

Symptom Checklist

Date: _____

Fabry disease is a progressive, in some cases life-threatening inherited disorder caused by an enzyme deficiency, which can lead to serious complications such as stroke or kidney failure. If the signs and symptoms of Fabry disease are recognized early, treatment can begin early.

Complete the checklist below, noting any descriptions that may apply to you, as it may facilitate a discussion with your doctor. The bottom section includes signs and symptoms that only your physician may detect.

Signs and symptoms

- Burning, tingling pain in the hands and feet

How often have you experienced this type of pain in the last month? _____

Where did you feel the pain? _____

What does it feel like? _____

Are you taking any medication for this pain? _____

- Pain crises that radiate throughout the body

How often have you experienced pain crises in the past month? _____

Where? _____

How long did they last? _____

Are you taking any medication for this pain? _____

- Fever

How often have you had fevers (for no known reason) in the past month?

How long did they last? _____

- Impaired sweating: a decreased ability to sweat/perspire when exercising or exposed to warm temperatures

- A purple-red skin rash (known as angiokeratomas), generally in the area from the belly button to the knees

- Exercise intolerance

How often in the past month have you found yourself unable to participate in activities that involve exertion? _____

- Intolerance to heat and cold changes

How often in the past month have you found yourself affected by changes in temperature?

Please describe these episodes:

Gastrointestinal problems

How often in the past month have you experienced gastrointestinal problems such as pain, nausea, cramping, diarrhea, constipation, or vomiting?

Please describe these symptoms:

 Hearing problems

How often in the past month have you experienced hearing problems such as ringing in the ears or not being able to hear well?

Please describe these symptoms:

Family history

- Family relatives with kidney problems or who have died as a result of kidney failure, especially at a young age
- Family relatives with heart problems or who have died as a result of heart failure, especially at a young age
- Family relatives who have had a stroke, especially at a young age
- Known family history of Fabry disease

Signs and symptoms your physician may detect

This section should be completed by your physician. The following are symptoms of Fabry disease that only your physician will be likely to detect.

- Proteinuria (excessive protein in the urine)
- Increased serum creatinine levels
- Corneal whorling (does not usually affect vision, detectable only through slit lamp)
- Cardiac hypertrophy (enlarged heart)
- Mitral valve prolapse or other valvular abnormalities of the heart
- Evidence of transient ischemic attacks or stroke

About diagnosis and treatment

For information about testing for Fabry disease, or for information on treatment (enzyme replacement therapy) for Fabry disease, please contact Genzyme Medical Information at 800-745-4447 or 617-768-9000.