

Monitoring your Fabry disease

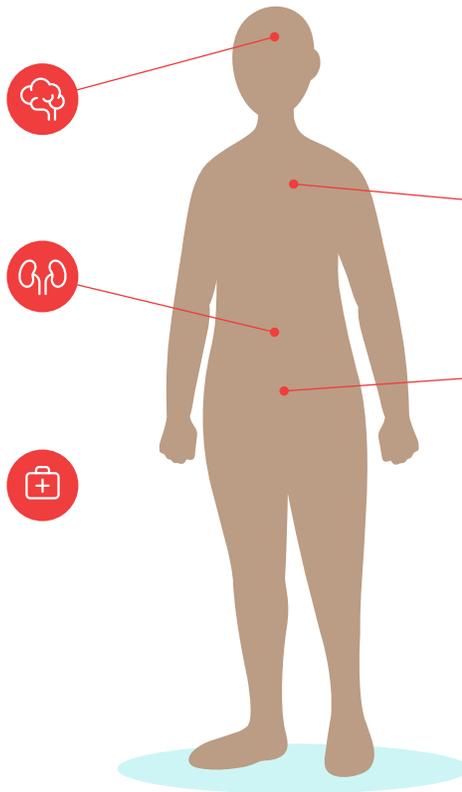
Understanding your test results and why they matter

Fabry disease is a complex condition that can affect many different parts of you, which can make it challenging to stay on top of all of its possible signs and symptoms. How are you supposed to know what to look for, or what to ask your doctor about? That's why it's so important to partner with a care team that includes doctors who specialize in different parts of the body. Your care team may include:

A **neurologist** to keep track of your pain, sweating, and other symptoms related to your nervous system

A **nephrologist** to see how well your kidneys are working and measure levels of protein and vitamins in your urine and blood

A **primary care physician** (PCP) to monitor your overall health. If you aren't sure which specialist to talk to, your PCP will be able to help



A **geneticist** to test if you inherited the disease

A **cardiologist** to monitor your blood pressure and any irregularities in your heart

A **gastroenterologist** to check for possible damage to your gastrointestinal system

Other specialists, depending on your signs and overall health

Together, these doctors make up your care team—they are all working together to keep you as healthy as possible. As part of that, they may order specific types of tests to help them keep track of, or monitor, what is going on in your body.

Why monitoring matters

Monitoring can help your care team stay on top of how your disease is progressing by identifying new signs early. Some signs of Fabry disease are difficult to notice without testing, but the earlier they are identified, the earlier they can be addressed—and the earlier you can get back to your life.

Your care team will monitor your lyso-Gb3 levels as standard practice. However, your care team may also want to perform other types of monitoring tests to keep track of your Fabry disease. Over the next few pages, you'll read about these tests. **These tests are recommended by an international panel of Fabry disease specialists, who published their recommendations in the medical journal *Molecular Genetics and Metabolism*.** You may not need all of these tests, but the tests that you do get will help your care team stay on top of your health. And, by tracking your results, you can ensure that you're an active participant in your health, too.

Fabry disease monitoring tracker – Neurologist



Brain- and nervous system–related monitoring

Fabry disease may cause your brain to not get enough blood, which could result in a stroke. There is also a risk of damage to white matter or to the peripheral nerve system. Brain scans will help your care team identify if this could be a concern for you.

These tests are recommended by an international panel of Fabry disease specialists, who published their recommendations in the medical journal *Molecular Genetics and Metabolism*.

Test Name	Frequency of Testing <i>How often I can expect to receive this test</i>	Notes <i>Any other relevant details—date of test, upcoming tests, doctor’s comments, etc</i>
Stroke		
Brain MRI <i>A test that creates images of your brain. Also used to look for white matter damage</i>	Every 3 years or as requested by your doctor	
CT imaging <i>A test that creates images of your body. Also used to look for white matter damage</i>	As requested by your doctor (in case of acute stroke and only if an MRI is not possible)	
Autonomic symptom evaluation <i>A blood pressure test that is done while you are lying down and standing</i>	Once a year or as requested by your doctor	
Blood pressure	At every visit or as requested by your doctor	Acceptable results: <120 (systolic)/<80 (diastolic) My results:
Cholesterol <i>A test that measures the cholesterol in your blood</i>	Recommended every 6 months if on ERT and once a year if not on ERT, or as requested by your doctor	Acceptable results: total cholesterol <200 mg/dL My results:



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Fabry disease monitoring tracker – Neurologist



Brain- and nervous system–related monitoring (continued)

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Pain		
Pain evaluation and history	Once a year or as requested by your doctor	
Cold and heat intolerance test <i>A doctor looks at how your body temperature changes while you are walking or jogging on a treadmill, versus when you are resting</i>	Once a year or as requested by your doctor	
Vibration test <i>A doctor uses a specific tool to test for feeling in your hands and/or feet</i>	Once a year or as requested by your doctor	



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Fabry disease monitoring tracker – Neurologist/ Other Specialists



Brain- and nervous system–related monitoring (continued)

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These tests are recommended by an international panel of Fabry disease specialists, who published their recommendations in the medical journal *Molecular Genetics and Metabolism*.

