TAKING BETTER CONTROL OF FABRY DISEASE

How Regularly Scheduled Assessments Can Help

FEATURING A CONVERSATION WITH DR. CHRISTINE ENG

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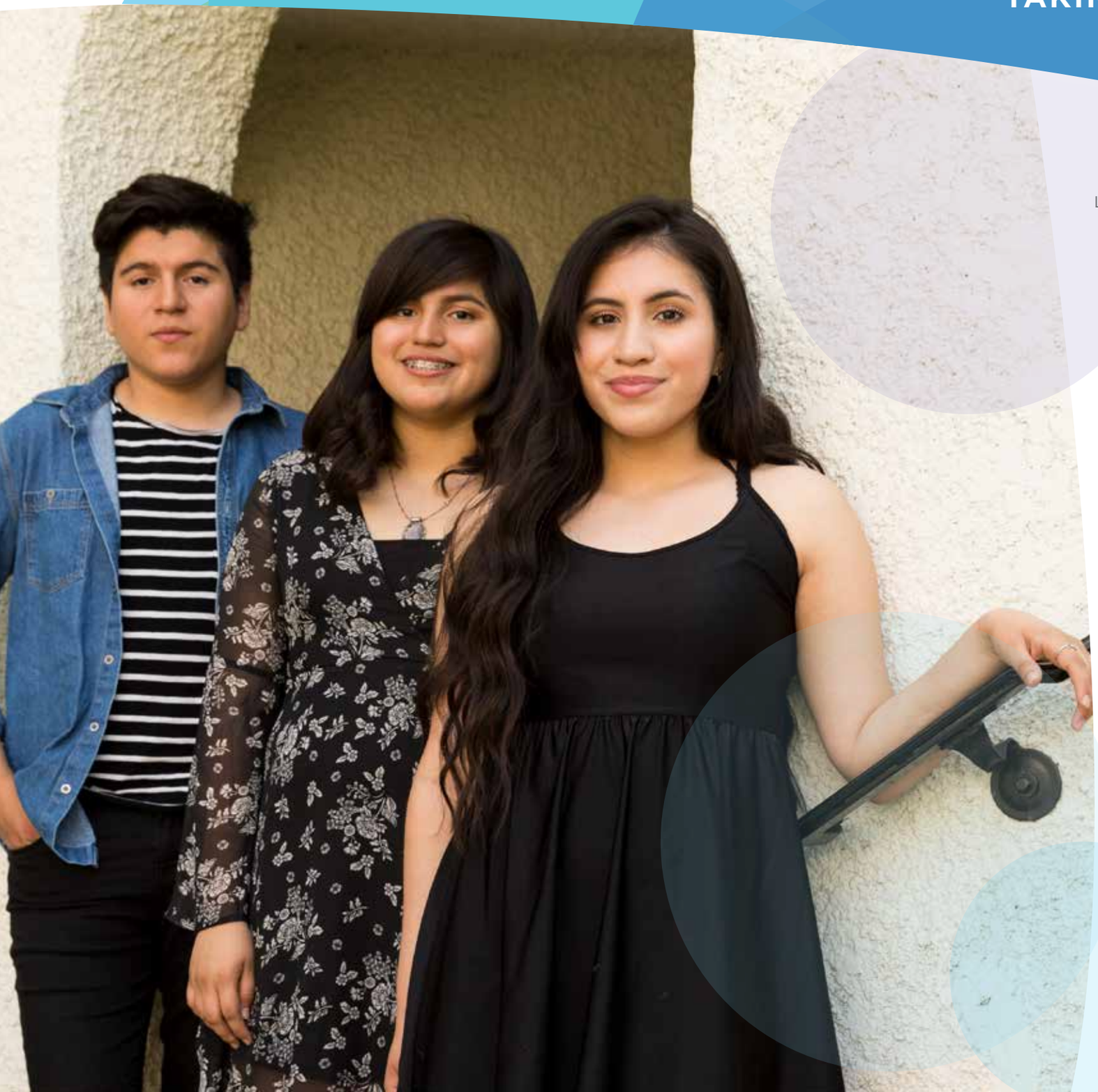

Living with Fabry disease can be challenging, but the more you understand about the condition and how it affects your body, the more control you'll have over your health. Regular visits to your doctor are one of the most important ways to monitor your disease. Through a variety of non-invasive tests and procedures, your doctor can discover and take action on Fabry symptoms at an early stage. This brochure includes a description of the types of tests your doctor should conduct and a specific timetable for taking them. This timetable is called the schedule of assessments.

Developed by experts in treating Fabry Disease

The Fabry disease schedule of assessments was developed by the Fabry Registry, an organization that collects clinical data on patient outcomes to improve patient care. The Registry is comprised of specialists with years of experience and expertise in treating patients with Fabry disease. The Fabry Registry is a global program organized and sponsored by Sanofi Genzyme. The following pages include a conversation with one of these specialists, Dr. Christine Eng, as well as an overview of the recommended assessments.

