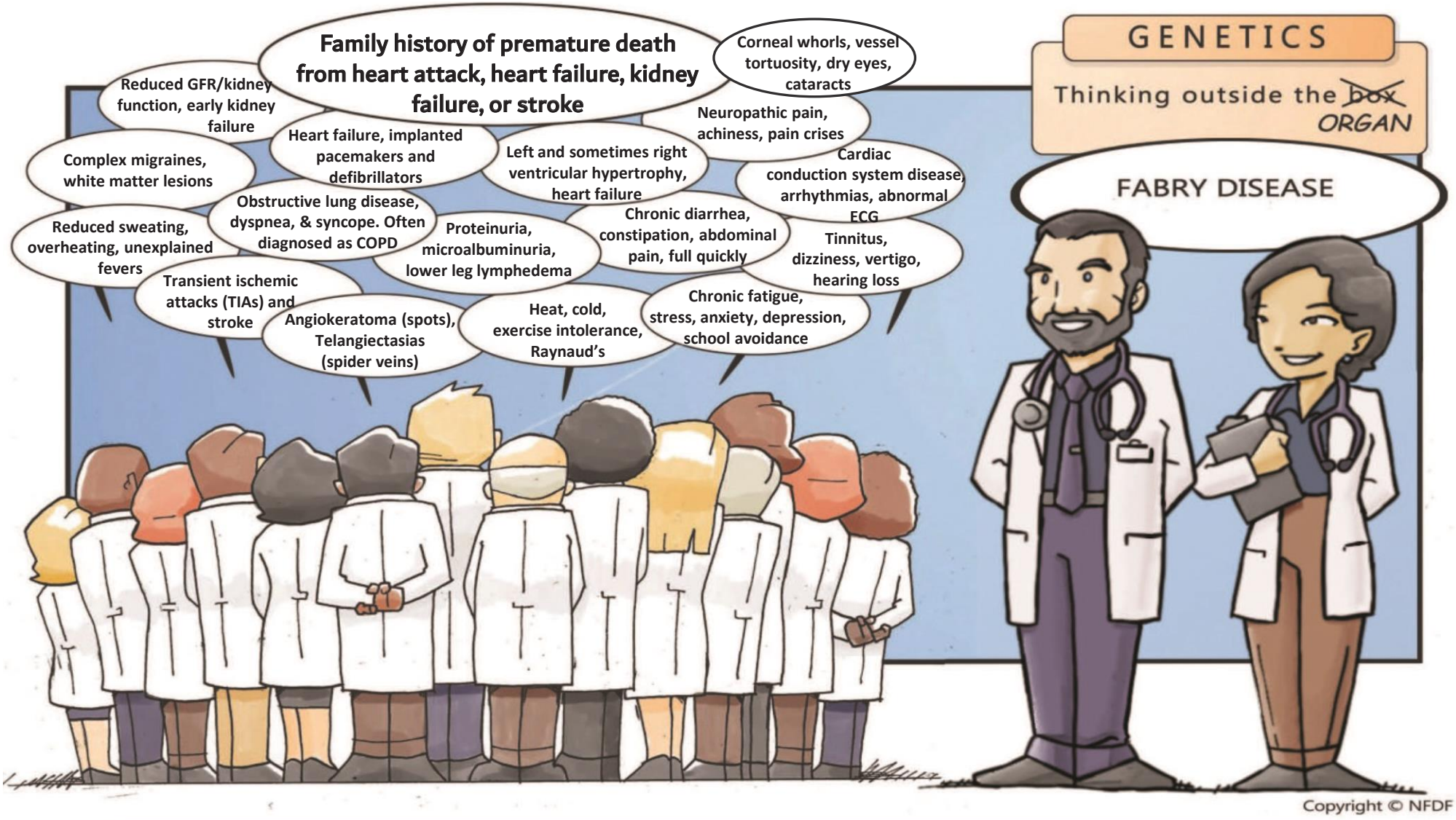
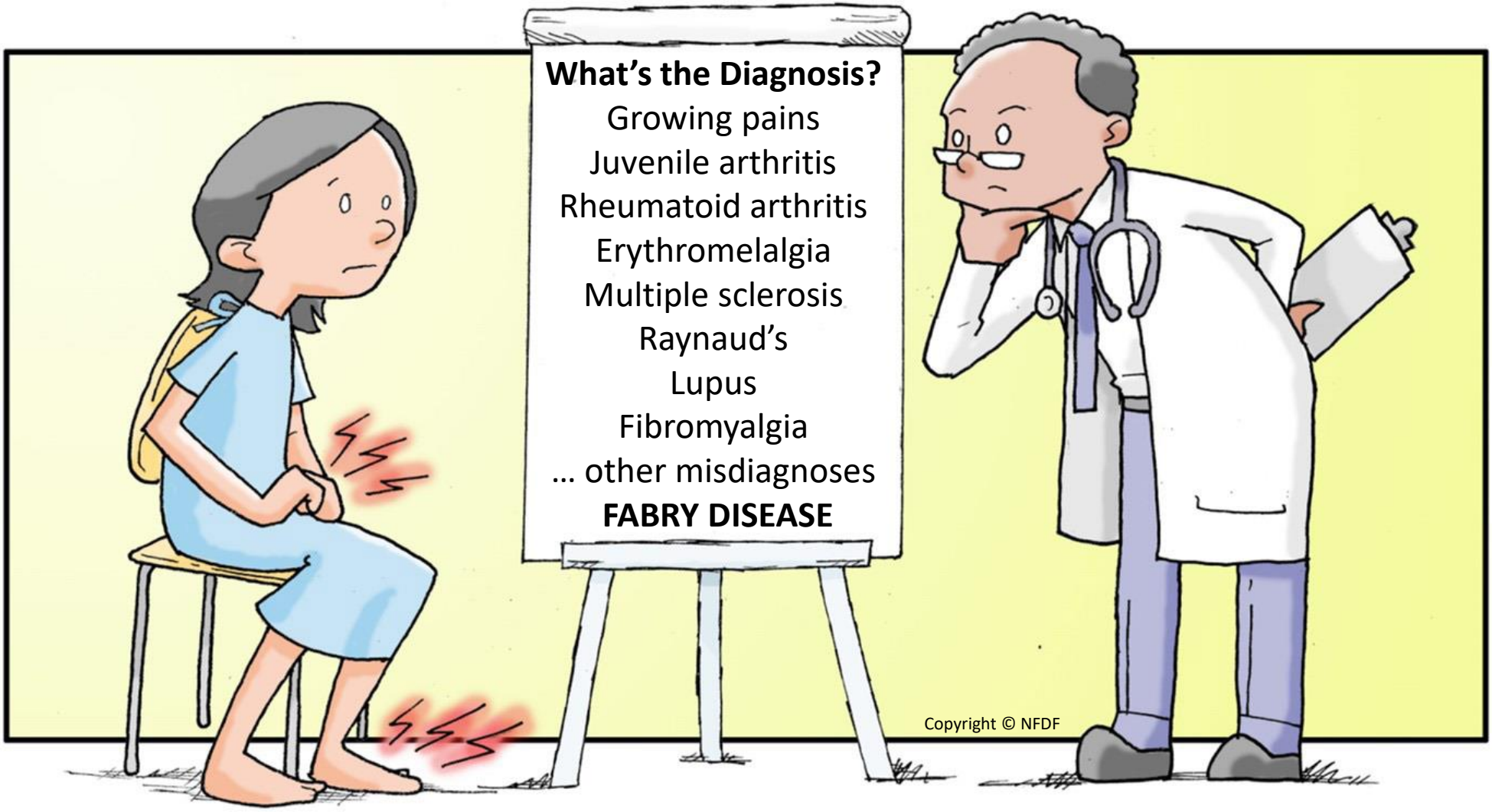
National
FABRY DISEASE
Foundation

