



# Making Sense of Your Genes

A GUIDE TO GENETIC COUNSELING

---

# Contents

What is genetic counseling?	1
Why might I see a genetic counselor?	1
How can I prepare for a genetic counseling visit?	2
Prenatal genetic counseling	5
General and pediatric genetic counseling	8
Psychiatric genetic counseling	11
Cancer genetic counseling	13
Resources	18
Acknowledgements	20

Information included herein is not professional advice and is not intended to replace the judgment of a practitioner with respect to particular patients, procedures, or practices. To the extent permissible under applicable laws, NSGC and Genetic Alliance disclaims responsibility for any injury and/or damage to persons or property as a result of any actual or alleged libelous statements, infringement of intellectual property or other proprietary or privacy rights, or from use or operation of any ideas, instructions, procedures, products or methods contained in the material therein.

---

## What is genetic counseling?

The goal of genetic counseling is to help you learn more about the causes of genetic conditions and how they affect you.

Genetic counselors can:

- Review your family and medical histories.
- Explain how genetic conditions are passed down through families.
- Figure out if you or your family members are at risk for disease.
- Find and give you information about genetic conditions.
- Offer guidance to help you make informed choices or life plans.
- Provide information about testing options and help you decide what is best for you and your family.
- Help you find referrals to medical specialists, advocacy and support networks, and other resources.

## Why might I see a genetic counselor?

You might see a genetic counselor in many situations, such as:

- You are pregnant or considering becoming pregnant and are concerned about the health of your baby (*See prenatal genetic counseling, page 5*).
- Your baby had an abnormal result from newborn screening (*See general and pediatric genetic counseling, page 8*).
- You, your child or a family member has been diagnosed with a genetic condition (*See general and pediatric genetic counseling, page 8*).



- You are concerned that you, your child, or a family member has a genetic or inherited condition and would like more information (*See general and pediatric genetic counseling, page 8*).
- Your family has a history of developmental disability, birth defects, and/or mental retardation (*See general and pediatric genetic counseling, page 8*).
- Your family has a history of mental illness (*See psychiatric genetic counseling, page 11*).
- Your family has a history of cancer (*See cancer genetic counseling, page 13*).



## How can I prepare for a genetic counseling visit?

Although steps to prepare for specific types of visits are provided later in this booklet, here are a few common areas to think about before your visit. Come to the visit with a list of questions you would like to ask. This will help the counselor focus on your concerns. Genetic counseling visits usually involve collecting family history information.

It can be useful to ask your relatives about what types of medical conditions occur in your family before your visit. If you have medical records relating to your concerns, you may want to bring them or ask your doctor to send them to the genetic counselor before your visit.

### WHAT CAN YOU EXPECT FROM YOUR VISIT?

At the beginning of the session, you and the genetic counselor should outline what to talk about in the session.

Common topics include:

- Talking about your family health history and ethnic heritage.
- Helping you understand the causes of genetic conditions.
- Helping you understand testing options, diagnosis, or, in some cases, the reason why no diagnosis has been made.
- Guiding you through decision-making about genetic testing, family planning, or medical planning.
- Helping you deal with emotions associated with having or not having a known genetic condition, having a relative with a genetic condition, or being at risk for a genetic condition.
- Finding supportive resources to help you manage a genetic condition.
- Understanding the chance of passing a genetic condition on to your children.

Your input is very important to the genetic counseling session; the details you provide will allow the genetic counselor to understand your health concerns fully.

**A genetic counseling session is a conversation.  
Your input is very important to the session.**

---

## QUESTIONS YOU MIGHT ASK YOUR GENETIC COUNSELOR

- Does the disease in question run in families?
- If my family member has a disease, might I get it?
- If I have a disease, are my family members at risk of getting it?
- Is any kind of genetic testing available? If so, what are the benefits and limitations of the testing? How will I pay for it?
- What kind of information can genetic testing give me?
- What does the genetic testing process involve?
- If I decide to have genetic testing for myself or my child, when can I expect to hear about the results? Will the results be given to me over the phone or in person?
- How can knowing more about a genetic risk help me?
- Could I be exposing myself or my family to discrimination based on genetic information?



