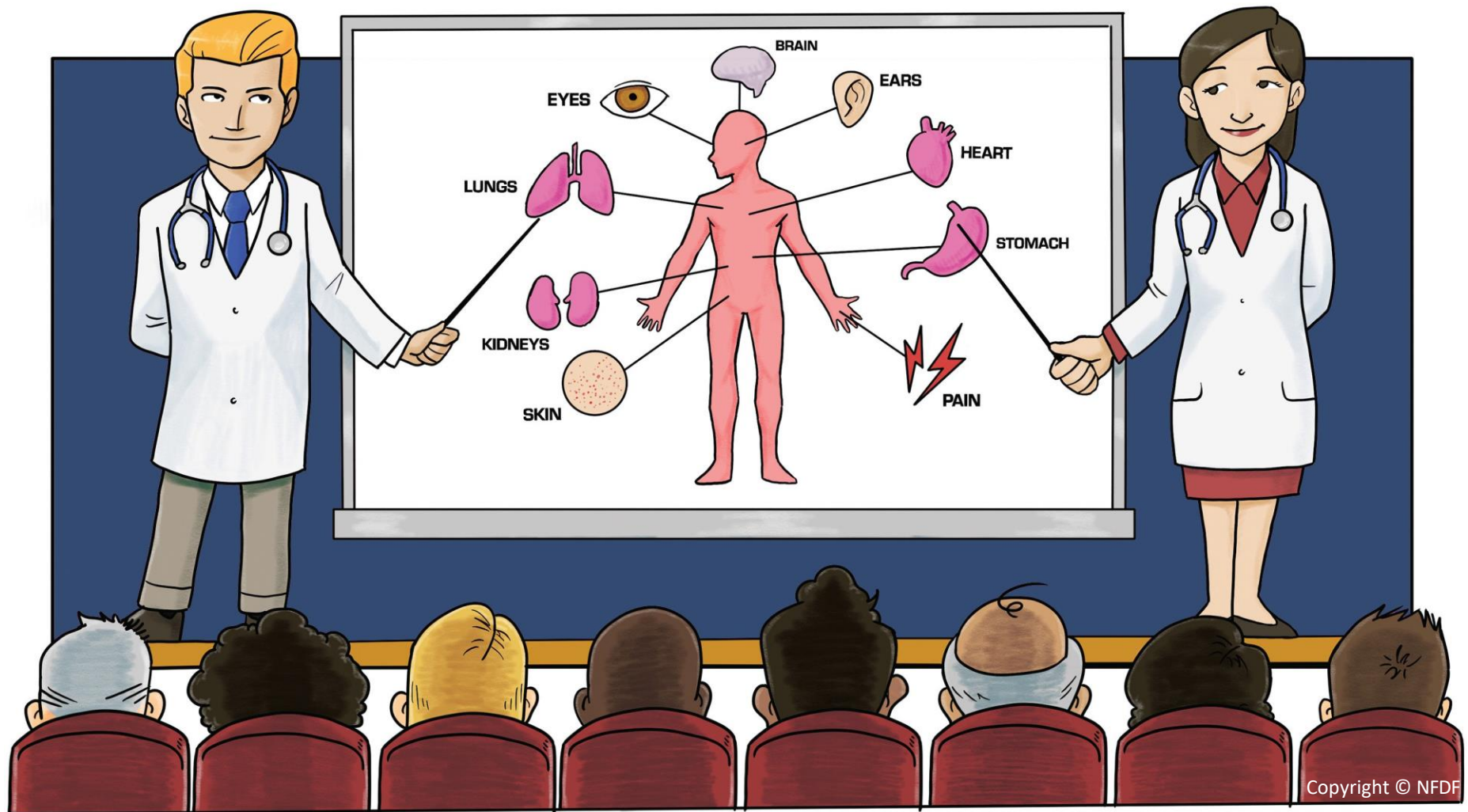




National  
**FABRY DISEASE**  
Foundation

## Understanding the many Symptoms of Fabry Disease



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### Fabry Disease Symptoms

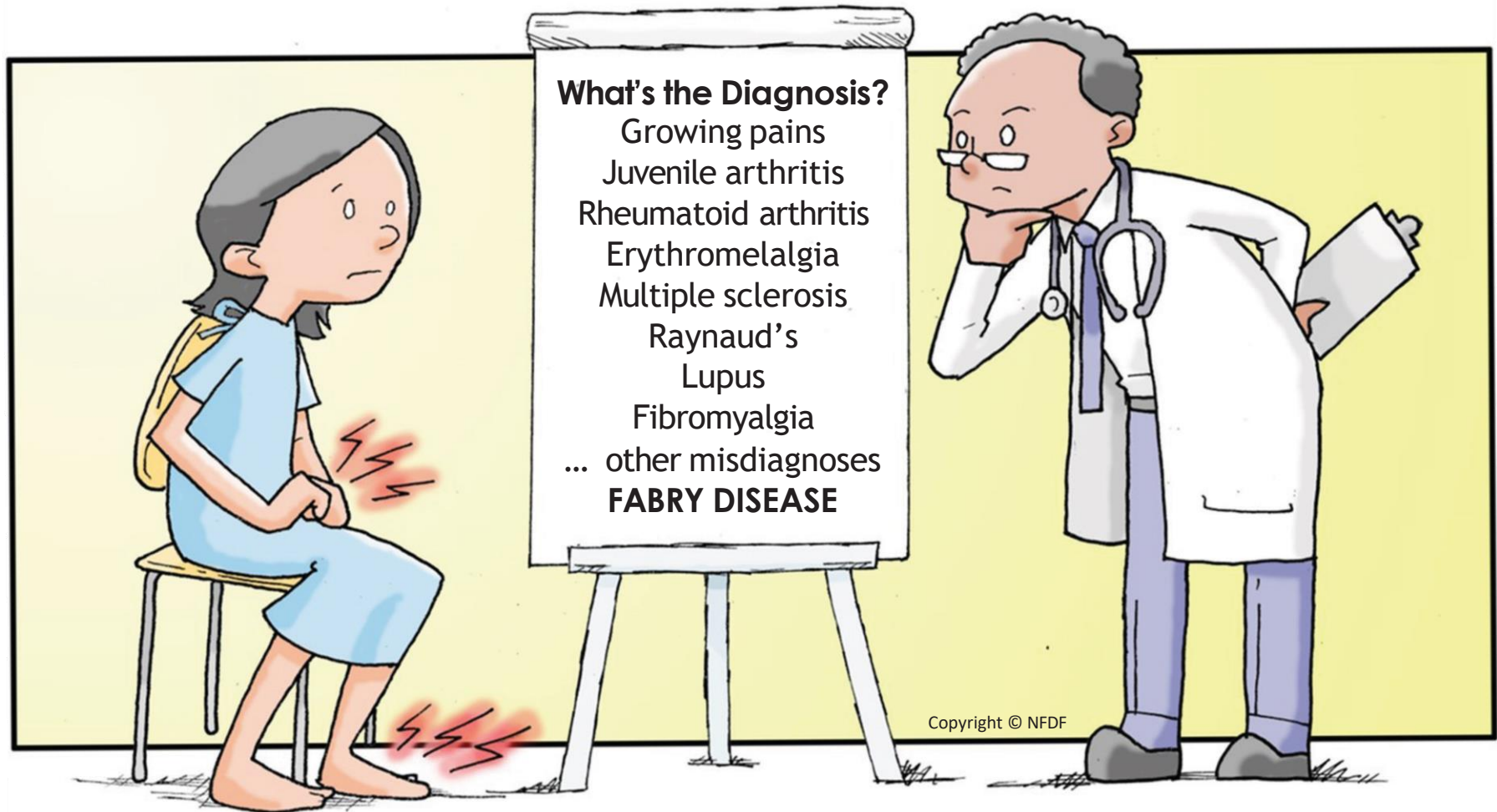
... these common symptoms can vary greatly among individuals.

