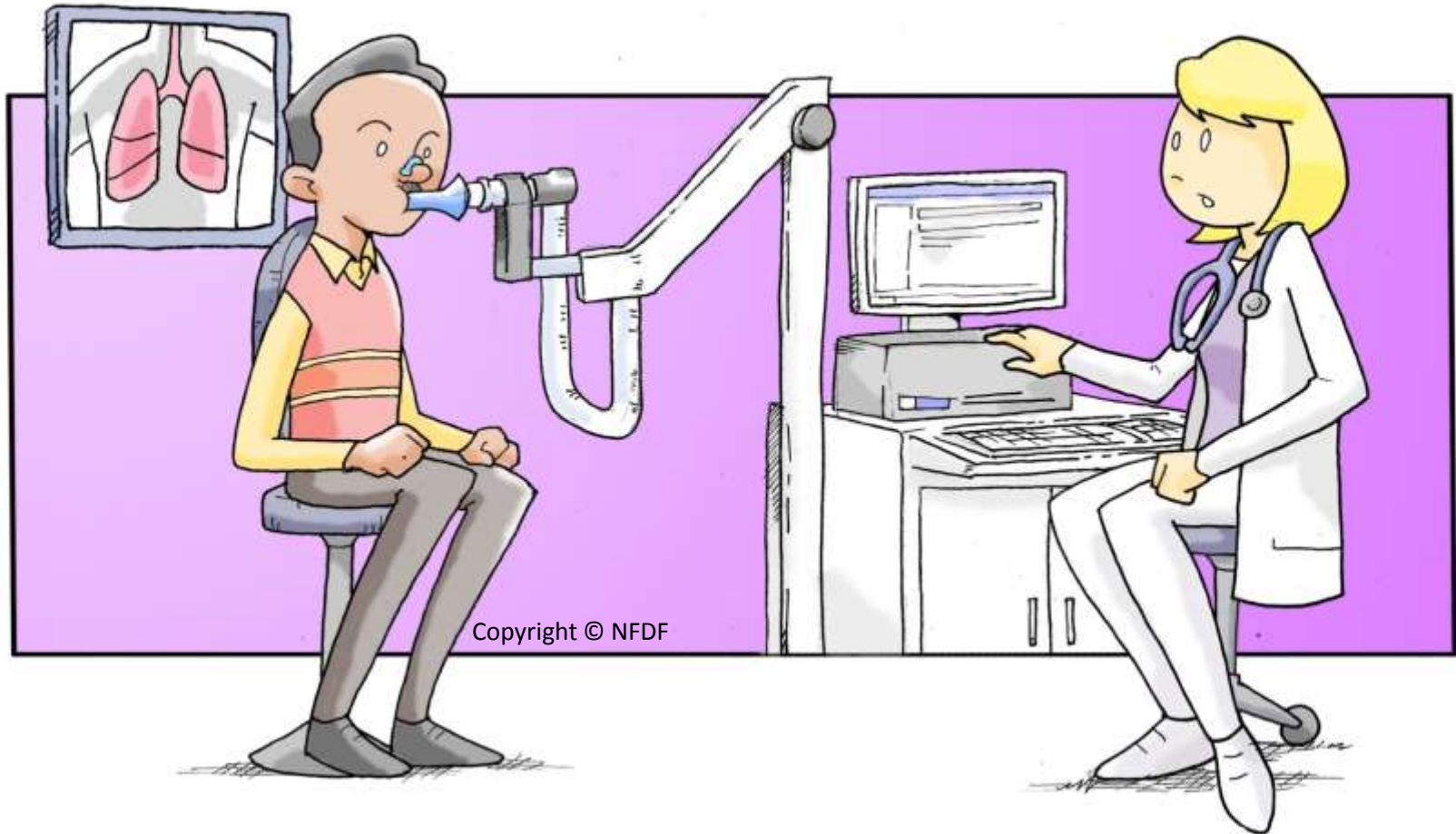
Anxiety, depression, antisocial behavior, and school avoidance are common symptoms of Fabry disease. Free, confidential counseling is provided to the Fabry community 7 days a week/24 hours a day through the NFDF's Family Assistance Program at 1-800-648-9557.