Understanding the many symptoms of Fabry Disease



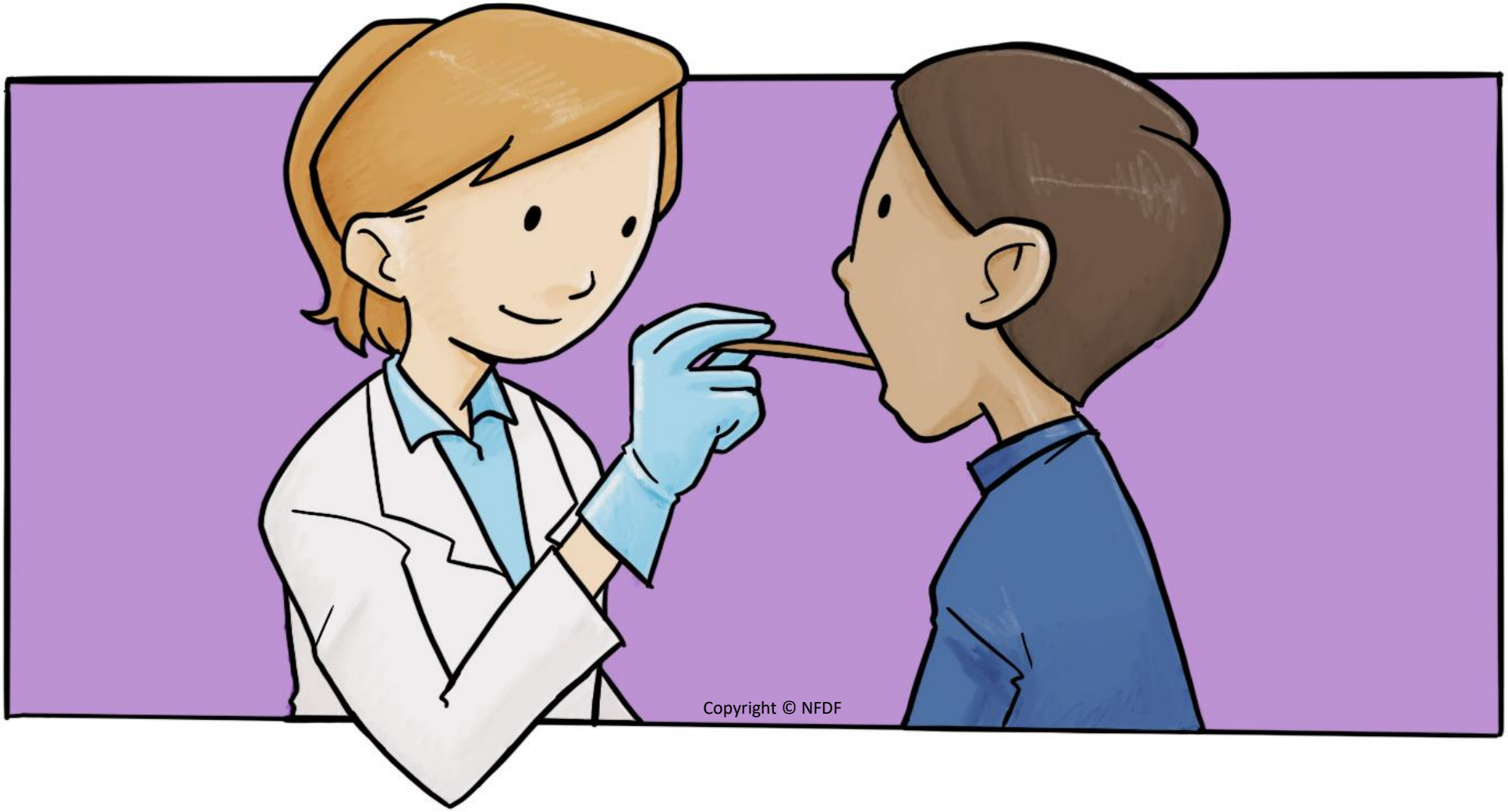
Fabry Disease Symptoms
These common symptoms may vary greatly among individuals.

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



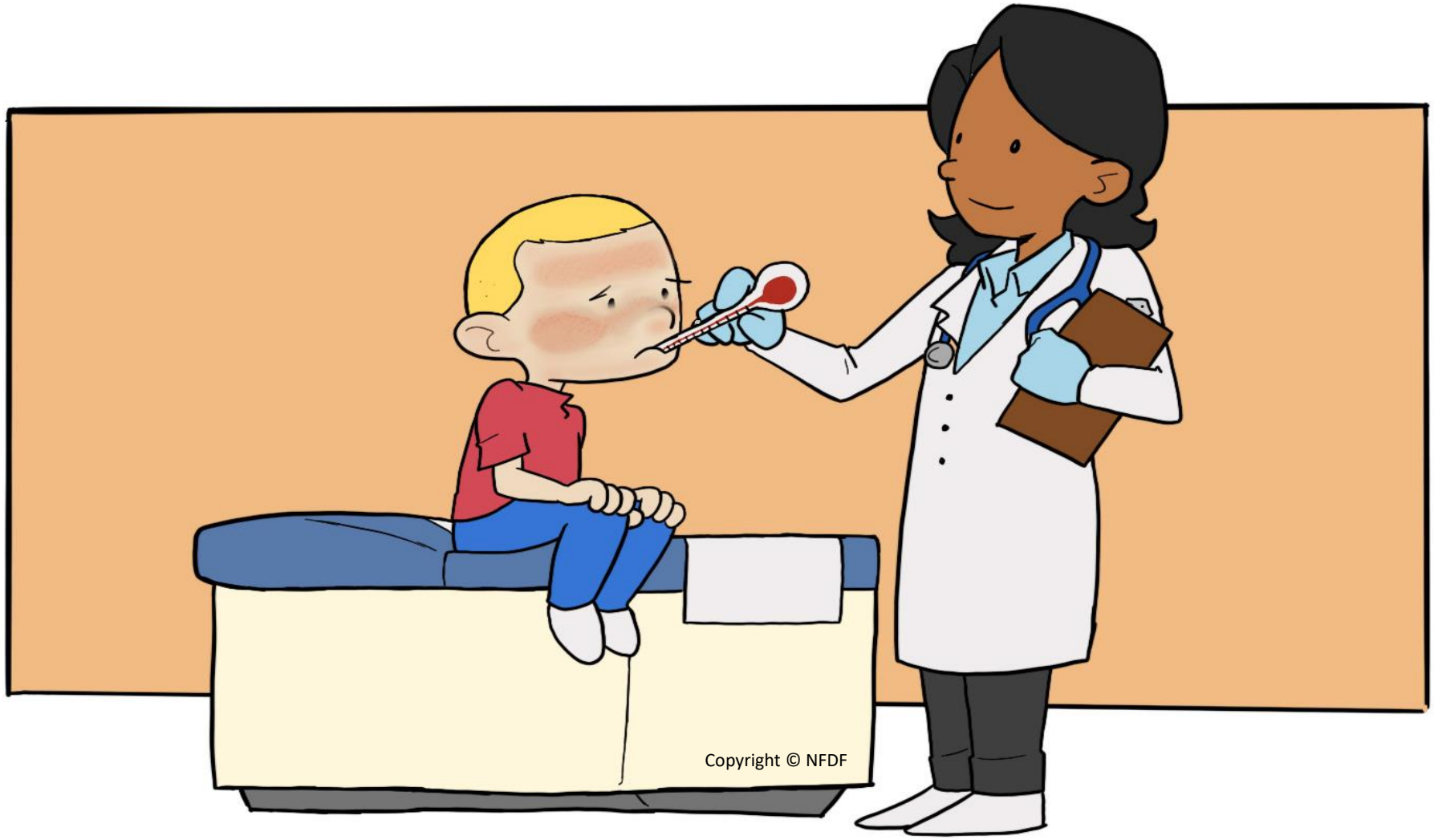
Small fiber peripheral neuropathy is a painful part of Fabry disease for many. Beginning in childhood, burning, and tingling pain in the hands and feet is very common. Other types of pain include chronic overall achiness, burning pain in other areas of the body, sharp, shooting pain in the extremities, and extended pain episodes/crises. Fabry pain is often misdiagnosed as other conditions. [1-4]