---

## Prenatal genetic counseling

### WHAT IS PRENATAL GENETIC COUNSELING?

Prenatal genetic counselors work with individuals, couples, or families who have an increased chance of having a child with a birth defect or genetic condition. Those who are already pregnant or are considering having a child in the future can meet with a prenatal genetic counselor to learn more about the condition in question, understand their risks more clearly, and discuss options for prenatal screening, testing, and/or assisted reproduction techniques such as sperm and egg donation. During pregnancy, if a baby is found to have a birth defect or genetic condition you may be referred to a prenatal genetic counselor. The counselor will help the expecting couple understand the medical information, what to expect, and how to prepare for the birth of a child with special needs, as well as discuss options such as pregnancy termination or adoption.

Prenatal counselors also help many families who do not have an increased chance of having a child with a birth defect or genetic condition understand prenatal screening and testing options. Procedures such as blood tests and ultrasounds may be able to give a better idea if a developing baby has a chance of having birth defects or a genetic condition.

### WHY SEE A PRENATAL GENETIC COUNSELOR?

You and your partner:

- Are worried about a genetic condition or a disease that runs in your family
- Have a child who is affected with a genetic condition and are thinking about having another child in the future
- Have family members with mental retardation or birth defects
- Have a history of infertility or pregnancy losses (miscarriages or stillbirths)
- Are concerned that your health or lifestyle poses a risk to the pregnancy

- Are concerned about risks to the pregnancy associated with increasing parental age
- Receive abnormal prenatal screening or ultrasound results
- Are concerned that you are at increased risk of being a carrier of a genetic condition because of your ethnic background (some diseases are more common in certain ethnicities)
- Are pregnant and the baby has been diagnosed with a birth defect or genetic condition
- Have taken a medication or drug during pregnancy or have been exposed to a chemical and are concerned that it might cause a problem for the baby

### WHAT CAN I DO TO PREPARE FOR MY APPOINTMENT?

The counselor will ask some questions about your family and medical history, so you may want to prepare a few things before your appointment. Information to bring with you might include:

- Your (or your partner's) pregnancy history, including your current and past pregnancies. The counselor may ask about:
  - births and miscarriages
  - terminations or abortions
  - pregnancy complications
- Your (and your partner's) medical and health history:
  - major illnesses
  - chronic conditions such as diabetes or heart disease
  - medications you are taking
- The health history of other members of your and your partner's family, including:
 

- children	- aunts
- siblings, half-siblings, and their children	- uncles
- parents	- first cousins
	- grandparents





---

The counselor may ask if any of your relatives have had major illnesses or chronic conditions. If any of those relatives have died, the counselor will probably ask their ages and causes of their deaths.

The counselor may collect information about any miscarriages, stillbirths, and/or children who died young in either family. The counselor may also ask if anyone in the family was born with a birth defect or had developmental delays, learning disabilities, or mental retardation.



The counselor may also ask about your (and your partner's) ethnic background or from where your ancestors came (what country or part of the world). This is because some genetic conditions are more common in certain ethnic groups.

### WHAT WILL HAPPEN DURING MY APPOINTMENT?

Depending on the reason for the visit, some things a genetic counselor may do during an appointment are:

- Go over your family and medical history with you
- Figure out and explain your chances of having a child with specific genetic conditions
- Help you explore and make decisions about your options for screening and testing before and during pregnancy
- Help you interpret screening or testing results
- Help you understand medical and genetic information
- Provide you with information about any problems detected during pregnancy and help you understand your options
- Provide counseling and emotional support
- Refer you to support and advocacy networks

---

## General and pediatric genetic counseling

### WHY SEE A GENETIC COUNSELOR AT A GENERAL GENETICS CLINIC?

General genetics clinics serve children, adults, and families with known or suspected genetic conditions and birth defects. Some clinics serve both children and adults; whereas, others serve primarily children (pediatric genetics clinics) or adults (adult genetics clinics). A team of physicians and genetic counselors are available to diagnose and provide support and help for those with any genetic condition. Some families start out in a general genetics clinic and, if a diagnosis can be made, they may then be referred to a specialty clinic.