People with Fabry disease undergo one test for a diagnosis and a life-time of tests for resilience.



The genetic test to diagnose Fabry disease is called the *GLA* gene test. In males, an enzyme analysis is diagnostic for Fabry disease. It can be used to confirm whether a male has Fabry disease or not. In females, an enzyme analysis is not sufficient. Females with a Fabry *GLA* gene variant may have normal enzyme levels. For this reason, DNA analysis of the *GLA* gene must be performed for females.

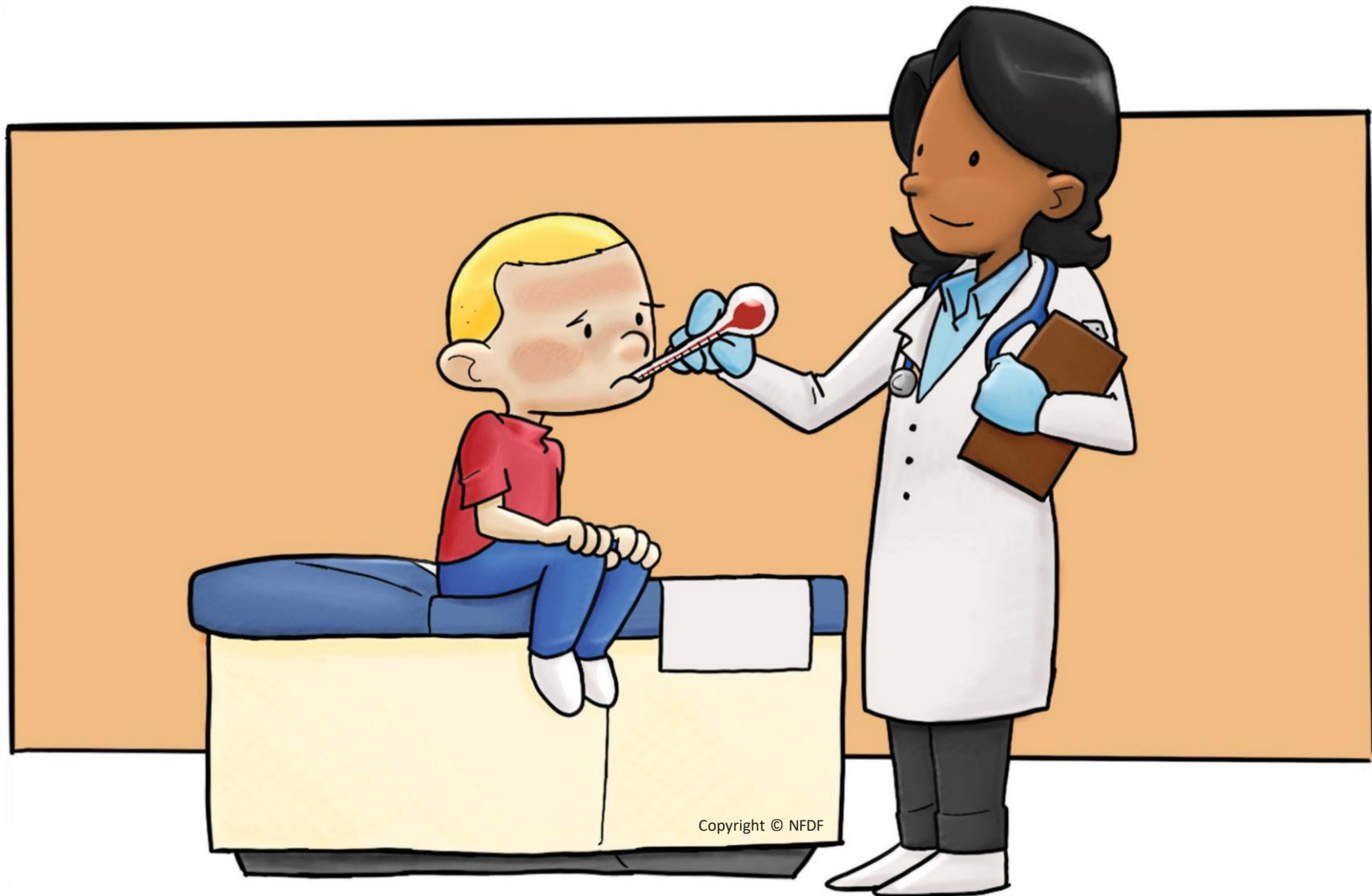
# Getting people to understand Fabry disease can be a very painful experience!



Small fiber peripheral neuropathy is a painful part of Fabry disease. Beginning in childhood, burning and tingling pain in the hands and feet is very common. Other types of pain include chronic overall achiness; burning pain in other areas of the body; sharp, shooting pain in the extremities; and extended pain episodes/crises. Fabry pain is often misdiagnosed as other conditions.