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



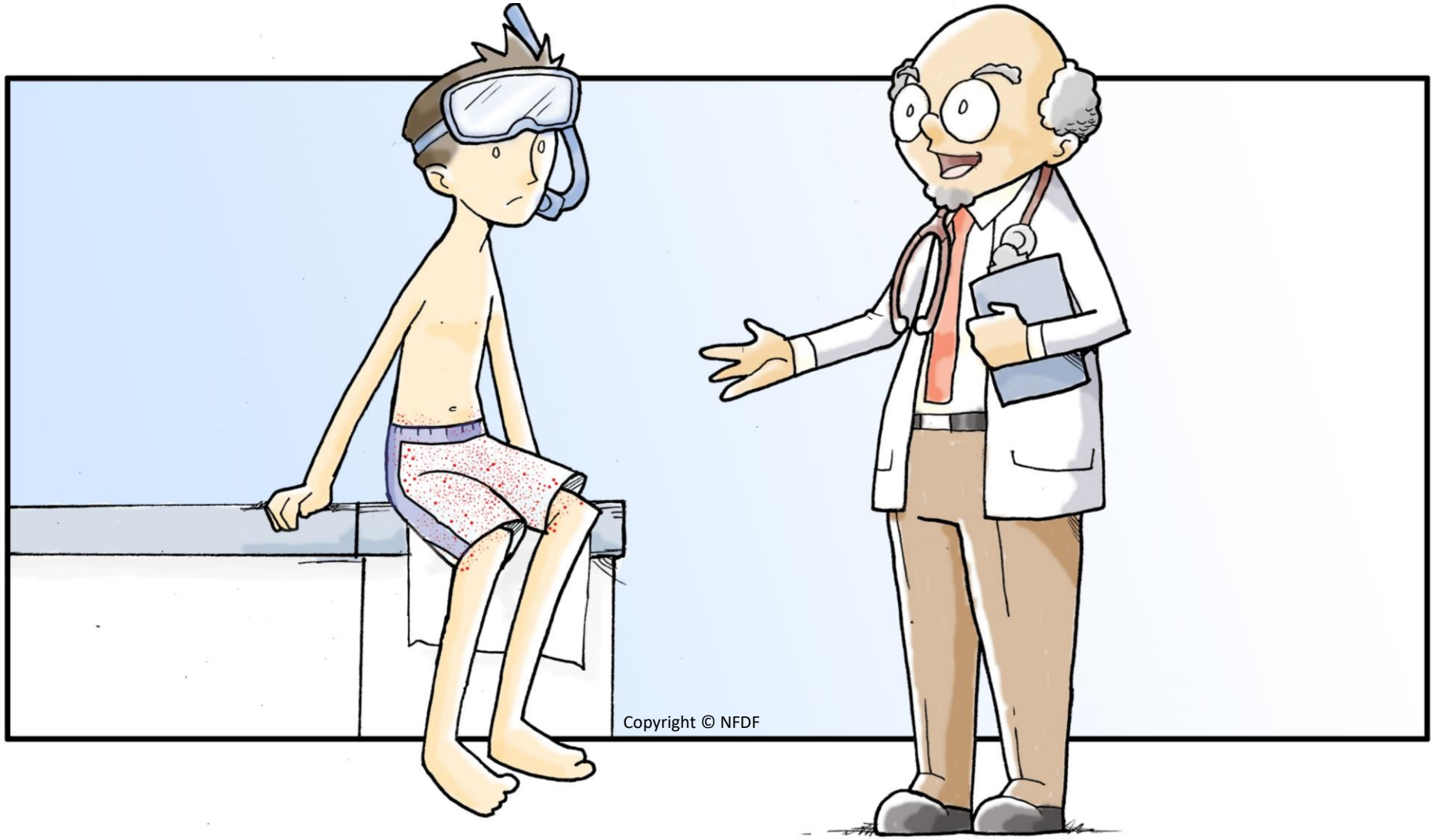
The genetic test to diagnose Fabry disease is called the *GLA* gene test. In males, an enzyme analysis is diagnostic for Fabry disease. It can be used to confirm whether a male has Fabry disease or not. In females, an enzyme analysis is not sufficient. Females with a Fabry *GLA* gene variant may have normal enzyme levels. For this reason, DNA analysis of the *GLA* gene must be performed for females. [5]

Some people work feverishly to manage their Fabry disease symptoms.



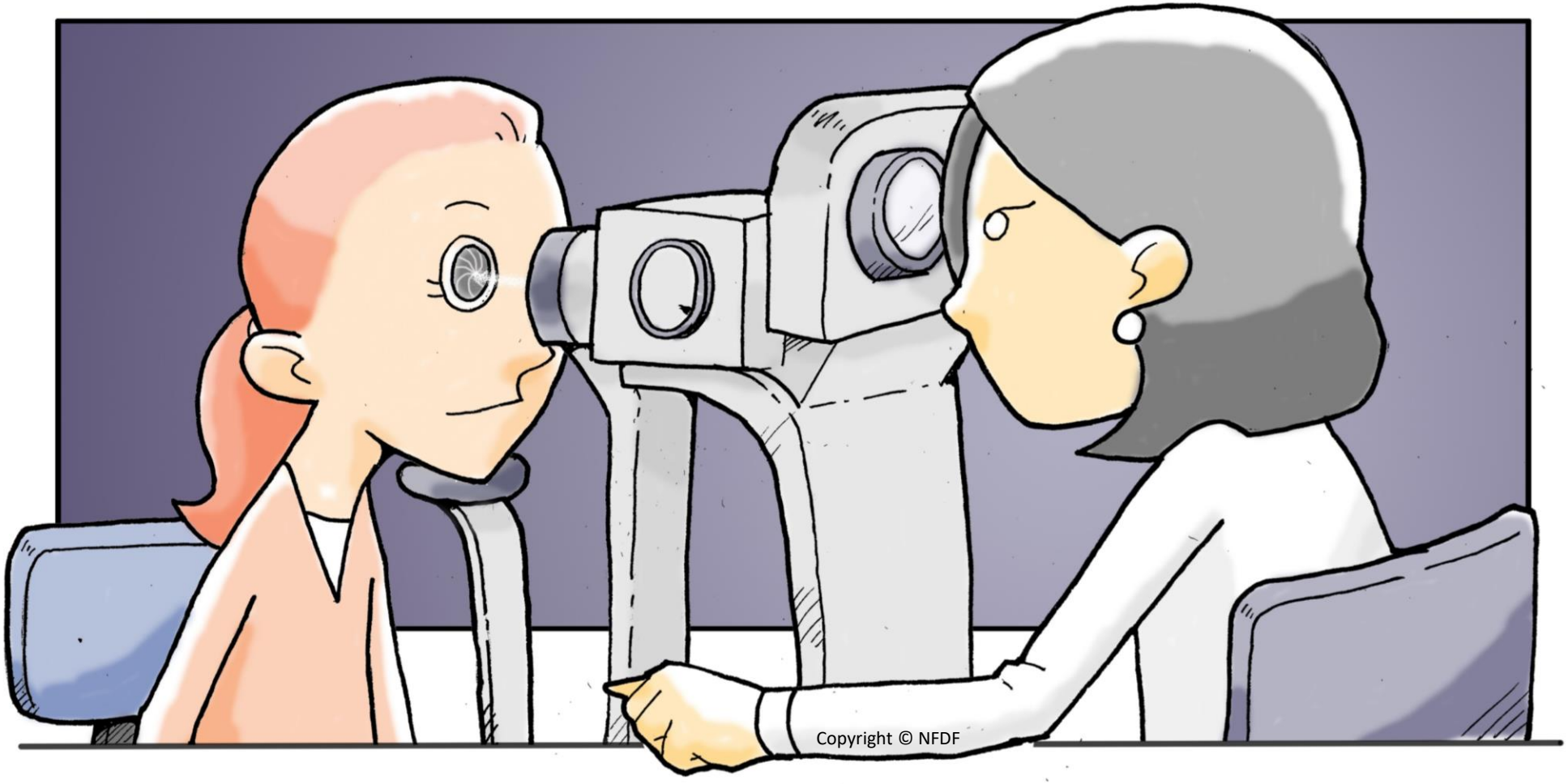
Many individuals with Fabry disease experience episodes of unexplained high fever, especially in childhood and adolescence, but also throughout other stages of life. Recognizing recurrent fever of unknown origin as a symptom of Fabry disease could lead to an early diagnosis. [4,6,7]

Connecting the right dots can lead to a rare discovery.