Some general genetics clinics provide outreach services to rural areas. If you are far from a major medical center, contact your nearest center and ask about outreach programs. Some outreach programs also specialize in prenatal services, in addition to the genetic consultation services provided in a general clinic.



---

## WHAT CAN I DO TO PREPARE FOR MY APPOINTMENT?

It is best to start preparing a few weeks in advance, if possible, so you have enough time to gather all of the information needed.

1. **Contact your insurance company** to find out if the genetic counseling consultation will be covered or if you may need to pay for some or all of the appointment.
2. **Find out** as much as you can about your family's health history. Talk to your family members and try to find medical information about your siblings, parents, aunts, uncles, cousins, grandparents, children, and grandchildren. To find out if a condition runs in the family, the genetic counselor will ask questions that relate to the problems for which the person was referred.
3. **Gather all of your medical records** and, if possible, for any family members who may also be affected by the particular genetic condition.
4. **Think about bringing someone**, either a friend or family member, with you for support and to help you remember all of the information that will be provided to you.
5. **Write down your questions** and concerns in advance. You may want to bring a notepad and pen to take notes during the meeting.

---

**If you do not live near a major medical center, contact your nearest center and ask about outreach programs.**

---



### WHAT WILL HAPPEN DURING MY APPOINTMENT?

The genetic counseling session can last an hour or longer, and the information covered will vary depending on the genetic condition. (More complex cases could take longer.) Depending on the reason for the visit, some key things may happen during the counseling session:

- If the patient, couple, or family was referred to the genetics clinic, the genetic counselor will review the reason for referral.
- The team will take a family health history of at least three generations, documenting all genetic conditions or health problems in each family member.
- A physician may provide a full physical examination and decide on laboratory tests to rule out or diagnose a genetic condition or refer you to other medical specialists.
- The genetic counselor can provide supportive counseling and information about resources or support networks.
- The genetic counselor will explain the diagnosis and any issues about the condition, including how the condition is expected to progress, the management of the condition, treatment options, whether genetic testing is available, and the chances of the condition being present in future pregnancies.

This may be done all in one counseling session or over the course of numerous sessions. The healthcare team may ask you to come back for follow-up appointments.

**The genetic counseling session can last an hour or longer, and the information covered will vary depending on the genetic condition.**

## Psychiatric genetic counseling



### WHY SEE A PSYCHIATRIC GENETIC COUNSELOR?

If you have a personal or family history of a psychiatric or mental health condition, the psychiatric genetic counselor will help answer questions you have about the cause of the condition and the chance it can happen again in your family.

These types of conditions may include autism spectrum disorders,

depression, bipolar disorder, schizophrenia, and others. If you are pregnant or considering pregnancy, the counselor will discuss the possibility of your children having a psychiatric condition, based on your personal or family health history. If you are pregnant and taking medications for a psychiatric illness, the genetic counselor can work with you and your mental health care provider to evaluate the safety of the psychiatric medication during pregnancy.

In general, genetic testing is not offered during psychiatric genetic counseling sessions.