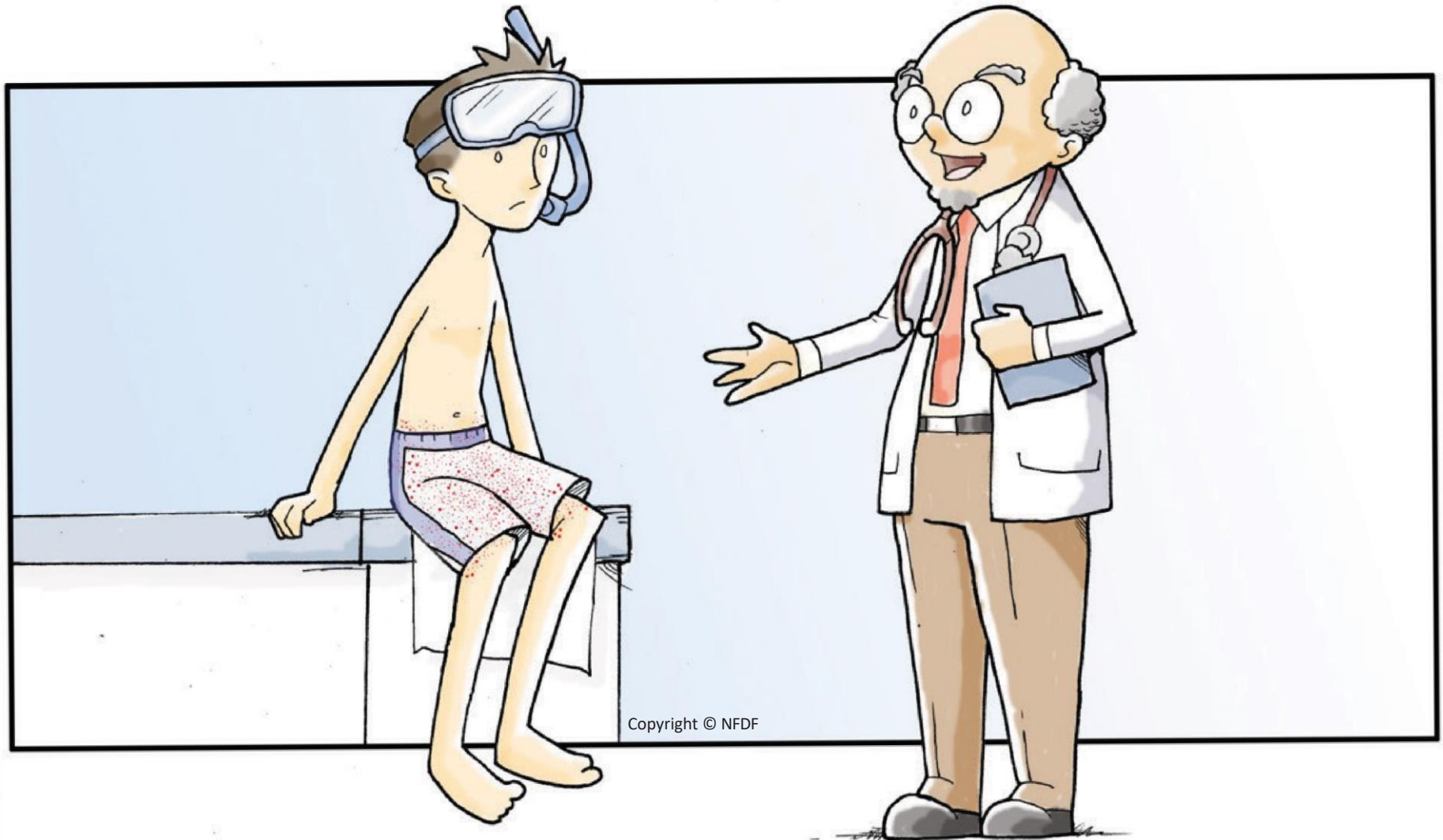


Some people work feverishly to manage their symptoms of Fabry disease.



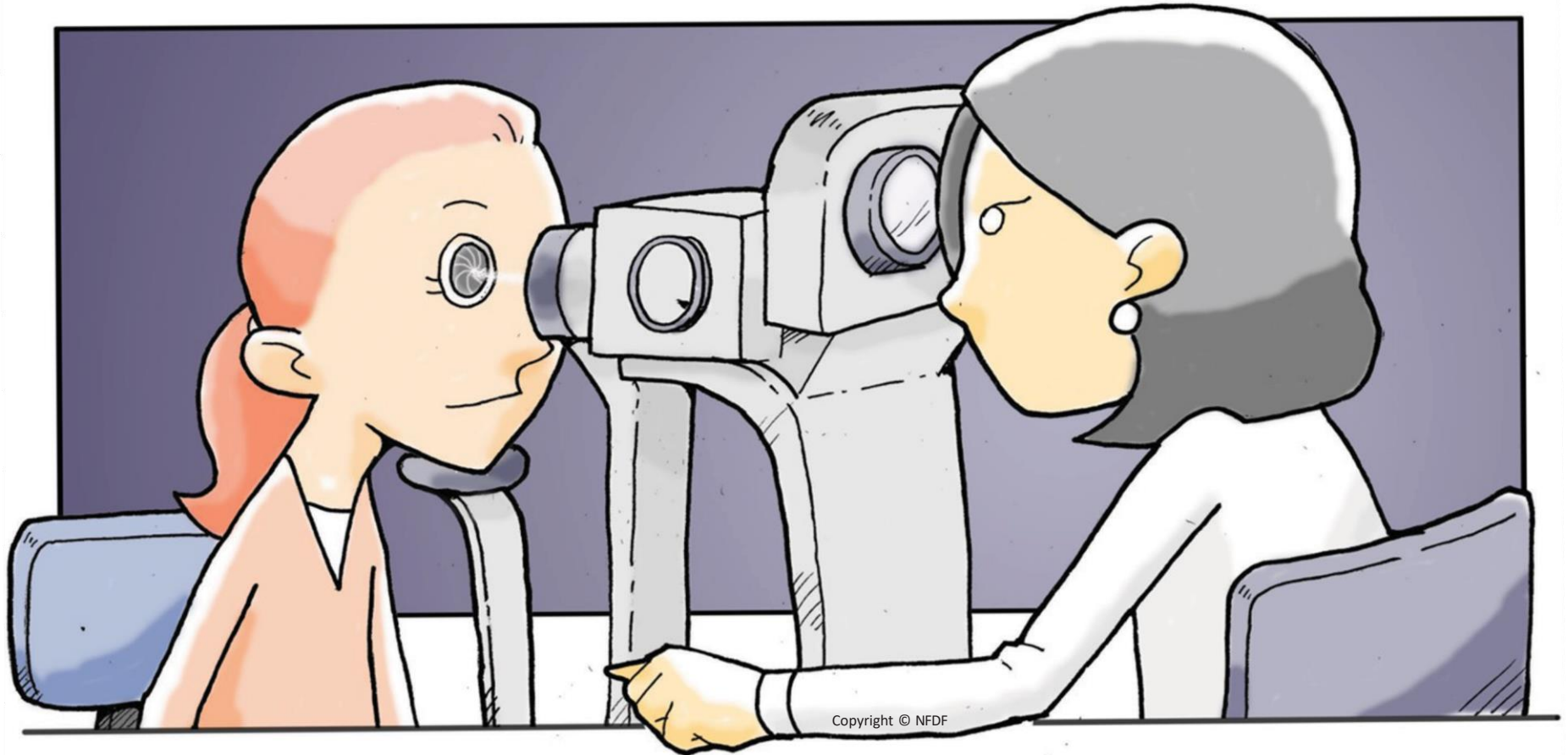
Especially in childhood and adolescence, but also throughout other stages of life, many individuals with Fabry disease experience episodes of unexplained high fever. Recognizing recurrent fever of unknown origin as a symptom of Fabry disease could lead to an early diagnosis.

Connecting the right dots can lead to a rare discovery.



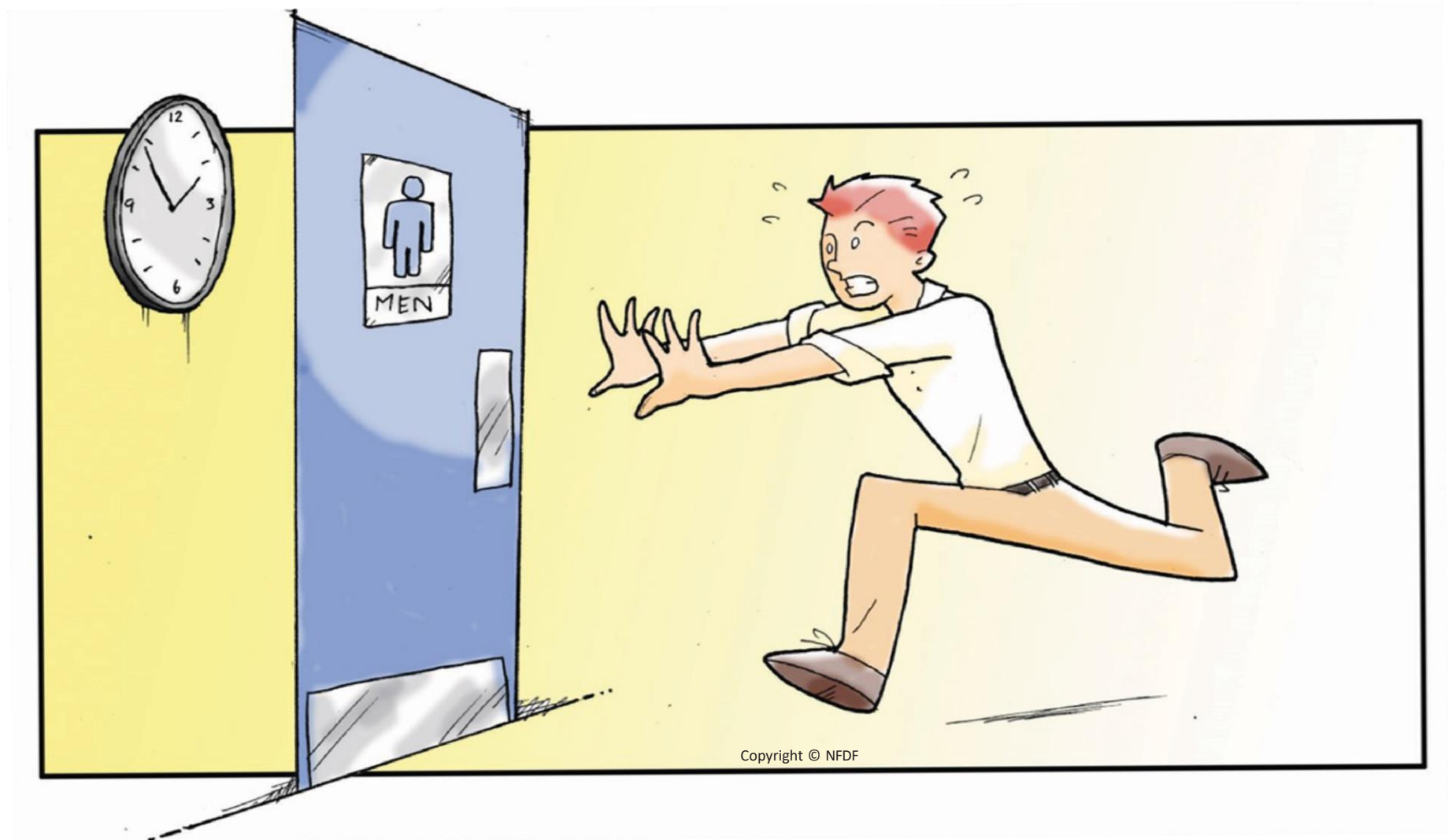
Fabry angiokeratoma are small, sometimes clustered, red or reddish-purple skin lesions that usually occur in the bathing trunk area but can be seen on other parts of the body. They usually appear by ages 5 to 13 in boys and increase in number and size with age. Reports indicate they are present in about 30% of kids under 16 years old, about 2/3 of adult males, and more than 1/3 of adult females.

