

What is this study about?

We invite you to join a research study of neurofibromatosis type 1 (NF1) in adults at the National Institutes of Health (NIH). We are trying to answer a simple question: in a family with NF1, why do some family members have more serious disease than others? We hope to find genes that control the seriousness (or severity) of NF1. Finding those genes may allow doctors to predict the kinds of medical problems a person with NF1 might have.

Who can participate?

To be in our study, a person must:

- 1) Have a diagnosis of NF1.
- 2) Be willing to travel to the NIH in Bethesda, Maryland. We can pay for travel and lodging expenses.
- 3) Have gone through puberty. People in our study will be 16 years or older.
- 4) Be able to have an MRI of your spine (this involves lying still for about an hour).
- 5) Have both parents willing to be involved with the study. *It does not matter if they have neurofibromatosis type 1.* We need to ask your parents some medical questions and draw some blood.
- 6) Have a doctor to follow-up with you after your visit to NIH.

What is involved in the study?

If you are eligible for the study, we will:

- 1) Help arrange travel for you (and family members, in some cases) for a 2-day visit to NIH in Bethesda, Maryland.
- 2) Ask you questions about your medical history and do a physical exam (like in any doctor's office). In addition, we will take some pictures of your eyes and your skin, get an MRI of your spine and draw some blood. We will ask you to consider a skin biopsy. You will stay at a local hotel and *not* in the hospital.
- 3) Answer your questions about NF1. We can help you find doctors near you for follow-up.
- 4) Discuss our findings with you and share the results from your MRI of the spine. We will send you a letter summarizing your visit.

What is the cost of the study?

The visit and all testing are free and there is no cost to you, your family or your insurance company for this study.

What about travel?

We will pay your travel and parking expenses. If you live locally (near the NIH) we will cover your expenses to come to NIH for 2 days, including some meals. If you live further away, we will pay for your transportation to NIH (plane, car, train), hotel, meals and expenses. Travel must be arranged by NIH.

Will I get paid for joining the study?

No, participants in the study do not receive any compensation other than travel and lodging expenses.

Why should I join the study, or what's in it for me?

There may be no benefit to you, but hopefully the results from the study will benefit families with neurofibromatosis type 1. People join research studies for many reasons. In NF1, there is a long history of cooperation between patients and doctors. Animal models of the disease are helpful only up to a point.

Where can I learn more about the study?

You can read more about the study at 2 websites:

- 1) The NIH website:
http://clinicalstudies.info.nih.gov/cgi/detail.cgi?A_2005-HG-0152.html
- 2) The NHGRI website:
<http://www.genome.gov/16015146>

How do I enroll?

You may contact either Dr. Jennifer Sloan (genetic counselor) at jsloan@mail.nih.gov or Dr. Douglas Stewart (principal investigator) at drstewart@mail.nih.gov. Additional contact information is listed below.

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