### WHAT CAN I DO TO PREPARE FOR MY APPOINTMENT?

To get the most out of the appointment, try to provide as much information as possible about the psychiatric conditions in your family. Try to find out about:

- Your own mental health history and current status
- Who in your family has or had mental health conditions
- The name(s) of the condition(s) that run in your family, if known
- Ages that people in your family began having symptoms and were diagnosed

## WHAT WILL HAPPEN DURING MY APPOINTMENT?

The genetic counseling session will probably last about an hour, and what is covered during the session will be different for each person. General things that are likely to occur during your appointment include:

- The counselor will take a detailed family history. The more information you can gather, the better the counselor can study and share the type(s) and pattern of psychiatric conditions that may run in your family.
- Using the family history information, the counselor will talk about the likely cause of the illness in your family. Some people are interested in talking about risks for themselves, their children, or other family members to develop the condition or related conditions.
- The counselor will explain the cause and symptoms of the condition(s).
- Although genetic testing is not yet available for most psychiatric conditions, the genetic counselor will explore with you how likely it is that other family members might become ill.
- The genetic counselor may also discuss environmental risk factors in and outside of the home, what to look for in a young person at risk for the condition, and when to seek treatment.
- The genetic counselor can help you develop strategies for dealing emotionally with the risk in your family and the uncertainty related to whether a family member will or will not become ill.



---

## Cancer genetic counseling

### WHY SEE A CANCER GENETIC COUNSELOR?

Some kinds of cancers run in families. If you have had cancer at a young age, had two or more separate cancers, or have several family members that have had cancer, you may want to think about genetic counseling and perhaps genetic testing.

A cancer genetic counselor will evaluate your family health history and talk about risks for inherited cancer, as well as screening and management for those at increased risk. If genetic testing is available, the counselor will tell you about the tests and help you decide if testing would be useful to you.

If you have had cancer, genetic testing may be useful to you and your medical team for making decisions about cancer management.

If you have not had cancer, it can help you understand your risk for cancer and the risks for other members of the family.

In both cases, genetic testing may help you and your medical team make decisions about cancer screening and cancer prevention methods. It could also provide useful information for your family members about their cancer risks.

A genetic counselor can also refer you to support resources for people with cancer, an increased chance of cancer, or family histories of cancer.

---

**If you have had cancer at a young age, had two or more separate cancers, or have several family members that have had cancer, you may want to think about genetic counseling and perhaps genetic testing.**

---

---

## WHAT CAN I DO TO PREPARE FOR MY APPOINTMENT?

The counselor will ask you questions about your family's health history and your medical history. Here is some information that will help the genetic counselor work with you:

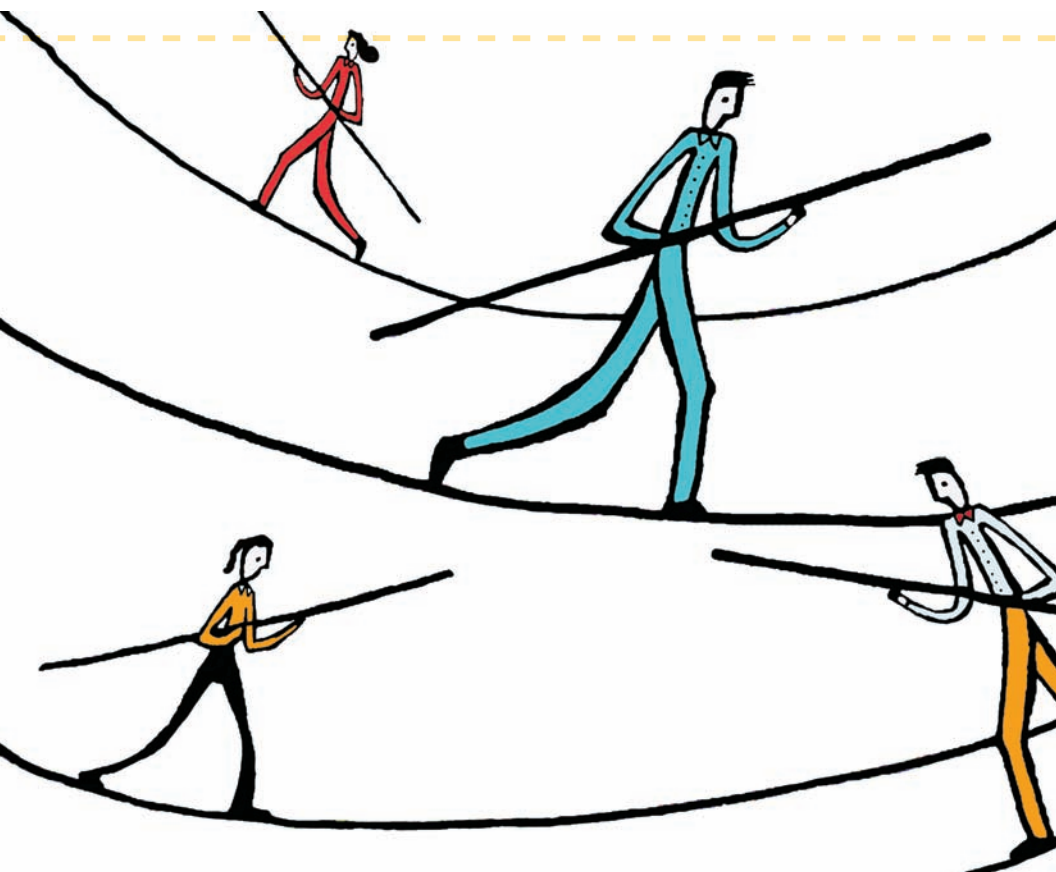
- S/he will ask about your own cancer experience, including type(s), part of the body affected, and age of diagnosis. To better understand your cancer history, the counselor is likely to ask for access to your medical records. You should bring any medical records or pathology reports related to your or your family's cancers or cancer treatment, if possible.
- Have any other members of your family had cancer, and what type of cancer did they have? Different types of cancer can run together in families.
- What was the age of diagnosis for members of the family with cancer? Generally, the younger a person is when a cancer occurs, the more likely s/he is to have a form of cancer that can run in the family.
- It may also be helpful to talk with your family about possible genetic tests.

---

Although testing is an individual decision, the tests can have consequences for members of your family that do not have cancer. For example, a test result might mean that some relatives have a higher risk for cancer than others.

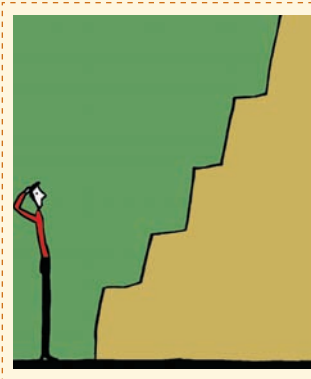
It might be useful for you to find out how your parents, siblings and children might feel about finding out information about inherited cancer risk.





Not all predispositions to cancer can be identified by a genetic test. Even if a genetic test is available for a certain type of cancer, genetic testing is not helpful for all individuals.

You might also want to contact your insurance carrier about whether or not genetic counseling and testing is covered under your policy. The counselor will have information about costs of the testing, but may not know what your insurance will cover. Often, the genetic counselor can work with you and your insurance provider to explain the medical importance of testing and determine your coverage before you go forward with testing.



### WHAT WILL HAPPEN DURING MY APPOINTMENT?

Every counselor has a different approach. But, in general, a few things will probably happen in the session.

- The counselor will go over your family health history with you. Some counselors might get this information from you ahead of time, and some will take the history with you in the session. The counselor will

ask you some questions about the health of your family members. This helps the counselor know what you might be at risk for, while using the information to create a family tree for your medical records.

- The counselor will talk to you about any cancers that you might be at risk for and the associated cancer screenings that you should do. This can be very useful, even if you decide against testing or if no testing is available. It can give you a good idea of what sort of screenings you should be doing and what you can do to reduce your risk. If you are considered to be at high risk, the counselor may talk to you about prophylactic surgeries (done to prevent a cancer, rather than treat one).
- If testing is available, the counselor will have information about cost and possible benefits and harms of the testing. The counselor will also help you explore what the results of testing will mean for you.