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



The most common eye finding in Fabry disease is a whorling pattern called cornea verticillata or corneal whorling. Cornea verticillata does not typically impact vision, but because it can be seen with a slit lamp in a routine eye exam, these whorls are very important to help diagnose Fabry disease. People with Fabry disease can also develop Fabry cataracts, tortuous vessels, dry eyes, and conjunctival lymphangiectasia as well as other possible eye symptoms.

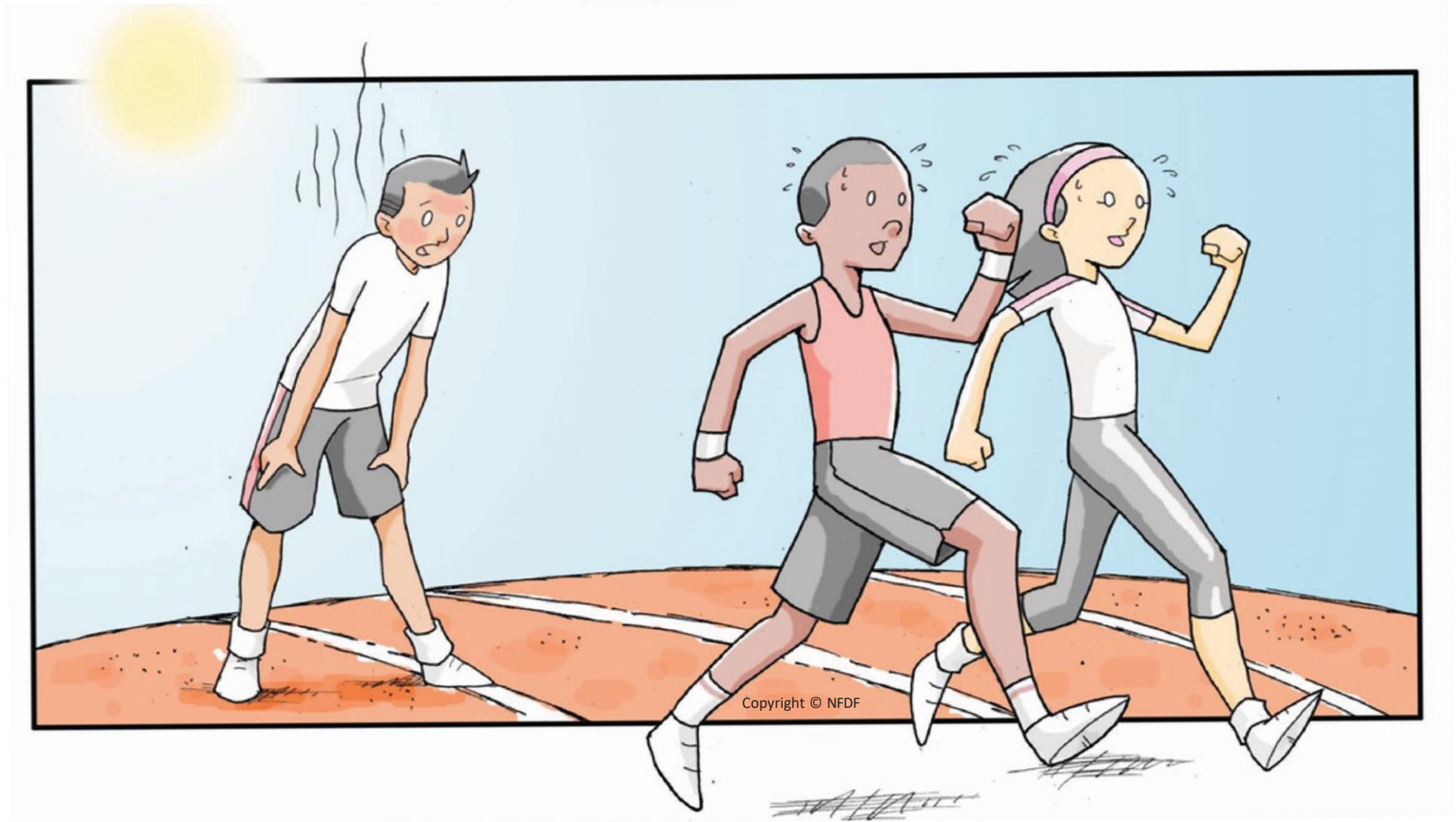
Some people with Fabry disease are always in a rush.



Chronic gastrointestinal (GI) upset is common in many people with Fabry disease. Diarrhea, constipation, early satiety (feeling full quickly when eating), bloating, heartburn, nausea and vomiting are all typical symptoms of Fabry disease. GI upset often interferes with daily activities and can have a significant negative impact on quality of life.



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



People with Fabry disease often have hypohidrosis which is a reduced ability to perspire that can cause them to overheat easily. They struggle with intolerance to hot temperatures, exercise, and strenuous physical activity. Hypohidrosis has been reported in about half of males and about one quarter of females with Fabry disease.