THE IMPORTANCE OF REGULAR ASSESSMENTS

A Conversation with Dr. Christine Eng

Dr. Christine Eng is a medical geneticist at the Baylor College of Medicine. She specializes in the study of Fabry disease and was one of the developers of the schedule of assessments. We conducted a Q&A with her about these assessments and their value in monitoring and treating Fabry patients. The information provided is not intended to be a substitute for professional medical advice. You should always seek the advice of your physician or other qualified healthcare provider with any questions you may have regarding a medical condition.

Q: What is a scheduled assessment?

Dr. Eng: An assessment is just another word for a test. It could be a lab test, x-ray, physical exam or any type of procedure designed to provide key information about your health. Regularly scheduled assessments are especially important in managing a chronic, progressive condition like Fabry disease. As you probably already know, Fabry disease causes a wide range of symptoms that can vary from mild to severe to life threatening, and many of these symptoms tend to worsen over time.

By assessing or testing various organ systems and body functions at regular intervals, your doctor can monitor your health and manage symptoms before they become serious.

Q: As someone living with Fabry disease, why do I need to have regular assessments?

Dr. Eng: The schedule of assessments is designed to help your doctor monitor the organ systems that are known to have complications in Fabry disease. Many patients with Fabry disease do not have outward symptoms, but the disease can progress "silently" in the body. These assessments can uncover some of these problems, so your doctor can intervene while symptoms are still manageable. Of course, it can also be just as useful to know that your tests have come back normal. Then you can be reassured that you are taking the right steps to manage your disease.

Q: What if my doctor doesn't know about the assessments?

Dr. Eng: The schedule of assessments is especially useful for doctors who do not have a lot of experience with Fabry disease. While Fabry disease is rare, the types of assessments in the schedule are relatively common. Your doctor should be familiar with most or all the recommended tests and procedures. If your doctor does not have the schedule of assessments, he or she can download it from the Fabry Registry website, or you can download it yourself and provide it to him or her. It's one way you can help take control of your health and Fabry disease.

Q: Why are some assessments required more frequently than others?

Dr. Eng: It is primarily based on the rate of change—the pace at which Fabry symptoms tend to worsen.
For instance if an echocardiogram (a test that uses sound waves to get a picture of the heart) was normal one year, a year's time is an adequate interval to catch a problem that may be developing. On the other hand, proteinuria (protein in the urine, which is a sign of problems with the kidneys) is something that can climb from zero to a problematic finding at a much faster rate. So kidney function tests are recommended at more frequent intervals.

Q: What happens if I skip an assessment?

Dr. Eng: While you shouldn't feel overly guilty about missing one or two assessments, you should try to keep as up-to-date as possible. This is increasingly important as you get older and the disease progresses. You don't want to miss the chance to intervene and possibly avoid a more serious complication.

Q: Let's say I am a woman with Fabry disease and am not experiencing any symptoms. Do I still need to follow the schedule of assessments?

Dr. Eng: Yes. Fabry can take a rather stealthy or secretive course, compromising your organ systems even if you are feeling well. While an outward lack of symptoms is reassuring, it is not an accurate reflection of what is going on internally. The assessments provide a way to check and see if you are truly as healthy as you feel.

ASSESSMENT OVERVIEW

Q: What kinds of tests will I be given?

Dr. Eng: There are several different kinds of tests or assessments. Some involve just talking to you and getting a medical history. Others include a complete physical exam, lab tests for kidney function, tests to monitor your heart and breathing and even tests that measure your quality of life.

Q: Why are there different tests for people under and over the age of 18?

Dr. Eng: Fabry disease has different manifestations, or symptoms, depending on your age. For example, children with Fabry tend to experience more pain, while adult patients have more kidney and cardiac issues. The assessment schedules reflect these differences in the course of the disease.

Q: Why doesn't the adult assessment schedule specifically mention gastrointestinal symptoms, pain, sweating and heat intolerance?

Dr. Eng: These symptoms are more prevalent in younger patients. In addition, there aren't any objective tests that monitor or measure those things. However, these kinds of symptoms can be discussed with your doctor when your medical history is taken.

Q: Why are these specific tests recommended?

Dr. Eng: The assessments measure the organ systems and other areas of the body that are most commonly affected by Fabry disease. The list on the next page gives an overview of some of these tests.

Kidney Function Tests

By early adulthood many people with Fabry disease will have significant kidney problems, sometimes requiring dialysis.

