



**MASSACHUSETTS
GENERAL HOSPITAL**

**Evaluation of
Neuropathic Pain
in
Fabry's Disease**

Research Coordinators:

**Ezekiel Fink, M.D.
(617) 724-3260**

**David Chen, M.D.
(617) 724-8602**

Principal Investigator:

Anne Louise Oaklander, M.D., Ph.D.
Departments of Anesthesiology and
Neurology
MGH Pain Center
Massachusetts General Hospital

PATIENT INFORMATION BROCHURE

Evaluation of Neuropathic Pain in Fabry's Disease

What is Fabry's disease?

Fabry's disease is a fat storage disorder caused by an enzyme deficiency. Many body systems may be affected, including the eyes, brain, heart, kidneys, nerves, and skin. Enzyme replacement therapy (ERT) has been shown to improve some symptoms in Fabry's disease.

Neuropathic Pain in Fabry's

Pain is a common symptom of Fabry's disease and it is believed to be associated with loss of pain-sensing nerve fibers in the skin. Some describe this as a burning sensation in the hands and feet; other describe it as a shooting sensation. One study has suggested that treatment with ERT leads to decreased pain, but the mechanisms are still unknown.

What is the purpose of the study?

At MGH, we are trying to understand whether there is an improvement in pain and nerve injury with ERT.

We use 2 methods:

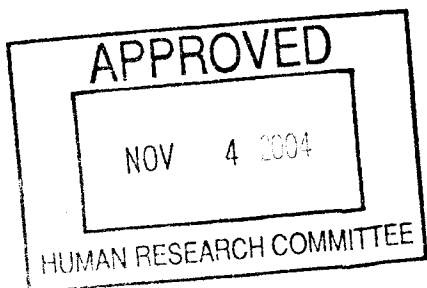
1. We examine the nerve endings in the skin by removing a small piece of skin using a local anesthetic.
2. We measure the skin response to histamine, an index of nerve function. This is a non-invasive test.

This project is *NOT* a treatment study. There are no medications or new procedures being tested. We will not stop or change any medications your physician has prescribed.

Who can participate in this study?

Adults, over age 18, who:

- ◆ have a confirmed diagnosis of Fabry's disease
- ◆ have or do not have symptoms of pain
- ◆ are not on blood-thinners
- ◆ do not have other problems affecting nerves, such as diabetes, HIV, or are undergoing chemotherapy treatment



What is required of the patient?

If you are eligible for this study, you will be asked to fill out a medical history form and bring it along with you to the appointment. We will also ask permission to review your medical records.

Subjects will be asked to come to the Massachusetts General Hospital for study (which lasts about 1 hour) every 9 months, as long as they are willing.

During each visit, one skin punch biopsy will be taken from an area above the ankle after it is numbed by local anesthetic. The piece of skin taken is about the size of the head of a pin. The biopsied area is covered with a sterile bandage that requires no care and will fall off after about a week.

Subjects will receive \$100 for each research visit.

Whom can I contact about the study?

If you are interested in more information about this research study, call Dr. Ezekiel Fink or Dr. David Chen, the Clinical Research Coordinators, at (617) 724-3260 or (617) 724-8602.