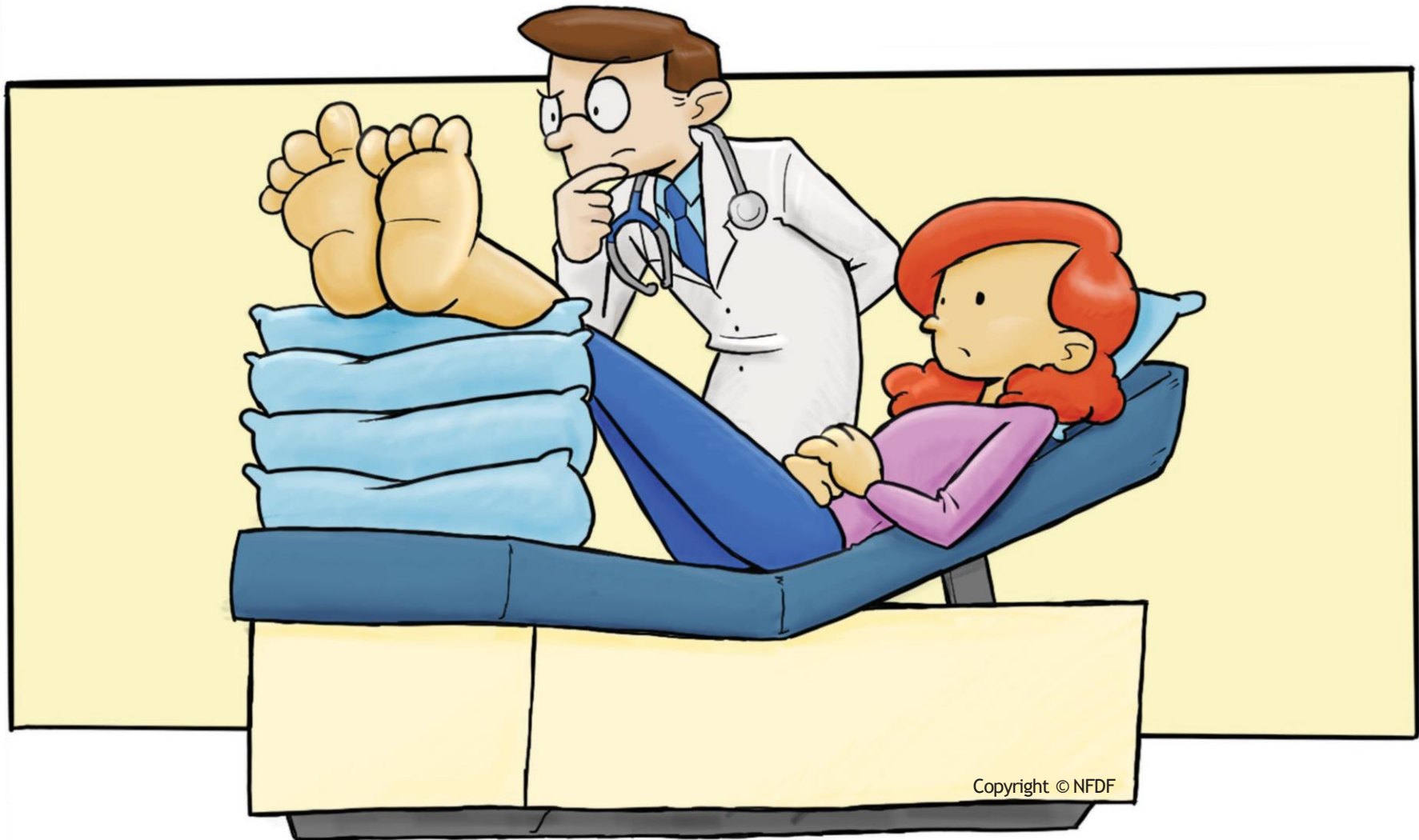


The burden of Fabry disease can be very stressful.



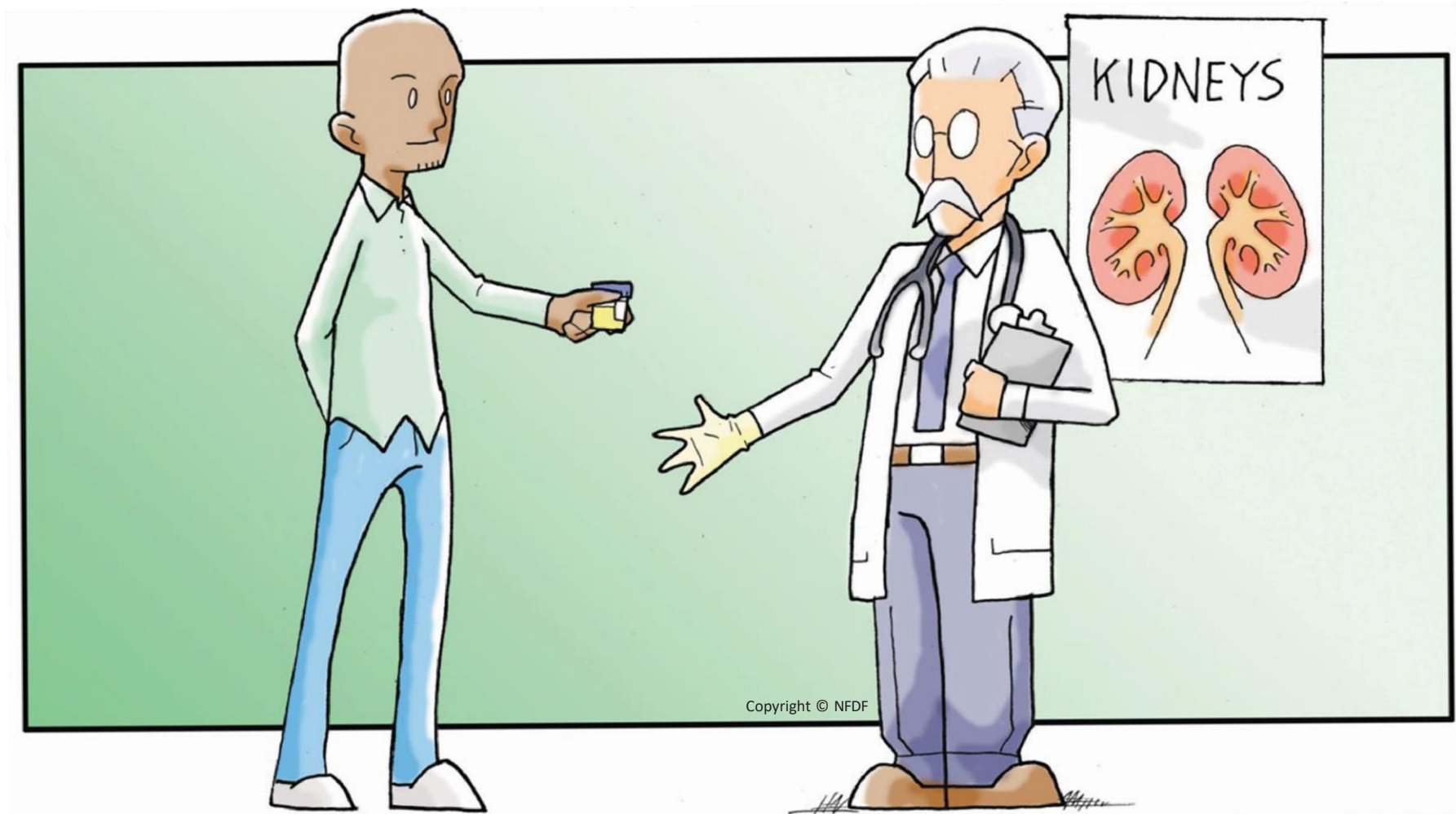
Anxiety, depression, and school avoidance can be common among people with Fabry disease. Free, confidential counseling is provided to people with Fabry disease and their family members 7 days a week, 24 hours a day through the National Fabry Disease Foundation's Family Assistance Program at 1-800-648-9557. Any personal issue is appropriate for discussion.

Many people with Fabry disease keep their heads held high ... and their legs held higher.



Unexplained swelling in the lower legs, ankles and feet ranging from mild swelling to pitting edema is common with Fabry disease. Pitting edema describes edema (excess fluid collection in the tissue) when pressure applied to the skin is released and an indentation is left behind.