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White Matter Damage		
<p>Mood/overall quality of life</p> <p><i>A doctor will ask you questions to assess your overall mood and quality of life</i></p> <p>May also be performed by your primary care physician or psychologist</p>	<p>At every visit or as requested by your doctor</p>	
<p>Productivity</p> <p><i>A doctor will ask you questions or give you test(s) to assess how well you are able to perform everyday tasks such as work or school</i></p> <p>May also be performed by your primary care physician or psychologist</p>	<p>At every visit or as requested by your doctor</p>	
Other		
<p>Skin biopsy</p> <p><i>A doctor looks at a small piece of your skin under a microscope to detect lipid deposits</i></p> <p>May also be performed by your dermatologist</p>	<p>As requested by your doctor</p>	



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Fabry disease monitoring tracker – Nephrologist

Kidney-related monitoring

Over time, Fabry disease may cause your kidneys to not function as well as they should, which may lead to kidney disease and failure. Monitoring can help your care team find and respond to any changes to your kidney health early.

These tests are recommended by an international panel of Fabry disease specialists, who published their recommendations in the medical journal *Molecular Genetics and Metabolism*.

Test Name	Frequency of Testing <i>How often I can expect to receive this test</i>	Acceptable Results <i>Typical results in someone who does not have Fabry disease</i>	My Results <i>As given by my doctor or lab results</i>	Notes <i>Any other relevant details—date of test, upcoming tests, doctor's comments, etc</i>
Serum creatinine <i>A test that measures the creatinine in your blood</i>	Recommended every 6 months if on ERT and once a year if not on ERT, or as requested by your doctor	0.84-1.21 mg/dL		
Estimated glomerular filtration rate (eGFR) <i>A blood test that looks at how well your kidneys are functioning</i>	Recommended once a year if low risk; every 6 months if moderate risk; every 3 months if high to very high risk; or as requested by your doctor	≥90 mL/min/1.73 m ²		
Urine protein test <i>A test that looks for protein, including albumin, in your urine</i>	Recommended once a year if low risk; every 6 months if moderate risk; every 3 months if high to very high risk; or as requested by your doctor	Undetectable		
Calcitriol (25-hydroxyvitamin D) <i>A blood test that measures your vitamin D level</i>	As requested by your doctor	≥20 ng/mL (50 nmol/L)		
Kidney biopsy <i>A test that inspects a small sample of your kidney tissue</i>	As requested by your doctor	No sign of abnormal decline or disease		



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Fabry disease monitoring tracker – Cardiologist

Heart-related monitoring

Fabry disease may damage your heart, which may result in heart attacks or other heart-related health problems. Monitoring can help your care team identify and manage problems early.

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Test Name	Frequency of Testing <i>How often I can expect to receive this test</i>	Acceptable Results <i>Typical results in someone who does not have Fabry disease</i>	My Results <i>As given by my doctor or lab results</i>	Notes <i>Any other relevant details—date of test, upcoming tests, doctor's comments, etc</i>
Blood pressure	At every visit or as requested by your doctor	<120 (systolic)/ <80 (diastolic)		
Heart rhythm analysis	At every visit or as requested by your doctor	Findings may vary ^a		
Electrocardiography (ECG) <i>Electrocardiography records the electrical activity of your heart</i>	Once a year or as requested by your doctor	Findings may vary ^b		
Holter monitoring <i>A portable device that monitors your heart for 24-48 hours</i>	Once a year or as requested by your doctor	Findings may vary ^c		
Echocardiography <i>An ultrasound of your heart</i>	Once a year or as requested by your doctor	Findings may vary ^d		
B-type natriuretic peptide <i>A blood test that measures certain proteins to assess your heart health</i>	Once a year or as requested by your doctor	Reference ranges vary between labs		

^a http://heartrhythmguide.com/sinus_rhythms.php

^b <https://ecglibrary.com/norm.php>

^c <https://www.heart.org/en/health-topics/heart-attack/diagnosing-a-heart-attack/holter-monitor>

^d <https://www.heart.org/en/health-topics/heart-attack/diagnosing-a-heart-attack/echocardiogram-echo>



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Fabry disease monitoring tracker – Gastroenterologist



Gastrointestinal system–related monitoring

Over time, Fabry disease may damage your gastrointestinal system, which can cause problems like constipation, diarrhea, or bloating after eating. Gastrointestinal monitoring can find these problems early.

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Test Name	Frequency of Testing <i>How often I can expect to receive this test</i>	Notes <i>Any other relevant details—date of test, upcoming tests, doctor’s comments, etc</i>
Endoscopic evaluation <i>A test that looks at your intestines using an endoscope, a tube with a light and camera at the end</i>	As requested by your doctor	



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Fabry disease monitoring tracker – Other specialists

Lung-related monitoring

Fabry disease may damage your lungs, which may make breathing more difficult. Identifying changes in how well your lungs are working can help your care team manage possible problems.

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Test Name	Frequency of Testing <i>How often I can expect to receive this test</i>	Acceptable Results <i>Typical results in someone who does not have Fabry disease</i>	My Results <i>As given by my doctor or lab results</i>	Notes <i>Any other relevant details—date of test, upcoming tests, doctor’s comments, etc</i>
Spirometry (lung function test) <i>A test that measures how well your lungs are working by having you breathe into a machine</i>	Every 2 years or as requested by your doctor	<ul style="list-style-type: none"> · FVC (or forced vital lung capacity) of >80% of the predicted value based on your height and weight · FEV₁ (or forced expiratory volume in 1 second) of >80% of the predicted value based on your height and weight · FEV₁/FVC of >70% 		

Vision-related monitoring

Fabry disease may damage your eyesight over time. Monitoring your vision regularly can help your care team identify any changes.

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Test Name	Frequency of Testing <i>How often I can expect to receive this test</i>	Notes <i>Any other relevant details—date of test, upcoming tests, doctor’s comments, etc</i>
Slit lamp exam <i>A test to examine your eyes</i>	As requested by your doctor	



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