## People with Fabry disease tend to lose things!



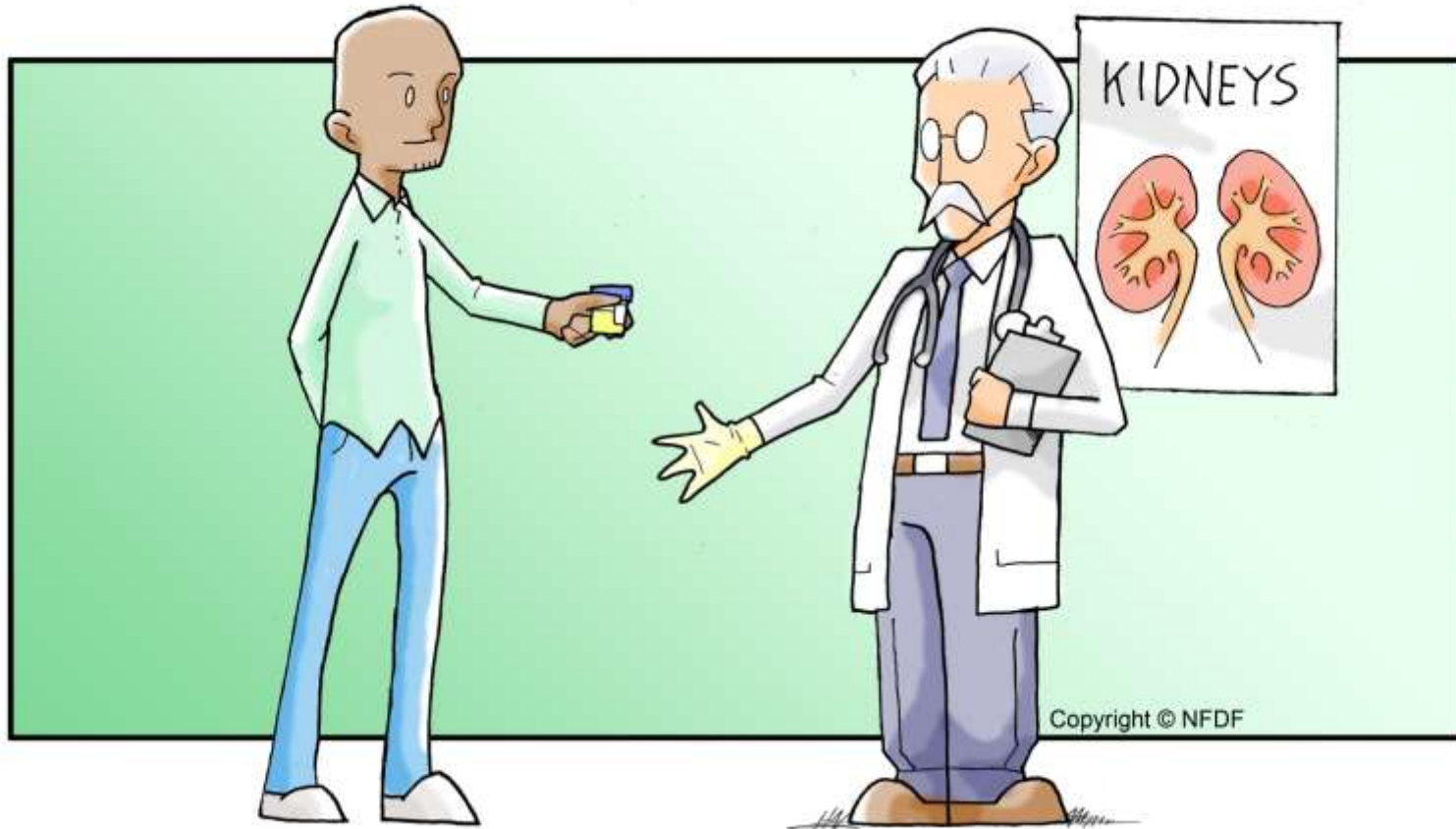
Common audiology symptoms include tinnitus and progressive or sudden hearing loss. Acute hearing loss is about 60 times more prevalent than in the general population. The onset of progressive hearing loss usually begins in men and women in the 2<sup>nd</sup> and 4<sup>th</sup> decades of life respectively. Use of hearing aids is common. 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. Chronic cough and exercise intolerance are common. 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!