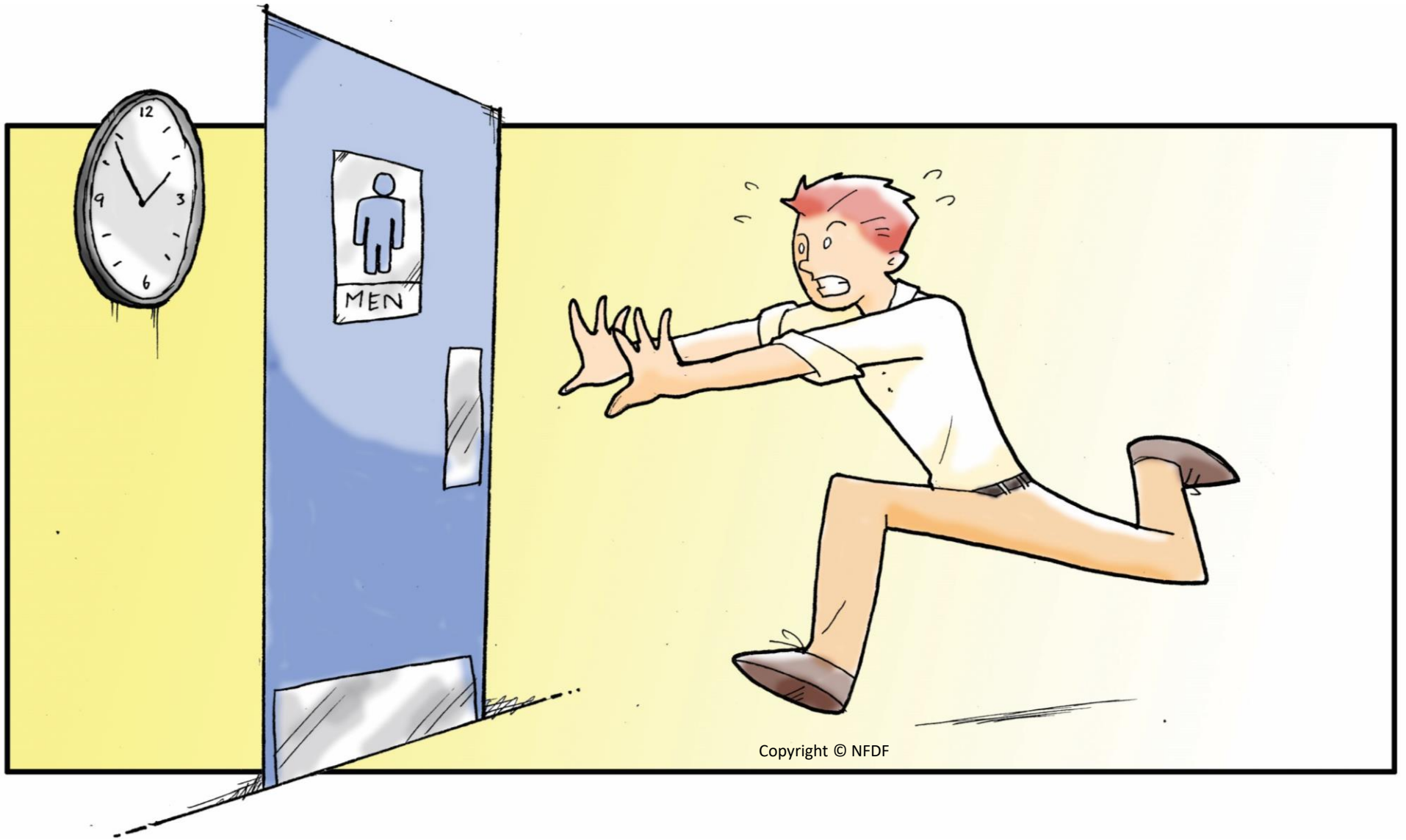
Fabry angiokeratomas are small, sometimes clustered, red or reddish-purple skin lesions (tiny spots/dots from dilated capillaries) that usually occur in the bathing trunk area (lower back to knees) but can be seen anywhere on the body. When present, they usually appear in childhood (a little later in girls than boys) and increase in number and size with age. Reports indicate they are present in 66% of males and 36% of females. Some people also have telangiectasias, which look like red broken blood vessels under the skin (spider veins). [4,8]

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



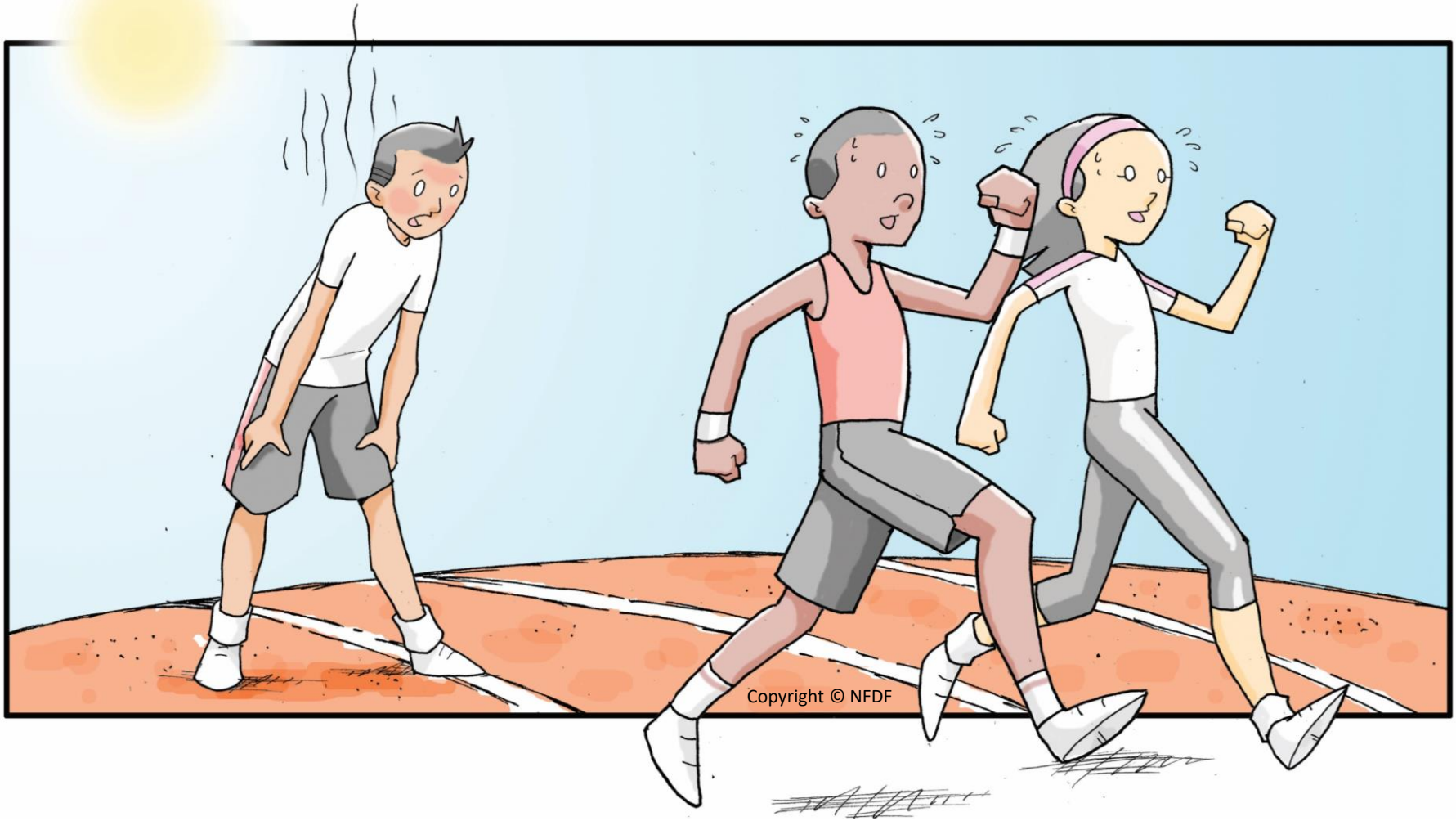
The most typical eye signs in Fabry disease are whorl-like opacities called cornea verticillata or corneal whorls that can be seen by an eye doctor in a routine eye (slit-lamp) exam. Eye manifestations do not usually cause significant visual impairment. Among other common manifestations are Fabry cataracts, which can usually be repaired. [4]