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



Studies show that renal manifestations of Fabry disease occur early in life in a significant proportion of children, in many women, and in almost all men with classic disease. These manifestations progress to renal failure in nearly all males and some females. Albuminuria/proteinuria (albumin or protein in urine) are indications of kidney cell damage. Damage to kidney cells can occur at a very young age even before lab values begin to increase.

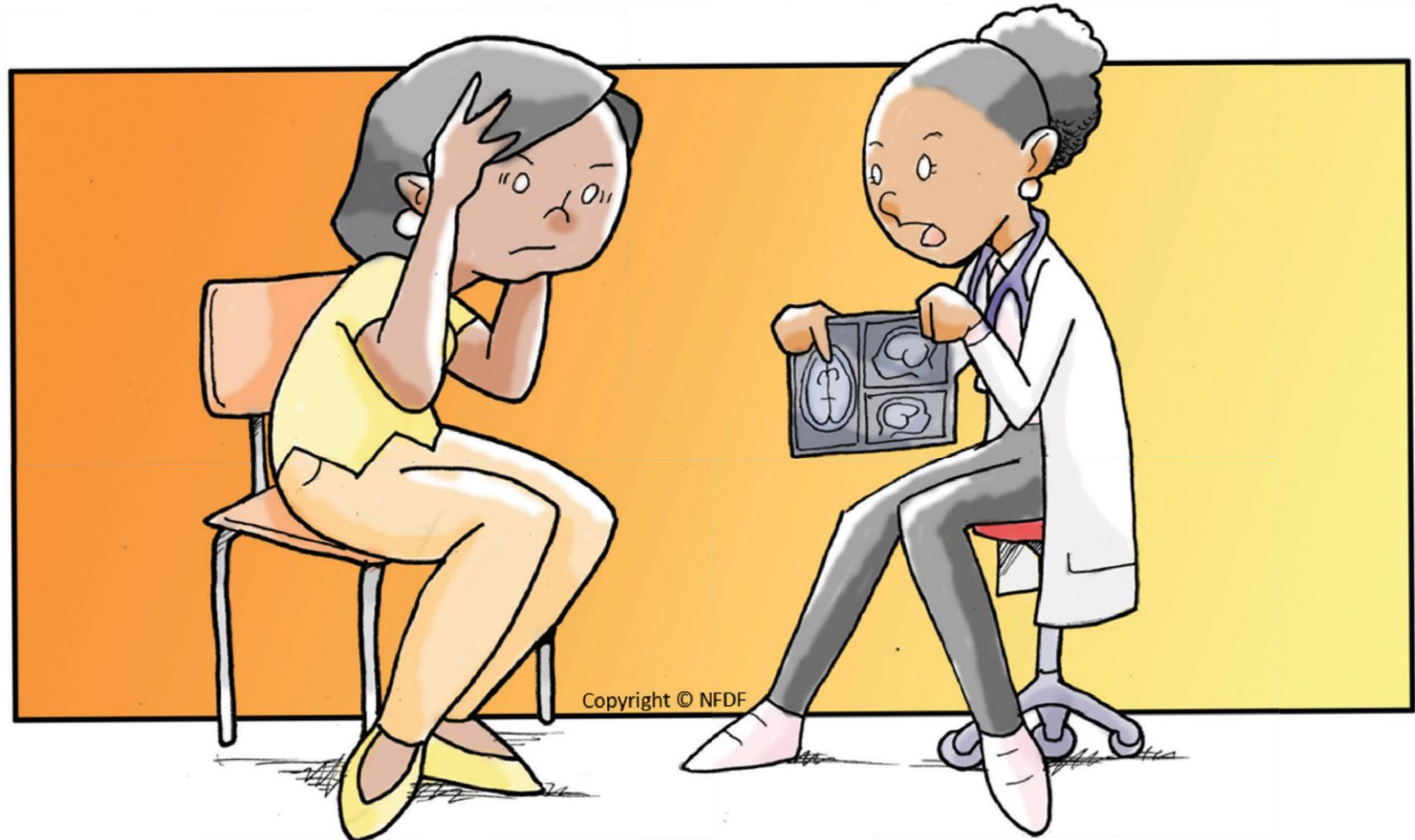


Many people with Fabry disease have a big heart and a courageous spirit.



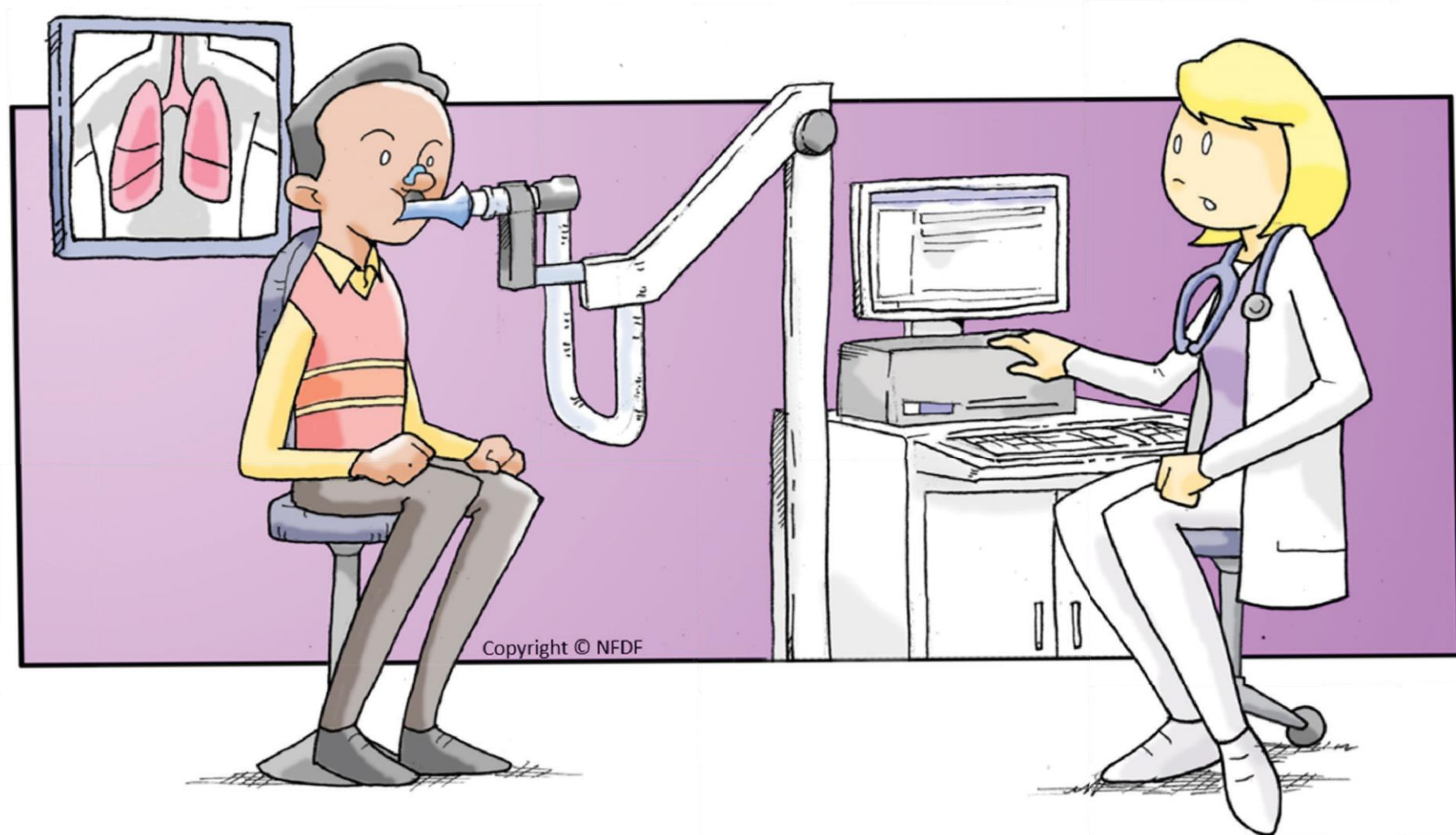
Common cardiac symptoms of Fabry disease include left ventricular hypertrophy (LVH), abnormal ECG which may include a fast or slow heart rate and conduction system disease (arrhythmias), fluctuating blood pressure, chest pain, palpitations, shortness of breath, dizziness, syncope (temporary loss of consciousness). Early heart attacks and/or heart failure may occur.