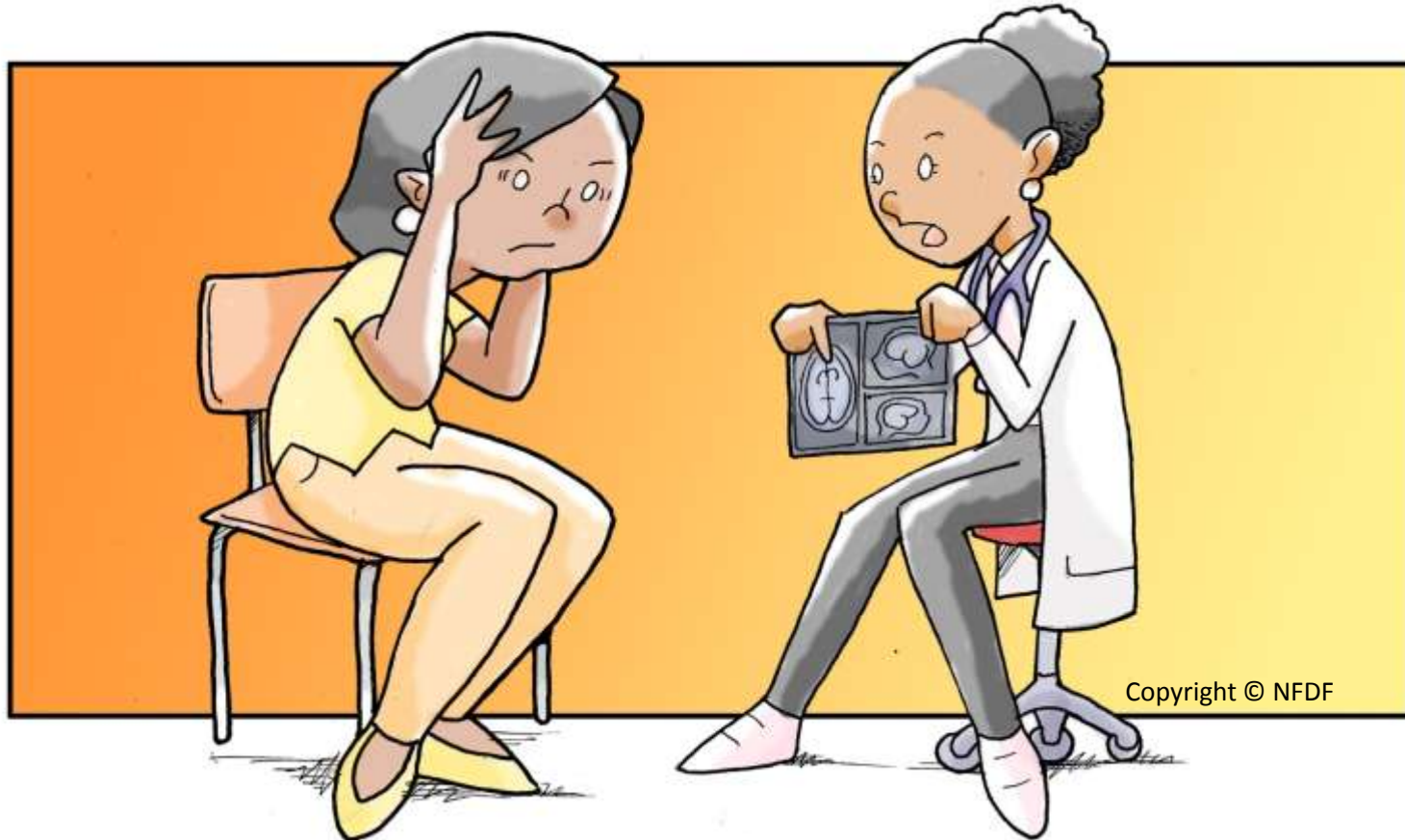
Overt proteinuria (protein accumulating in the urine) and progressive GFR decline are important signs of Fabry neuropathy. Proteinuria is usually the earliest sign of kidney involvement and is found in about 10% of children under 18 years old. It is found in about half of all males with Fabry disease by age 35. 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, palpitations, arrhythmias, and early heart attack/heart failure. One study reports that more than 50% of people with Fabry disease have cardiac symptoms at the age of 36. Raynaud's syndrome secondary to Fabry disease is also common.