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, which vary among individuals. They may interfere with daily activities and can negatively impact quality of life. [9]

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

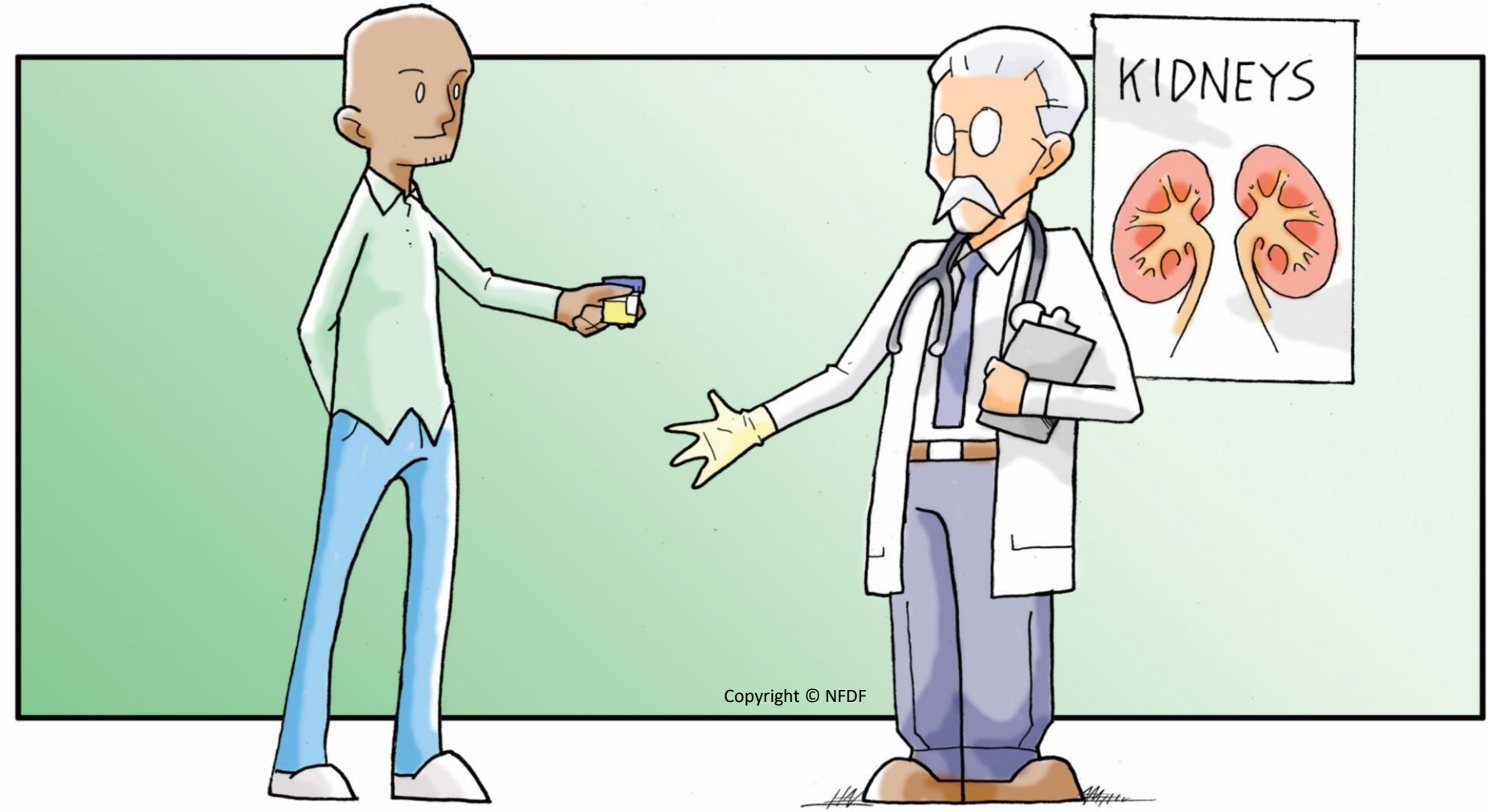


People with Fabry disease often have hypohidrosis, which is a reduced ability to perspire that can cause them to overheat easily. Many individuals struggle with intolerance to hot temperatures, exercise, and strenuous physical activity. Hypohidrosis has been reported in about half of males and about one-third of females with Fabry disease. [4,10,11]

The burden of Fabry disease can be very stressful.

Anxiety, depression, and school avoidance are common among people with Fabry disease. Free, confidential counseling and referrals are 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. Visit https://www.fabrydisease.org/index.php/programs-projects/family-assistance-program. Webinars and other resources are also available. Use "nfdf" as your company code if asked. [3]