# People with Fabry disease often have a lot on their mind.



Neurological symptoms such as complex migraines, white matter lesions, transient ischemic attacks and stroke of unknown causes at a relatively early age are common with Fabry disease. Other neurological manifestations include peripheral nervous system and central nervous system involvement. Mild to debilitating neuropathic pain is common as well as symptoms of autonomic dysfunction such as reduced sweating, gastrointestinal issues, and cardiac rhythm issues.

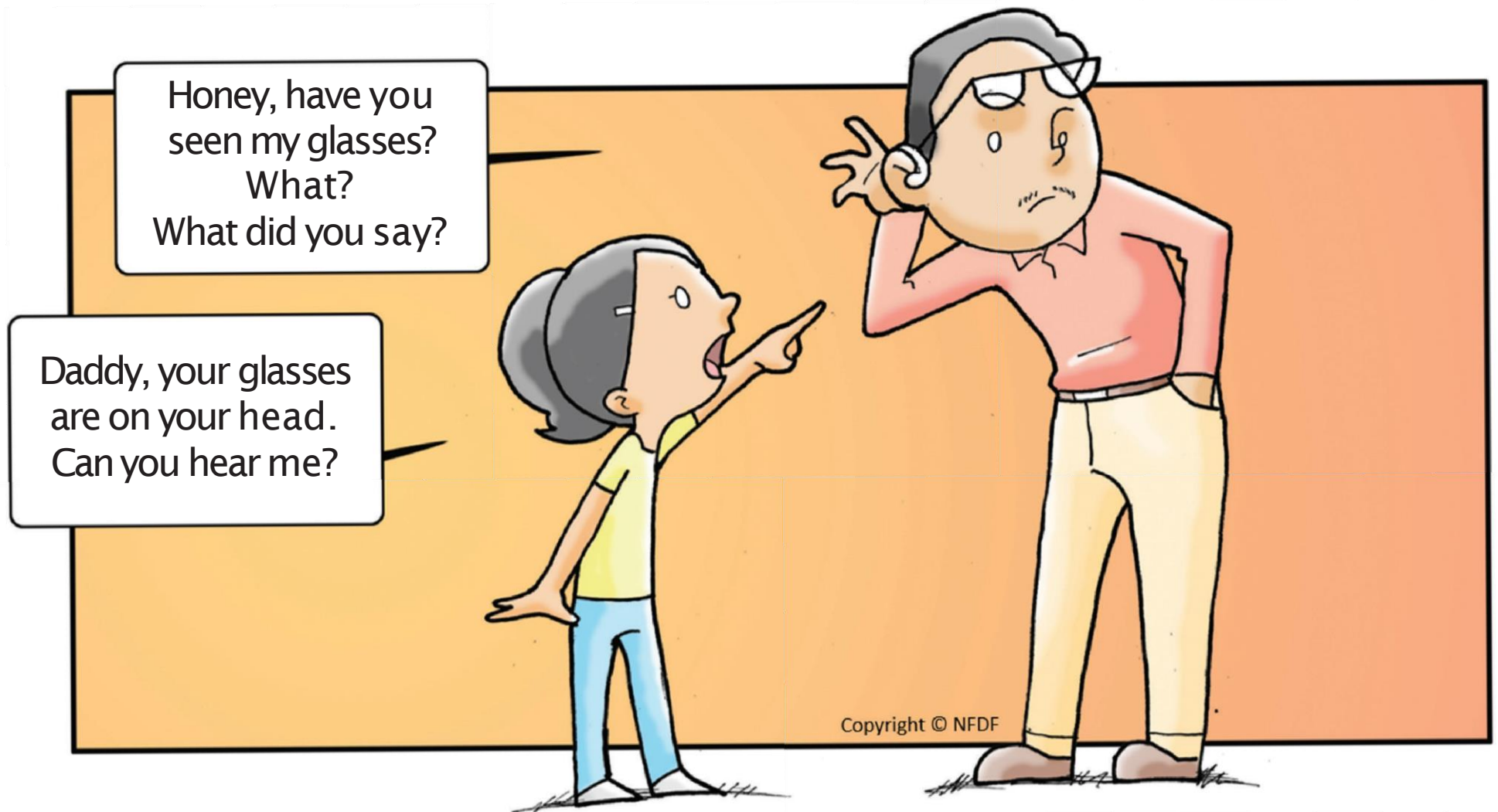
# Having Fabry disease can be a breathtaking experience.



Obstructive lung disease is common with Fabry disease. It often presents as wheezing, dyspnea (shortness of breath), or bronchitis and chronic cough sometimes occurs. A diagnosis of chronic Obstructive Pulmonary Disease (COPD) is common. A spirometry test is usually administered to determine how well the lungs function.