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



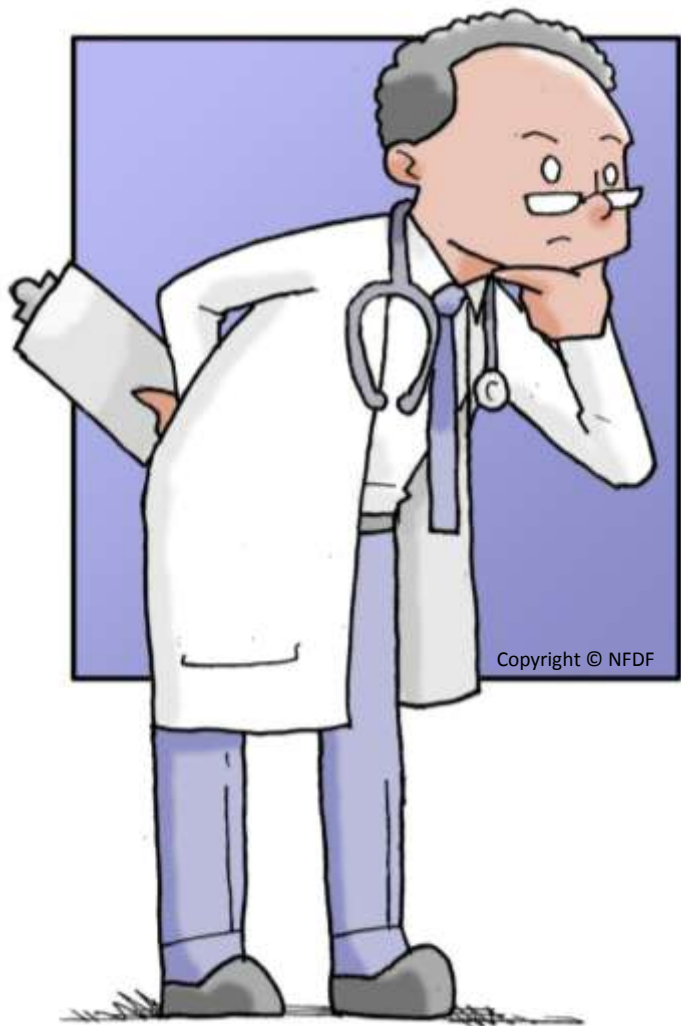
Neurological symptoms such as complex migraines, white matter changes, transient ischemic attacks and strokes of unknown causes at a relatively early age are common with Fabry disease. Almost 25% of people experience a cerebrovascular event. The events occur for men and women at the average ages of 34 and 54 respectively. 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 to 55 years old and about 10 to 15 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!

# Fabry Disease and The NFDF... The big picture



**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, but most people have many. Unlike most rare disorders, an FDA approved treatment is available but it is not getting to those who need it!

The National Fabry Disease Foundation's (NFDF's) vision is: ***"No longer will any individual's quality of life be diminished, nor will their lives be shortened because of Fabry disease."***

The NFDF is dedicated to providing support and assistance to people with Fabry disease and to providing education programs to increase recognition and diagnoses. Everyone with Fabry disease should have an opportunity to receive treatment and to live better and longer lives.

# Over 2600 peer-reviewed medical journal articles have been written about Fabry disease.

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Even though a treatment for Fabry disease was approved by the FDA in 2003, most of the thousands of affected people remain unrecognized and undiagnosed because of inadequate physician and family education programs. 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 and assistance programs by making a donation online from the donation link on our website homepage at [www.fabrydisease.org](http://www.fabrydisease.org) or send a check payable to “NFDF” to the above address.

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

This presentation was designed and prepared by Jerry Walter and was illustrated by Mike Johnson who both have Fabry disease.

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