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

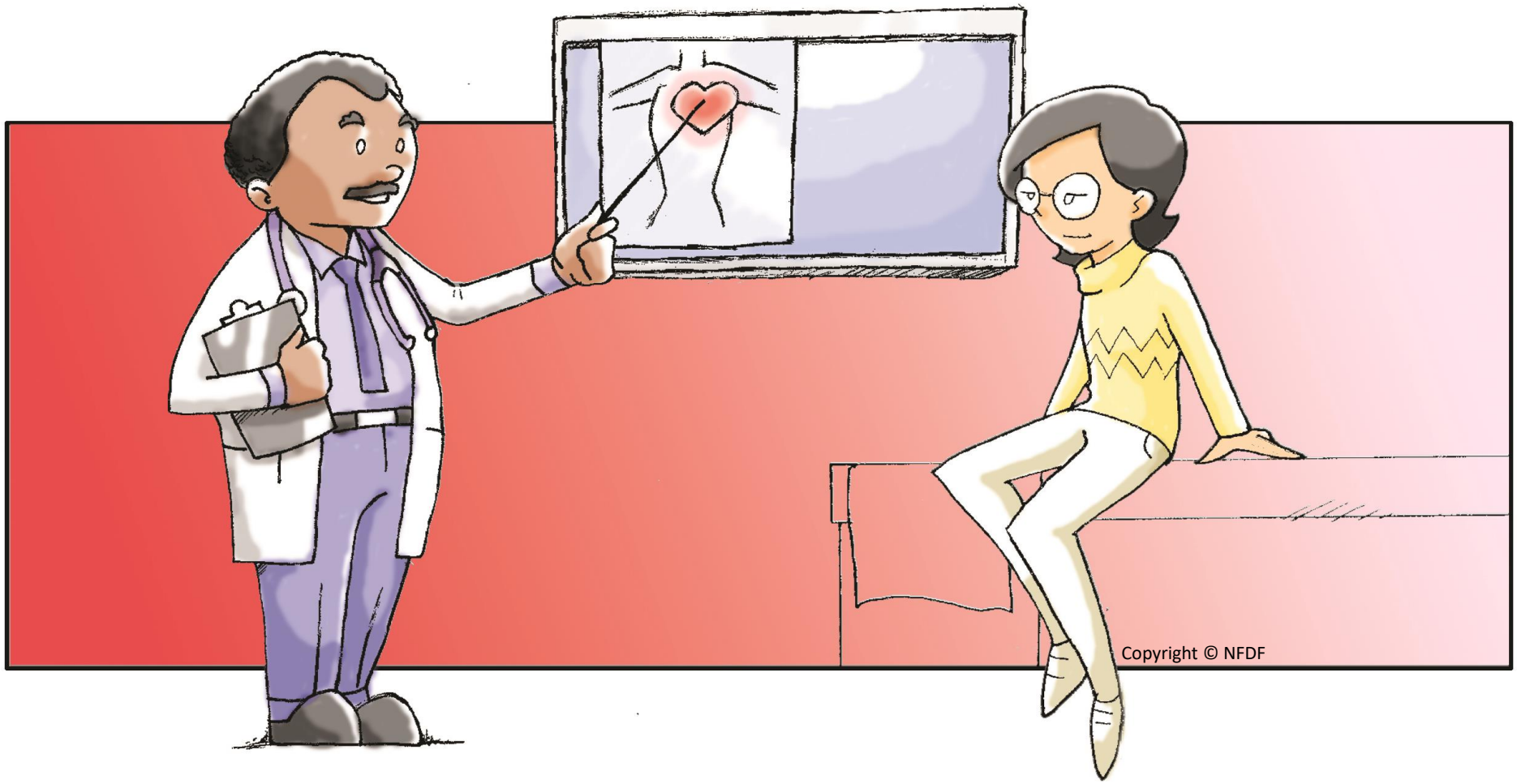


Studies have shown that GL-3 accumulation in the kidneys of people with Fabry disease can occur even before birth. Microalbuminuria and proteinuria (albumin or protein in urine), podocyturia, and reduced glomerular filtration rate (GFR) are early indications of kidney cell damage. Without Fabry-specific and adjunct treatments, kidney manifestations may cause progressive kidney dysfunction, failure, and transplant in adulthood. [4,12,13,15]

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

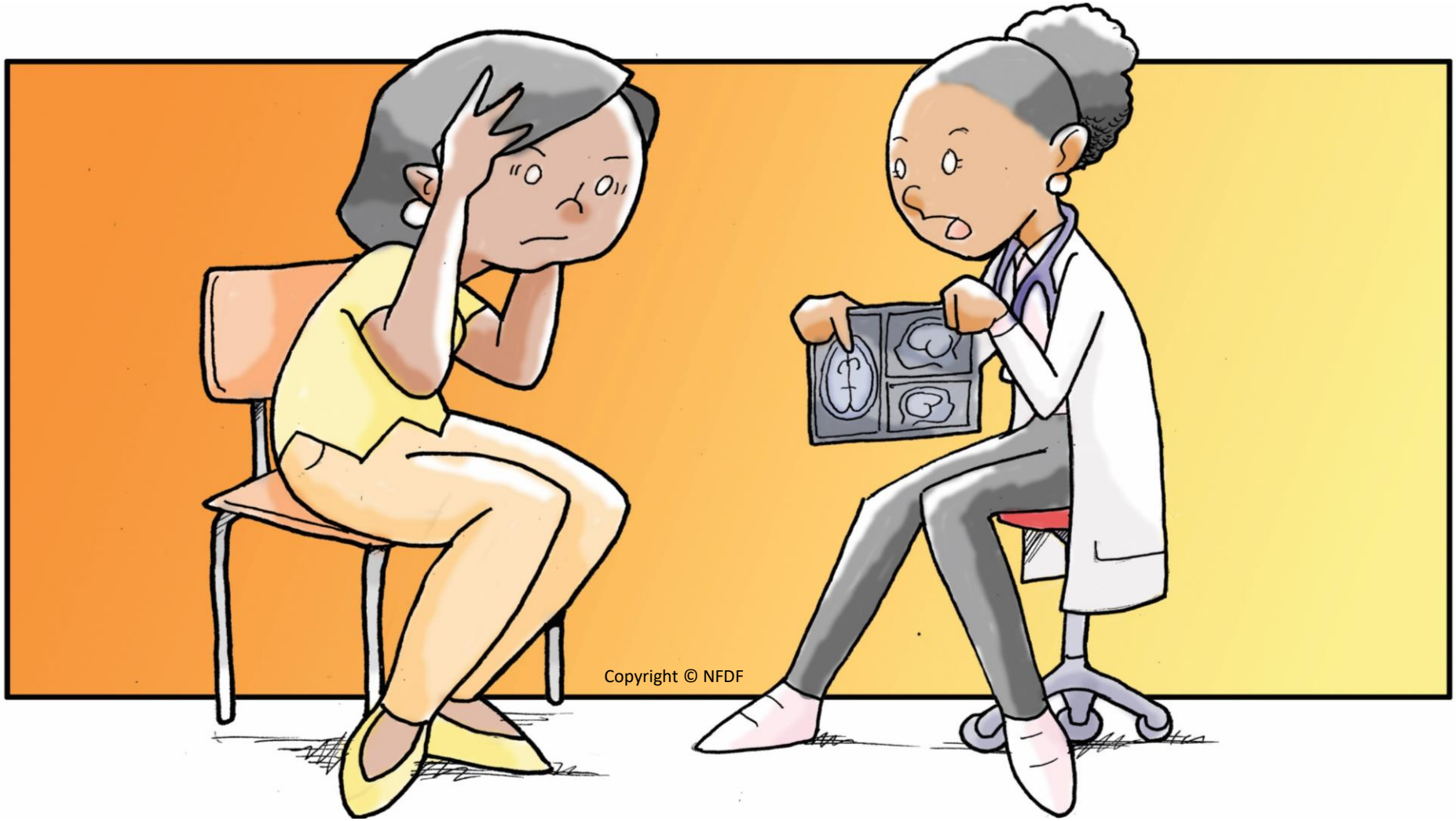
Unexplained swelling in the lower legs, ankles, and feet is common with Fabry disease, ranging from mild swelling to pitting edema. Pitting edema describes edema (excess fluid collection in the tissue) when pressure applied to the skin is released, and an indentation is left behind. [10]

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



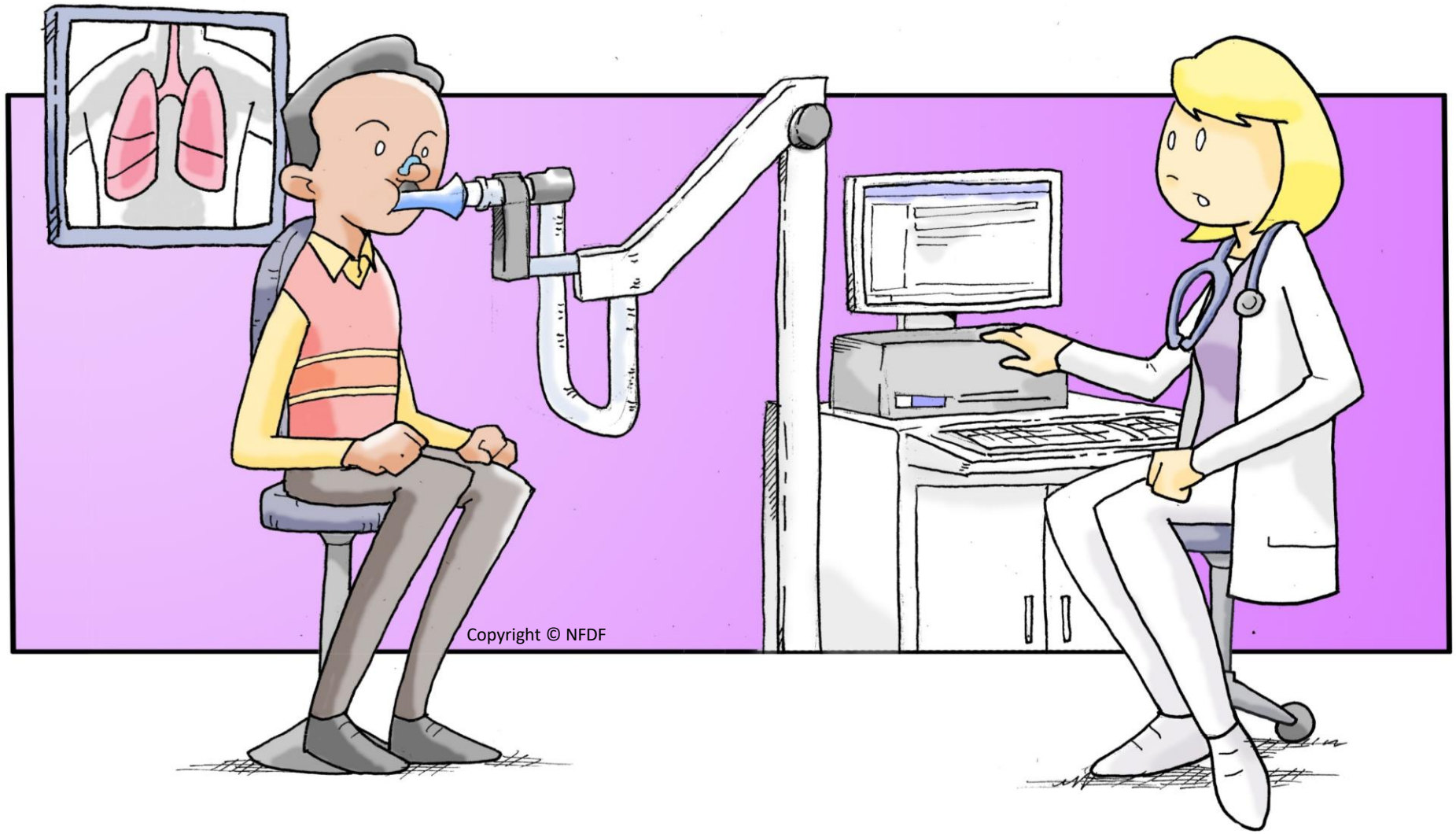
Common cardiac symptoms of Fabry disease include left ventricular hypertrophy (LVH), sometimes right ventricular hypertrophy (RVH), abnormal ECG, which may include a fast, slow, or irregular heart rate, conduction system disease (arrhythmias), fluctuating blood pressure, chest pain, palpitations, shortness of breath, dizziness, syncope (temporary loss of consciousness). Early heart attacks and/or progressive heart failure may occur. [3,4,14,15]

