

# Monitoring Your Health

A Guide for Patients with Fabry Disease



sanofi

# Monitoring your health: For patients with Fabry Disease

Fabry disease is a condition that can affect the whole body. Below are some common tests to monitor your symptoms and your condition. You should consult with your healthcare provider regarding any specific questions you have about your medical care.

	<b>Laboratory Tests</b>	<ul style="list-style-type: none"> <li>• A Lipid Panel measures various levels of fats in the blood</li> <li>• Plasma Globotriaosylceramide (GL-3) measures the level of a particular type of fat in the blood that is of interest in Fabry disease</li> </ul>
	<b>Kidney Function Tests</b>	<p>Blood chemistry tests include:</p> <ul style="list-style-type: none"> <li>• Serum Creatinine: a waste product that comes from normal wear and tear on your muscles</li> <li>• Blood Urea Nitrogen (BUN): a waste product from the breakdown of protein in your food</li> <li>• Glomerular Filtration Rate (GFR) measures how well the kidneys remove waste and excess fluid from your blood</li> </ul> <p>Urine Protein Tests compare the presence of specific proteins using ratios</p>
	<b>Ophthalmology Evaluation</b>	<p>A microscope with a bright light (slit lamp) is used to look at the front and inside of your eyes. This examination checks for:</p> <ul style="list-style-type: none"> <li>• Corneal whorling</li> <li>• Changes in the arteries supplying blood to the eye</li> <li>• Cataracts, or clouding of the lens</li> </ul>
	<b>Audiology Evaluation</b>	<p>Audiology tests to examine hearing loss may include:</p> <ul style="list-style-type: none"> <li>• Tympanometry: tests eardrum movement by recording changes in air pressure</li> <li>• Otoacoustic emissions: tests how well sounds are heard at different frequencies</li> <li>• Auditory evoked potentials: tests your brain's response to sound</li> </ul>
	<b>Brain MRI</b>	Creates detailed pictures of your brain to look for changes that may relate to a stroke
	<b>Cardiac Function Tests</b>	<p>Routine assessments to evaluate cardiac function include:</p> <ul style="list-style-type: none"> <li>• Echocardiogram (ECHO): an ultrasound of your heart that measure its size and how it pumps</li> <li>• Electrocardiogram (ECG): measures the electrical activity of your heart</li> <li>• Holter Monitor: a portable device that tracks your heart's activity over a period of time</li> <li>• Cardiac MRI: creates detailed pictures of your heart</li> </ul>
	<b>Lung Function Test/ Spirometry</b>	A lung function test measures how much and how forcefully you can blow out air from your lungs in one breath. To see how well your lungs are functioning, you will be asked to take in a big breath, and then blow as hard and long as you can into a machine called a spirometer.
	<b>Quality of Life</b>	Surveys to assess quality of life may include the SF36® Health Survey, EuroQOL, or PedsQL® Measurement Model
	<b>Pain Evaluation</b>	Measures the presence and severity of pain using the following scales: BPI, Fabry Specific Pain and QOL Questionnaire and the Neuropathic Pain Symptom Inventory
	<b>GI Symptom Monitoring</b>	Your doctor monitors and records gastrointestinal (GI) symptoms such as abdominal pain, bloating, diarrhea, nausea, vomiting, quickly feeling full, and difficulty gaining weight
	<b>Other</b>	Other assessments determined by your healthcare provider

	Result at Baseline	Date and Result	Date and Result	Date and Result	Date and Result	Date and Result
Date of Assessment						
<b>PATIENT INFORMATION</b>						
DNA Analysis or Enzyme Assay						
<b>GENERAL</b>						
Height						
Weight						
Body Mass Index (BMI)						
Blood Pressure						
Heart Rate						
<b>LABORATORY TESTS</b>						
<b>BLOOD TESTS</b>						
Lyso-GL-3						
Lipid Panel						
<b>Kidney Function Tests</b>						
Serum Creatinine						
Blood Urea Nitrogen (BUN)						
Glomerular Filtration Rate (GFR)						
<b>URINE TEST</b>						
Urine Protein Tests (total protein/creatinine and albumin/creatinine ratios)						
<b>CLINICAL ASSESSMENTS</b>						
Ophthalmology Evaluation						
Audiology Evaluation						
Brain MRI						
Echocardiogram (ECHO)						
Electrocardiogram (ECG)						
Holter Monitoring						
Cardiac MRI						
Lung Function Test or Spirometry						
Quality of Life						
Pain Evaluation						
GI Symptom Monitoring						
<b>OTHER TESTS</b>						

Sanofi does not provide medical advice, diagnosis, or treatment. The health information contained herein is provided for general educational purposes only. Your healthcare professional is the best source of information regarding your health. Please consult your healthcare professional if you have any questions about your health or treatment.

# Schedule of Assessments

The Schedule of Assessments helps you and your doctor monitor your Fabry disease over time. Depending on your individual medical needs, your healthcare team will determine which tests you should have and how often.