# People with Fabry disease tend to lose things!



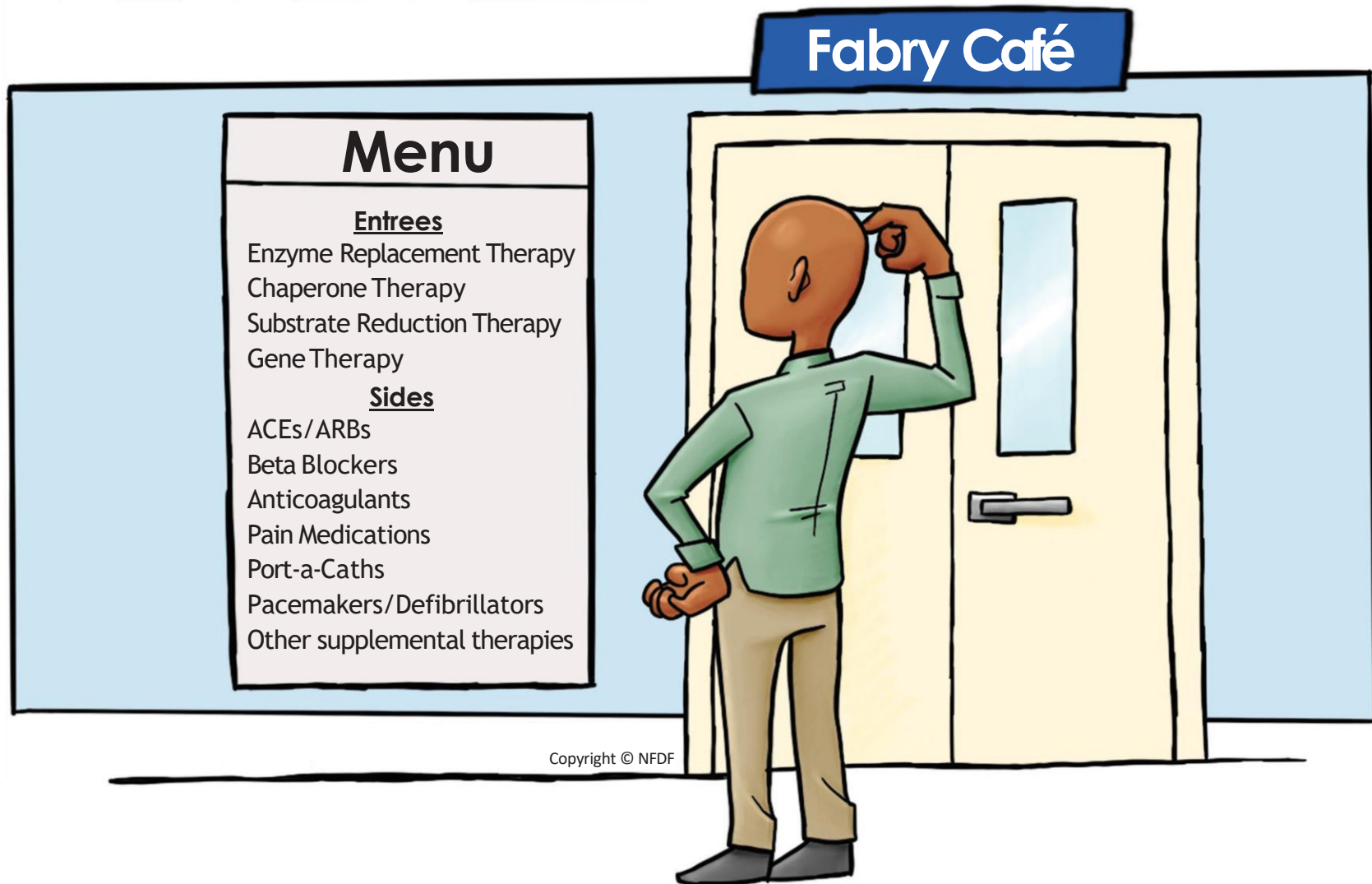
Common audiology symptoms caused by Fabry disease include progressive or sudden hearing loss and tinnitus. Acute hearing loss is about 60 times more prevalent than in the general population. Progressive hearing loss usually begins in men and women in the 2nd and 4th decades of life, respectively. Use of hearing aids is common. Dizziness and vertigo are also common.

# Adults with Fabry disease often raise questions about Raynaud's syndrome.



Symptoms of Raynaud's secondary to Fabry disease are cold fingers or toes, color changes in the skin in response to cold or stress, numbness, and prickly feeling or stinging pain upon warming or stress relief. Affected skin usually turns white while unaffected skin appears more red.

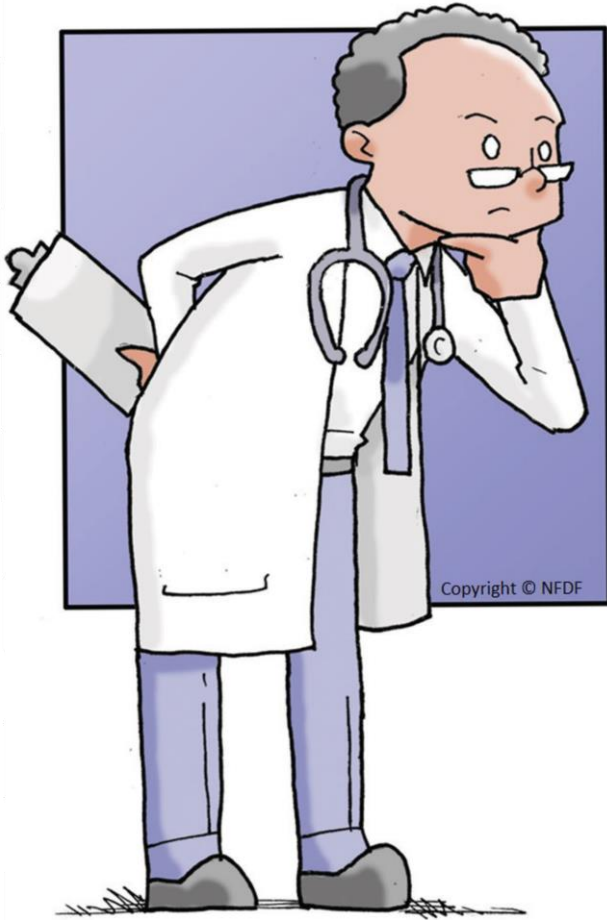
There are two approved treatment options currently available. The future may bring even more to our menu.



Enzyme Replacement Therapy (ERT) was first approved in Europe and many other countries in 2001, then in the U.S. in 2003. Oral Chaperone Therapy was approved first in Europe and other countries in 2016 and then the U.S. in 2018. A new ERT was approved in 2023. There are several other potential treatment options being investigated.



# A lot has been learned by studying the classics. The later-onset variations are a work in progress.



Fabry disease is a rare, progressive, destructive and often life threatening multi-system genetic disorder that is severely unrecognized or misdiagnosed.

The symptoms discussed in this calendar represent the most common symptoms of Fabry disease but not all possible symptoms. While there can be some variability in age of onset and severity, most males with classic disease experience most of these symptoms. The symptoms for females with classic disease and people with later-onset Fabry disease vary more widely.

In recent years, newborn screening has revealed a much higher than anticipated incidence of later-onset (also called non- classic) Fabry disease than previously known. People with later- onset Fabry disease **usually** do not experience early symptoms but "MAY" experience primarily heart dysfunction at a relatively young age.

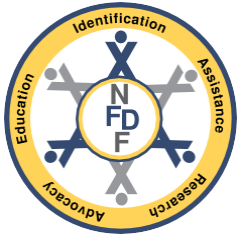
To stay informed and to receive resources, please register at the top of our website at [www.fabrydisease.org](http://www.fabrydisease.org) and subscribe to our e-newsletter from the orange newsletter icon on the righthand side of the website. Please see our programs handout and other resources on the website and please join our fight by participating in surveys and other important initiatives.

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



Children should not have to live a poor quality of life and adults should not have to die young from heart failure, kidney failure, and strokes. The future for individuals with Fabry disease is much brighter with the initiation of a Fabry specific treatments (ERT, chaperone therapy, substrate reduction therapy, and gene therapy), adjunctive therapies, leading a healthy lifestyle, and managing other risk factors. Earlier recognition, diagnoses, improved management, and treatment could have a profound positive effect on the lives of people with Fabry disease.

**Widespread physician and family education is critical to living better and longer lives!**



National  
**FABRY DISEASE**  
Foundation

National Fabry Disease Foundation  
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## **Giving more life to our children's years and more years to our children's lives!<sup>1</sup>**

As a charitable non-profit organization, we rely on the generosity of our donors to enable us to provide valuable education, assistance and support programs to individuals with Fabry disease and their families; and to provide education to physicians, others involved in medical management, and the public to improve the recognition, understanding, management, and treatment of Fabry disease.

**Please make a tax-deductible charitable donation on our website at [www.fabrydisease.org](http://www.fabrydisease.org) or by check to the above address. For our non-U.S. friends, please make a donation to the Fabry support group in your country.**

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

This calendar was designed and developed by Jerry Walter and illustrated by Mike Johnson who both have Fabry disease. To request a calendar, please email [jerry.walter@fabrydisease.org](mailto:jerry.walter@fabrydisease.org). For more information, email [info@fabrydisease.org](mailto:info@fabrydisease.org) or phone 800-651-9131 (U.S. toll free) / 919-732-2799.

Note 1 – Slogan borrowed and adapted from the Belgium Fabry Support Group. Thank you!