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



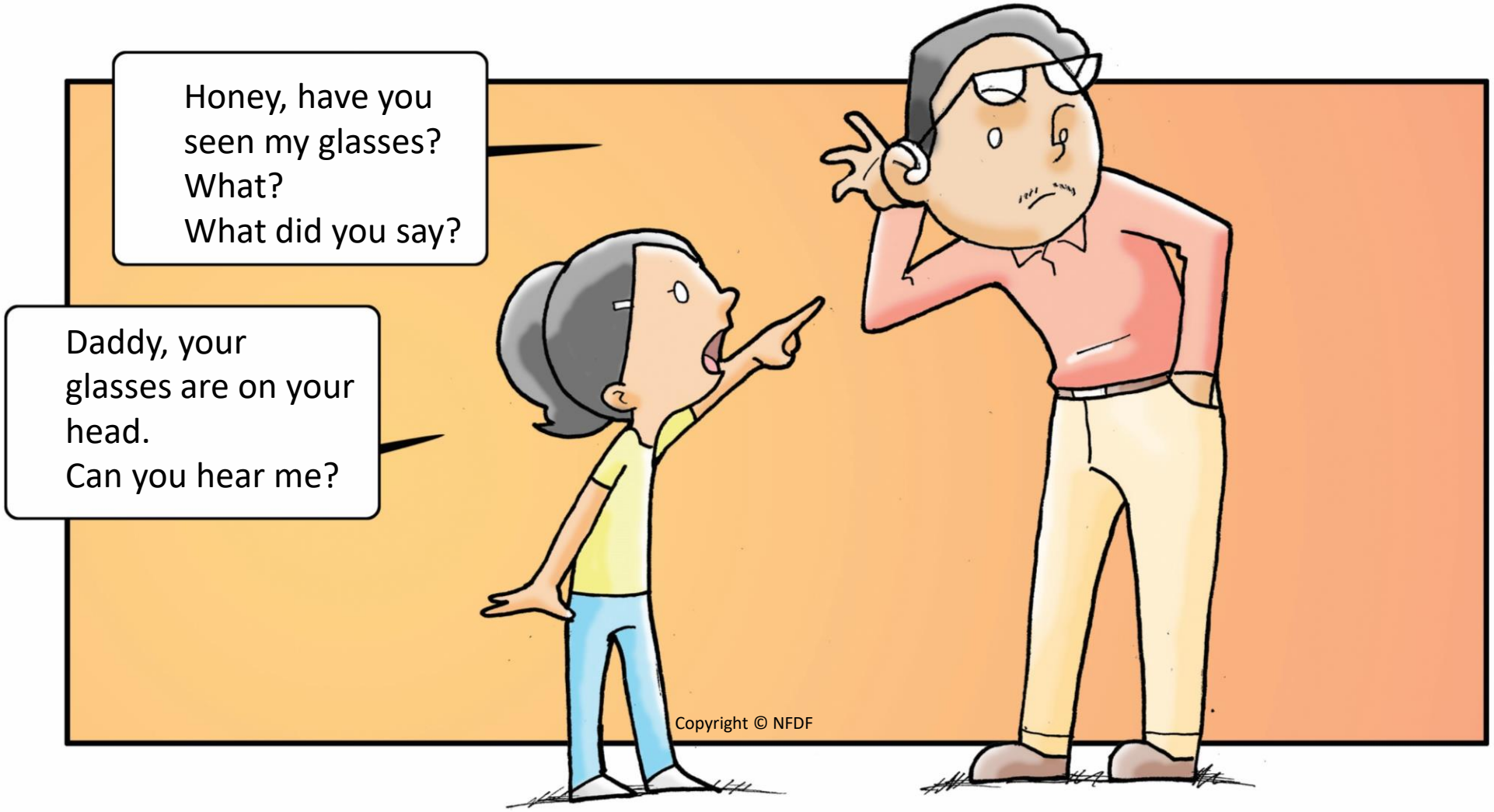
Neurological symptoms may include complex migraines, brain white matter lesions, transient ischemic attacks (TIAs), and stroke at a relatively early age in adults. Other neurological manifestations include peripheral nervous system and central nervous system involvement, mild to debilitating neuropathic pain, as well as symptoms of autonomic dysfunction such as reduced sweating, gastrointestinal issues, and cardiac rhythm issues. [4,15,16]

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. [2]

People with Fabry disease tend to lose things!