	Younger than 18 years old			18 years old or older		
	BASELINE	EVERY 12 MONTHS	EVERY 24-36 MONTHS	BASELINE	EVERY 12 MONTHS	EVERY 24-36 MONTHS
<b>PATIENT INFORMATION</b>						
<b>Confirmation of Diagnosis: DNA Analysis or Enzyme Assay</b>	X			X		
<b>Family History</b>	X <sup>1</sup>			X	X <sup>2</sup>	
<b>GENERAL</b>						
<b>Height</b>	X	X <sup>2</sup>		X	X <sup>2</sup>	
<b>Weight</b>	X	X <sup>2</sup>		X	X <sup>2</sup>	
<b>Body Mass Index (BMI)</b>	X	X <sup>2</sup>		X	X <sup>2</sup>	
<b>Blood Pressure</b>	X	X <sup>2</sup>		X	X <sup>2</sup>	
<b>Heart Rate</b>	X	X <sup>2</sup>		X	X <sup>2</sup>	
<b>LABORATORY TESTS</b>						
<b>BLOOD TESTS</b>						
<b>Lyso-GI-3</b>	X	X <sup>3</sup>		X	X	
<b>Lipid Panel</b>				X	X	
<b>Kidney Function Tests</b>						
<b>Serum Creatinine</b>				X	X	
<b>Blood Urea Nitrogen (BUN)</b>				X	X	
<b>Glomerular Filtration Rate (GFR)</b>	X	X		X	X <sup>4</sup>	
<b>URINE TEST</b>						
<b>Urine Protein Tests (total protein/creatinine and albumin/creatinine ratios)</b>	X	X		X	X <sup>5</sup>	
<b>CLINICAL ASSESMENTS</b>						
<b>Ophthalmology Evaluation</b>	X <sup>6</sup>			X <sup>6</sup>		
<b>Audiology Evaluation</b>	X	X <sup>7</sup>		X	X	
<b>Brain MRI*</b>				X <sup>8</sup>		X
<b>Echocardiogram (ECHO)</b>	X <sup>7</sup>		X		X <sup>6</sup>	
<b>Electrocardiogram (ECG)</b>	X <sup>7</sup>		X		X <sup>6</sup>	
<b>Holter Monitoring</b>	X <sup>6</sup>				X <sup>6</sup>	
<b>Cardiac MRI<sup>13</sup></b>				X		X <sup>12</sup>
<b>Lung Function Test or Spirometry</b>				X		X
<b>Quality of Life<sup>9</sup></b>	X	X <sup>2</sup>		X	X <sup>2</sup>	
<b>Pain Evaluation<sup>10</sup></b>	X	X <sup>2</sup>		X	X <sup>2</sup>	
<b>GI Symptom Monitoring<sup>11</sup></b>	X	X <sup>2</sup>		X	X <sup>2</sup>	

**Note:** Clinical assessments and frequency may vary due to the onset of new symptoms, a medical event, or where there is a clinical indication.

1. Provide genetic counseling as they mature
2. Every clinic visit
3. Monitor annually in males, every 2-3 years in females
4. Check eGFR annually if low risk, every 6 months if moderate risk, and every 3 months if high or very high risk
5. Test Urine Protein annually, every 6 months if moderate risk, and every 3 months if high or very high risk; microalbuminuria can be used for pediatric patients
6. As clinically indicated
7. Every 2 years evaluations starting at age 10 and as clinically indicated
8. Regular evaluations in males starting at age 21; females starting at age 30
9. Quality of Life Survey (SF36® Health Survey, EuroQOL, or PedsQL® Measurement Model)
10. Pain Evaluation (BPI, Fabry Specific Pain and QOL Questionnaire, or Neuropathic Pain Symptom Inventory)
11. GI symptoms (abdominal pain, bloating, diarrhea, nausea, vomiting, feeling full early, difficulty gaining weight) and endoscopic studies if indicated
12. Whenever there is evidence of clinical progression of disease or at an interval of 2-3 years
13. Cardiac MRI is recommended in patients <25 years if cardiac hypertrophy or significant arrhythmia is present. For adults 25+ years, Cardiac MRI is recommended at the time of diagnosis. If the first MRI is abnormal, patients with moderate or severe left ventricular hypertrophy (LVH) should undergo cardiac MRI at least every 2 years or at a frequency depending on cardiac disease severity and physician clinical judgment. Males with no or mild LVH should undergo MRI every 2 years.

## Additional studies may be recommended for individuals with more cardiac or renal involvement:

Brain Natriuretic Peptide (BNP), Bone Density, Loop Recorder, Vitamin D

\*If an MRI is contraindicated, a CT scan may be recommended

# Monitoring Your Health

Since you have Fabry disease, your healthcare providers will likely want to see you regularly to monitor your health. Fabry disease is progressive, meaning it can get worse over time. Therefore, it is important to track your health and let your healthcare providers know if you have any new or worsening symptoms.

## Just a phone call or email away

Whether your needs are large or small, your CareConnectPSS® team will work closely with you and your family to ensure you receive the confidential and personalized support you need. To learn more about our range of support offerings or to reach a member of your CareConnectPSS team, please call **1-800-745-4447** and select **Option 3** or email us at **Info@CareConnectPSS.com**

For more information, visit us at **www.CareConnectPSS.com**

