INFORMED CONSENT:
PARTICIPATION IN
GENETIC RESEARCH STUDIES
Introduction

This brochure provides information that can help you decide whether or not to participate in a genetic research project. It will tell you why it is important to think about this decision carefully. Questions you might want to ask the researcher before you make your decision are listed in the back of the brochure.

What is genetic research?

There are many different types of genetic research. Genetic research may be about finding genes (mapping), learning how genes work, or about treating or curing genetic conditions. To map a gene is to find its specific location, or a marker close by in your genetic code. Once a gene has been mapped, it is much easier to study the genetic basis of a condition. Scientists have to find out what the gene does (its function). They can then begin to develop tests for the gene and come closer to developing treatments and cures. Each genetic research project will have its own specific aims. These should be explained to you before you decide to participate in a project.

Genetic research projects often take place in hospitals or university medical centers. Before a project begins, it is usually reviewed by a committee called an Institutional Review Board or IRB. An IRB includes both scientists and non-scientists, such as clergy, community representatives, social workers, ethicists, lawyers, and nurses. The purpose of the IRB review is to assure that the interests of individuals participating in the research are well protected. However, the decision to participate is yours to make, and the informed consent process that precedes each study is designed to help you make your choice as freely as possible.
What is informed consent?

When researchers seek your consent, they are asking for your voluntary agreement to take part in a test, procedure, or research study. Informed consent means more than signing a written or printed consent form. This brochure is about informed consent for genetic research studies, and it also can help with informed consent for genetic tests and procedures.

To be informed, you need to know about benefits and risks of the research and how it may affect you, your family, and society. The research team should give you the information you need to make your own decision. Sometimes it is a good idea to talk about your decision with someone else not involved in the project. This person can be a family member, a good friend, or anyone you trust and respect and with whom you feel comfortable in discussing personal matters. Be sure to get the name and telephone number of a member of the research team you can contact in case you have any questions. In addition, the telephone numbers of other resources that you may wish to contact are listed in the back of this brochure.

If you think you do not have enough information to make an informed decision, or there is something you do not understand, ask questions. Keep asking questions until you do understand the project. Only when you are sure that you know what the research involves and how it affects you and your family should you give your consent. Make certain you carefully read any document given to you for your signature. Also, you should be given a copy of that form for your personal records.

Who benefits from genetic research?

Individuals and families participate in genetic research for many reasons. They may simply be interested in working with scientists to advance the understanding of a genetic disorder, without expecting immediate benefit to themselves. Other studies may offer the possibility of developing clinical tests or clues to treatments that can directly benefit the participants and their families. Sometimes, participating in a study can put you in contact with specialists and specialized care that may be otherwise inaccessible. A critical part of the informed consent process is a clear explanation to you by the research team of the purposes of a study and the benefits that you and your family might expect to gain from participating.

What are some of the risks of participating in genetic research?

Genetic information about you or other family members may be discovered during the research project. Genetic information about you may indirectly provide information about your entire family. One of the first steps in most genetic research studies is to draw a family tree (a pedigree) that also contains some medical information. Family circumstances such as paternity and adoption may be revealed. Consider discussing with relatives how you as a family would feel about knowing these things or allowing the information to be given to others. Decide for yourself whether or not you want this new information. It is possible that even a non-participating family member could somehow be made aware of certain information by a researcher or, more likely, by another family member in general conversation.

Another outcome of participating in a genetic research study is that you may have the opportunity to learn information about your own genetic status, such as your risk of developing symptoms of a genetic disorder. You would need to decide whether or not you want this information for yourself or made known to other family members.

Some family members do not want to participate in research or know about certain information that could be found during the research project. Even if they do not want to participate in the research project, this genetic risk information could also apply to them. If this is the case, it is important that these issues be
discussed with the researchers and a plan be worked out.

Information learned about you and your family, through your participation in genetic research, can become known to persons other than the research team. While researchers may try to protect you, no one can absolutely guarantee that at some point other researchers, insurance companies, employers, or other people will not get this information. You can ask for assurance that this information will not be put in your medical record. However, your insurance company will learn about it if you file a claim for any costs associated with this research project. Find out if your state has a law against discrimination based on genetic information.

There may be health-care costs to you for some types of genetic research. Your health insurance company may be unwilling to reimburse you for these costs. Inquire if your insurance company will pay for services related to the research project or experimental drugs. The informed consent agreement given to you by the research team should clearly spell out any costs which you may eventually have to pay.

Researchers will find it helpful to know why you or family members do not want to participate in their study, if this is the case. If you elect not to participate, your medical care and access to genetic counseling will not be affected. Ultimately, the decision as to whether or not to participate in the research study and whether to contact family members is yours.

Informed Consent:

Questions to Ask

The following is a list of questions that you might want to ask before deciding whether to participate in a genetic research study.

- **General Information**
  What is the purpose of the study? What are the names of the investigators? Who would be my contact person (and what is his or her phone number)? What agency is funding the research?

- **Benefits of Participating in Genetic Research**
  What are the benefits of participating in this research? For myself or family members? For others?

- **Risks of Participating in Genetic Research**
  What are the general risks of participating in this research? What physical risks may exist (beyond a blood sample)? What are some of the personal issues that could cause harm to me or my family (e.g., anxiety, stigma, discrimination, unpredictable disclosure of information)?

- **Treatment Issues**
  Will treatment be provided if unexpected problems arise while I am participating in the study? Who will pay for this treatment?

- **Support & Special Services**
  May I bring a friend or family member to help me, either while deciding to participate or while participating? Will special services be available for me if I need them (e.g., interpreters, braille, child care)?

- **Costs and Reimbursement**
  How will costs associated with participation in
this research be handled? Is there compensation for the time involved? Will the costs associated with travel/child care/special services be reimbursed? What additional health care costs may be associated with participation (e.g., will anything such as imaging scans and blood tests be billed to me or my insurance)?

☐ Storage of Genetic Information
What will happen to the stored DNA sample or any of my genetic information after this project is completed? What will happen if I decide to withdraw from this project? If this research plan changes in the future, if additional steps are added, or if new findings emerge, will I be notified and asked to sign another consent form? Will any of my genetic information be distributed (e.g., to pharmaceutical or biotechnology companies, genetic laboratories or government agencies)?

☐ Involvement of Other Family Members
What happens if you need to have other family members involved in the study? How will they be contacted and by whom? What will happen to cells, DNA, or personal genetic information if they choose not to participate at all or withdraw from the study?

☐ Study Results & Confidentiality Issues
What will happen to the results of my tests from the study? Will I receive them? If so, how? May I choose not to receive the results (can I change my mind)? Who will get them (will they be put anywhere except in the research records)? How will confidentiality of the records, including photographs, be maintained?

☐ Communication and Follow-Up
How will the results of the research project be communicated to participants? If genetic services, tests, or treatments are developed from this research, how will I be told about their availability? How will I be informed if you publish information about me and my family? What happens if I do not participate in the research?

Additional Resources

Alliance of Genetic Support Groups

(202) 966-5557
Ethical, Legal, and Social Implications Branch
The National Center for Human Genome Research
National Institutes of Health
(301) 402-0911

National Society of Genetic Counselors
(215) 872-7608

Office of the Director
Office For Protection from Research Risks
National Institutes of Health
(301) 496-7005