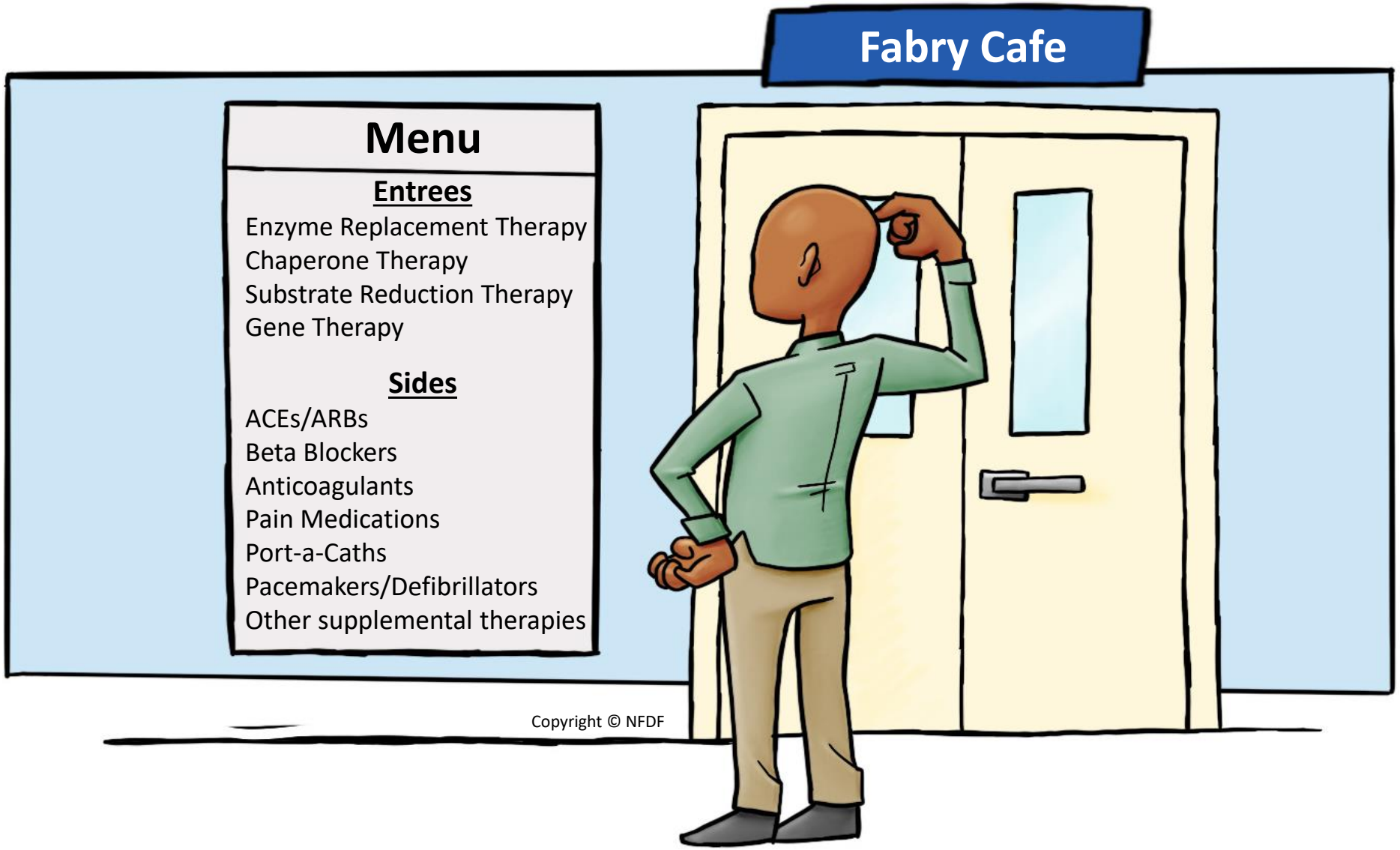


Hearing loss and tinnitus (ringing in the ears) are common in adults with Fabry disease. While most hearing loss happens slowly, there have been reports of sudden hearing loss (which can sometimes be recovered with quick medical intervention). Inner ear dysfunction from Fabry may also cause abnormal balance, dizziness, or vertigo (spinning dizziness). The use of hearing aids is common in adults, and cochlear implants are sometimes needed. [3,4,5]

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

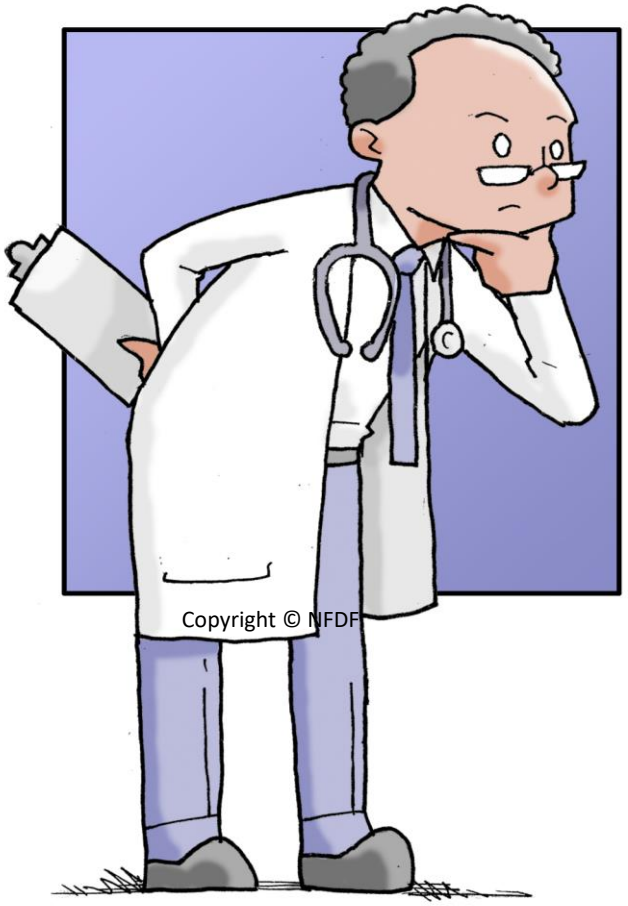
Symptoms of Raynaud's secondary to Fabry disease are cold fingers or toes, color changes in the skin in response to cold or stress, numbness, and prickly feeling or stinging pain upon warming or stress relief. Affected skin usually turns white, while unaffected skin appears more red. [17]

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



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

Much has been learned by studying the classics. The non-classic variants are a work in progress.



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

The symptoms described in these pages represent the most common symptoms of Fabry disease but not all possible symptoms. While there can be great variability in occurrence, age of onset, and severity, most males with classic disease experience most of the common symptoms. Females with classic variants can range from seemingly asymptomatic to severe. The non-classic forms, also called later-onset, do not usually cause the typical early symptoms of Fabry but may result in heart or kidney dysfunction at a relatively young age. Exceptions have been reported. [18]

In recent years, newborn screening has revealed a much higher than anticipated incidence of later-onset Fabry disease.

To stay informed and receive resources, please subscribe to our e-newsletter from the orange newsletter icon near the top right of our website at www.fabrydisease.org. Also, please see our many other programs and resources on the website, and please join our fight by participating in surveys and other important initiatives as they are distributed. See our Link Tree at https://linktr.ee/thenfdf.

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, heart attacks, kidney failure, or strokes. Fabry disease is a complicated disease. Much progress has been made to improve the understanding, management, and treatment of Fabry disease, but as questions are answered by research and solutions are implemented, more questions arise. Research continues, and the future of people with Fabry disease looks brighter!

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

TheNFDF

The National Fabry Disease Foundation

Visit our Link Tree for links to important NFDF resources and social media sites.

NFDF Resources:

- NFDF Website
- Sign up for our Newsletter!
- NFDF's YouTube Video Index
- Educational Symptoms Presentation
- Fabry Facts Playing Cards – Answers to FAQs
- U.S. Primary Financial Assistance Programs
- NFDF Family Assistance Program (Code: nfdf)
- Sponsored (cost-free) U.S. Genetic Testing
- 2023-2024 NFDF Webinar Meeting Space

NFDF Social Media:

- Facebook
- Instagram
- TikTok
- Twitter (X)
- YouTube
- LinkedIn

https://linktr.ee/thenfdf

The Annual Fabry Family Education Conference by the National Fabry Disease Foundation

The expense-free annual Fabry Family Education Conference in Greensboro, NC, is usually held in September but sometimes in October, just before the annual Fabry Family Weekend Camp. All individuals and families with Fabry may attend the conference, which includes an exhibit hall, educational presentations, family chat sessions, and chaperoned kids and teens activity rooms. Subscribe to the NFDF's Fabry Focus e-newsletter to stay up-to-date with the conference registration process and due dates.

The Annual Fabry Family Weekend Camp by the National Fabry Disease Foundation

The expense-free annual Fabry Family Weekend Camp at Victory Junction in Randleman, NC, is usually held in September but sometimes in October after the Fabry Family Education Conference. Families with at least one child with Fabry, ages 6 to 16, are eligible to attend the camp. The application process usually starts in June. Subscribe to the NFDF's Fabry Focus e-newsletter to stay up-to-date with the camp application process and due dates.

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National
FABRY DISEASE
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National Fabry Disease Foundation
144 E King Street
Unit 1419
Hillsborough, NC 27278

Giving more life to our children's years and more years to our children's lives! Note 1

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 educate 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 near the top right side of our website at www.fabrydisease.org, our Facebook page at www.facebook.com/FabryDisease, or by check to the above address.

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

This calendar was designed and written by Jerry Walter, MS, and illustrated by Mike Johnson, both of whom have Fabry disease. Casey McKenna, MS/CGC, NFDF staff member, contributed to the 2024-2025 revision. To receive a calendar, please contact jerry.walter@fabrydisease.org. For information, email info@fabrydisease.org or phone 800-651-9131 (U.S. toll-free) / 919-732-2799.

Note 1 – slogan borrowed with permission from the Belgium Fabry Patient Support Group. Thank you!