Fabry Disease Recommended Schedule of Assessments

The Recommended Schedule of Assessments represents the core Fabry disease-related assessments that allow evaluation of a patient's disease progression over time. Physicians will determine the actual frequency of necessary assessments according to a patient's individualized need for medical care and routine follow-up.

These recommendations were developed by the Fabry Registry Board of Advisors, a group of physicians who have experience in managing patients with Fabry disease. The Fabry Registry is sponsored and administered by Sanofi Genzyme.

Pediatric Patients (Under 18 Years of Age*)

	Upon Enrollment	Every 6 – 12 months[a]	Every 24-36 months	At time of an event or therapy change
GENERAL				
Medical History, with particular focus on:				
Gastrointestinal Symptoms				
Pain	■	■		■
Sweating				
Heat & Cold Intolerance				
Family History	■		■	
Physical Exam	■	■		■
Vital Signs, Height and Weight	■	■		■
Blood Pressure[b]	■	■		■
Enzyme Activity and Genotype	■			
Concomitant Medication Assessment	■	■		■
Pediatric Quality of Life Assessment – PedsQL™ Pediatric Quality of Life Inventory	■	■		■
Pediatric Quality of Life Assessment – PedsQL™ Multidimensional Fatigue Scale	■	■		■
Pediatric Pain Assessment – PedsQL™ Pediatric Pain Questionnaire™	■	■		■
LABORATORY TESTS				
Glomerular Filtration Rate (GFR)[c]	■		■	■
Albuminuria and Proteinuria[d]	■	■		■
OTHER STUDIES				
Audiologic Evaluation[e]	■		■	■
Cranial MRI – T1, T2 and FLAIR	■		■ [f]	■ [f1]
Electrocardiogram[g]	■		■	■
Echocardiogram[h]	■		■	■
Cardiac MRI[i]	■		■	■
Ophthalmology – Slit Lamp Exam[j]	■		■	

* Initiation of Laboratory Tests, Imaging, and Other Studies: There is variability in the clinical complications and progression of Fabry disease. Children are at risk for life threatening complications. There are no biomarkers available to discern mildly affected from severely affected patients. In children with a family history of early presenting or severe disease, complete evaluations should be done at the time of diagnosis. Other patients should be completely evaluated at no later than 5 years of age.

[a] Patients are recommended to undergo these evaluations every 6 months; for those with milder disease, once per year may be sufficient

[b] Blood pressure should be measured 3 times at each assessment; only the last 2 measurements should be recorded.

[c] GFR should be measured directly every 24-36 months until age 15, and annually thereafter. If direct measurement is not possible, serum creatinine levels should be obtained at the recommended intervals for an estimation of GFR, which is a less sensitive method.

[d] First morning voided urine for protein, albumin and creatinine in order to calculate a protein/creatinine ratio and albumin/creatinine ratio. Protein, albumin, and creatinine measurements can also be performed on timed samples (e.g. 24 hours).

[e] Audiologic evaluation should be performed at the earliest age that is practical.

[f] Cranial MRIs should be performed at ages 10, 15, and 18 years.

[f1] At the time of a cerebrovascular event, a cranial MRI should also include diffused weighted images and apparent diffusion coefficient (DWI/ADC).

[g] Electrocardiogram should be performed starting at age 10–12 years. If abnormal and/or clinical symptoms arise, Holter monitoring is recommended.

[h] Echocardiogram should be performed starting at age 10–12 years.

[i] Cardiac MRI is recommended to be performed in patients under age 25 if cardiac hypertrophy or significant arrhythmia is present.

[j] Monitor yearly if retinal vessel tortuosity noted.

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Fabry Disease Recommended Schedule of Assessments

Adult Patients (≥18 Years of Age)

	Upon Enrollment	Every 6 months	Every 12 months	Every 24-36 months	At time of an event or therapy change
GENERAL					
Medical History	■	■			■
Family History	■			■	
Physical Exam	■	■			■
Vital Signs, Height and Weight	■	■			■
Enzyme Activity and Genotype	■				
Concomitant Medication Assessment	■	■			■
Quality of Life (SF-36®, BPI)	■	■			■
LABORATORY TESTS					
Serum Creatinine[a] and BUN	■	■			■
Urine Protein Excretion[b]	■	■			■
Lipid panel	■		■		
OTHER STUDIES					
Audiologic Evaluation	■			■	■
Cranial MRI – T1, T2 and FLAIR	■			■	■[c]
Electrocardiogram[d]	■		■		■
Echocardiogram	■		■		■
24-Hour Holter Monitoring[e]	■		■		■
Cardiac MRI[f]	■		■[f1]	■[f1]	■[f2]
Respiratory – Spirometry Exam[g]	■			■	
Ophthalmology – Slit Lamp Exam[h]	■				

[a] Directly measuring glomerular filtration rate (GFR) is recommended if a more precise evaluation is desired.

[b] 24 hour or first morning void urine for protein, creatinine and albumin.

[c] At the time of an event, a cranial MRI should also include diffused weighted images and apparent diffusion coefficient (DWI/ADC).

[d] If electrocardiogram is abnormal and/or clinical symptoms arise, Holter monitoring is recommended.

[e] Annual 24-hour Holter monitoring is recommended for males 30 years of age or older and females 40 years of age or older.

[f] Cardiac MRI is recommended at Fabry diagnosis for patients ages 25 and older. It is recommended to be performed under age 25 if cardiac hypertrophy or significant arrhythmia is present.

[f1] If first MRI is abnormal: 1) patients with moderate or severe LVH receiving ERT should have MRI annually; 2) patients with significant arrhythmia should have MRI at least every 2 years or at frequency factoring cardiac disease severity and the physician's clinical judgment; 3) males with no or mild LVH receiving ERT should have MRI every 2 years.

[f2] If first MRI is normal, repeat every 5 years or earlier if ECG/ECHO results are abnormal on annual exam.

[g] If spirometry is abnormal, perform yearly.

[h] Monitor yearly if retinal vessel tortuosity noted.

For more information on Fabry disease, contact Sanofi Genzyme Medical Information at 800-745-4447, option 2.

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