- **Glomerular Filtration Rate:** a test to measure your level of kidney function and determine your stage of kidney disease
- **Tests for Albuminuria and Proteinuria:** conditions where there is too much protein in the urine. Excess protein in the urine is a sign of chronic kidney disease
- **Serum Creatinine and BUN** (blood urea nitrogen): these measures are closely related to kidney function
- **Urine Protein Excretion:** another test to detect protein in the urine

Heart Function Tests

Many Fabry patients may experience serious heart problems. Tests for these include:

- **Electrocardiogram (ECG):** a test that measures electrical activity in the heart
- **Echocardiogram (Echo):** a test that uses sound waves to get a picture of the heart
- **24-Hour Holter Monitoring:** a test that measures the heart's rhythms

Hearing Tests

Hearing tests are performed because many Fabry patients experience hearing loss or tinnitus, which is ringing in the ears.

Brain MRI

MRI stands for magnetic resonance imaging. It is a technique that uses a magnetic field and radio waves to create detailed images of the organs and tissues within your body. A brain MRI looks at portions of your brain to detect problems in the central nervous system, another common symptom of Fabry disease.

Eye Test

- Slit lamp exam. Nearly every person with Fabry disease has a distinct pattern on their cornea called corneal whorling or corneal verticillata. It doesn't affect vision and is only visible through a simple eye exam called slit lamp ophthalmoscopy. It is unique to people with Fabry (and people taking a certain heart medication) and is a good indication of Fabry disease.

THE TESTING PROCESS

Q: How long does it take to complete your testing?

Dr. Eng: Check with your doctor, but typically if you schedule your assessment far enough in advance, you should be able to complete your testing in a day or a day and a half. Of course, all the tests don't have to be done at the same time. As long as you follow the basic timelines, you can usually schedule your assessments when it's most convenient for you.

Q: Who performs the assessments?

Dr. Eng: Because there are multiple organ systems involved in Fabry disease, many specialists may be involved in your care. Initially, a geneticist or an internist can probably do the scheduled assessments and manage early complications. As your disease progresses, specialists in cardiac and kidney disease may need to be brought in. Often there is one physician who coordinates care between the different specialists.

Q: How do I find out the results of my assessments?

Dr. Eng: It can be done in a couple of different ways. Your doctor may make an appointment with you to review the results in person, talk to you on the phone and/or send the results in the mail. If there is a finding that needs immediate attention, your doctor will contact you and determine a course of action.

Q: If my child has Fabry disease, what can I do to help make the testing process less stressful?

Dr. Eng: These assessments can be a scary process for children. Fortunately, the tests are non-invasive and generally painless, other than the small needle prick of a blood test. However the hospital environment itself can be traumatic and a test like an MRI, while not painful, can be frightening. If possible, it can help to go to a children's hospital, where caregivers have a lot of experience in helping families and children dealing with chronic disease.

Q: Are the tests covered by insurance?

Dr. Eng: All of these assessments are considered standard of care and may be covered by your insurance company. That's one reason we came up with a standard schedule of assessments. If your insurance company will not pay for testing, you can resubmit your claim with the schedule as a clinically supported rationale for coverage.

FABRY REGISTRY SCHEDULES OF ASSESSMENTS

The following schedules are abbreviated versions of the recommended assessments for Fabry patients under and over the age of 18. Complete versions of these schedules can be downloaded at fabrycommunity.com.

Schedule of Assessments for Patients Under 18 Years of Age

	Upon Diagnosis	Every 6-12 months	Every 24-36 months	At time of a specific event or change in therapy
GENERAL				
Medical history including gastrointestinal symptoms, pain, sweating and heat and cold intolerance	✔	✔		✔
Family history	✔		✔	
Physical exam, including measurements of height, weight and blood pressure	✔	✔		✔
Quality of life	✔	✔		✔
LAB TESTS				
Kidney function test	✔		✔	✔
Test for protein in the urine	✔	✔		✔
OTHER STUDIES				
Hearing test	✔		✔	✔
MRI of the brain	✔		✔	✔
Cardiac function tests	✔		✔	✔
Eye exam	✔		✔	

Schedule of Assessments for Patients 18 Years of Age and Older

	Upon Diagnosis	Every 6 months	Every 12 months	Every 24-36 months	At time of a specific event or change in therapy
GENERAL					
Medical history	✔	✔			✔
Family history	✔			✔	
Physical exam, including measurements of height and weight	✔	✔			✔
Quality of life	✔	✔			✔
LAB TESTS					
Kidney function tests	✔	✔			✔
Test for protein in the urine	✔	✔			✔
Test for fats in the blood	✔		✔		
OTHER STUDIES					
Hearing test	✔			✔	✔
MRI of the brain	✔			✔	✔
Cardiac function tests	✔		✔		✔
Test of lung function	✔			✔	
Eye exam	✔				

Learn More

There are many resources available to learn more about Fabry disease and to connect with others affected by it. Below are some useful online resources.

Online Resources:

Sanofi Genzyme does not review or control the content of non-Sanofi Genzyme websites. Listing here does not constitute an endorsement by Sanofi Genzyme of information provided by any other organization.

www.fabry.org
www.fabrydisease.org
www.kidney.org
www.rarediseases.org

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