Spinocerebellar Ataxia: Informed Consent for Presymptomatic Testing

University of Iowa
Division of Medical Genetics
Testing Protocols

Patient's Name: _____

Hospital Number: _____

Date: _____

I _____ choose to participate in presymptomatic testing for the presence of the Spinocerebellar Ataxia (SCA) gene. I have been told that I am at risk for SCA because there is a history of this disorder in my family. I understand genetic material will be evaluated only for the gene change responsible for SCA and will not be used for any other testing. This test will most likely tell me if I have or have not inherited the gene that causes SCA.

I understand I must participate in a testing protocol that provides genetic counseling and any necessary follow up care. I agree to participate in the minimum of three counseling sessions required for the testing. There will be at least two sessions prior to the test result session. Sessions will last 1-3 hours. These sessions will include information about SCA, genetic testing, consequences of results and available support services.

I understand a neurological exam is recommended. I am aware this examination may disclose that I have clinical signs of SCA and I will be told of my results.

I understand the test does have some limitations. First, it is not known if everyone who is affected with SCA has the same change in the gene causing the disease. Second, occasionally the result of gene analysis cannot be interpreted with confidence.

I understand there can be three outcomes to the test:

1. The gene change in my family might not be interpretable, so my test results will not tell me more than I know now.

2. I do not have the SCA mutation.

3. The SCA gene mutation is present and I will develop SCA.

I understand my participation in this program is voluntary and I can terminate my participation at any time without jeopardy to my medical care. I understand if at any time my continued participation in this program is considered injurious to my health or disadvantageous to me, the testing staff may recommend postponement of the testing.

The information provided is technical and receiving it will be stressful. Because of this, I am encouraged to bring a close friend, spouse, or other family member not at risk for SCA to accompany me through the test procedures. I understand the professionals involved in this testing are willing to answer my questions and to discuss my concerns with me throughout the testing procedure. Counselors will assist me in arranging for any additional professional counseling and support I may need.

Risks of this testing include:

A. Concerns of a psychological nature. I understand the testing may be lengthy and does require a waiting period before results are available. These events may cause me to confront difficult psychological issues. Since I may withdraw from the testing protocol at any time and the process is a long one, decisions about my continued participation may evoke anxiety. An un interpretable result can be frustrating and can intensify the ambiguity of the risk situation. Knowing I do not have the common mutation for SCA can produce mixed emotions. I may also have to confront the fact I have inherited the mutation that causes SCA and will develop the disease. I am aware the testing cannot predict when I first will show signs of SCA nor can it predict the severity or rate of progression of SCA. This will also mean that my children will have a 50% chance of inheriting the SCA mutation.

B. Possible difficulties with employment or insurance.

C. Discomfort of a blood draw and a bruise. I understand this test requires me to provide 20cc (two tubes) of my blood.

D. Financial liability. I will be responsible for the costs of the laboratory testing and counseling regardless of the outcome of my testing. In some cases, medical insurance will not pay for this testing.

I understand results of my testing will not be available to any one other than the professionals involved and is not to be released to any second party without my written consent. Test results will be given only to me and only in person.

_____ has explained to me the purposes of the testing protocol and I agree to participate in this testing program. I acknowledge the possible risks, discomforts, and benefits I may experience. I have read and I understand this consent form.

_____
Participant / Date

_____
Witness / Date