- Cancer genetic testing is different from some other types of testing because a positive test result does not always mean that you will get cancer. If you test positive for a variation in a cancer gene, it means you are more likely to get cancer.
- Not all types of cancer are the same—different genetic tests mean different risks for different types of cancer.
- If you decide to have a genetic test, the counselor will help you arrange for the testing. S/he will also receive the results of the test and will contact you to explain your test results.
- Depending on the counselor or institution, results may only be given out at another in-person meeting. Some counselors or institutions will give results over the phone.
- The counselor will also talk about the meaning of the results with you, help you plan to talk about the results with family members, and share the results with your medical team.

**Cancer genetic testing is different from some other types of testing because a positive test result does not always mean that you will get cancer.**

## Resources

Other types of genetic counseling specialties besides the ones listed in this book are available. These include:

- Assisted Reproductive Technology/Infertility Genetic Counseling (for couples experiencing infertility or who want to have genetic testing done on an embryo)
- Cardiovascular Genetic Counseling (for people concerned about their risk for various types of heart disease)
- Metabolic Genetic Counseling (for children, adults, and their families who have genetic conditions affecting the way they break down (metabolize) certain substances in their body)
- Neurogenetic Genetic Counseling (for people and families who have inherited neurological conditions)

To find a genetic counselor in your area, for information about a genetic counseling specialty, or for information about genetic counseling as a field, visit [www.nsgc.org](http://www.nsgc.org)

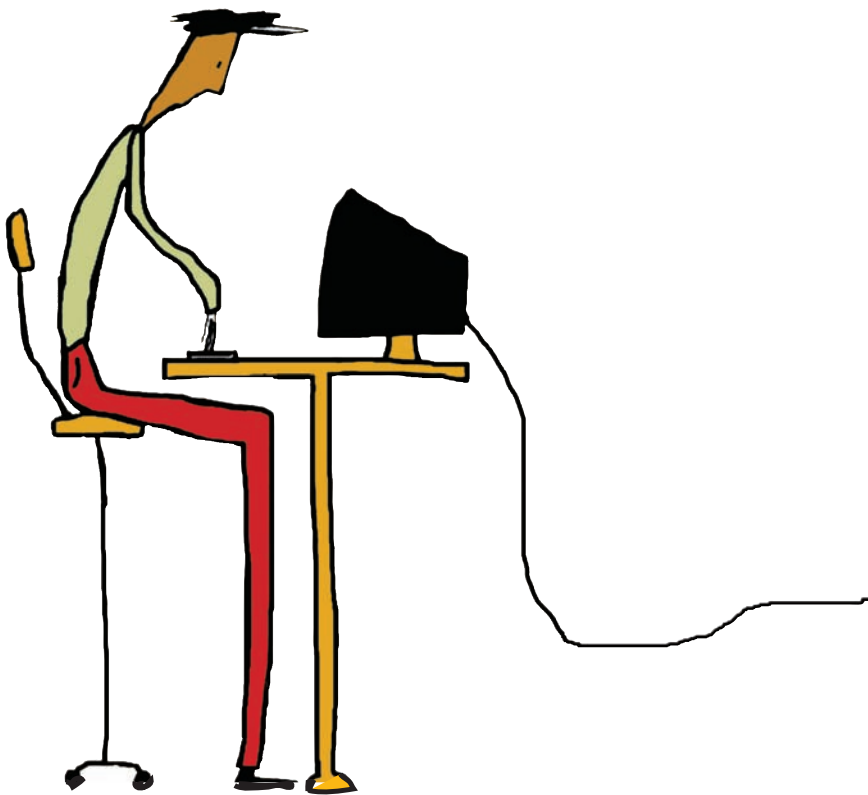


---

For more information on how to organize your family health history and construct a personalized family health history report, visit [www.familyhistory.hhs.gov](http://www.familyhistory.hhs.gov)

To learn about the Genetic Alliance Community Centered Family Health History project and access the *Does it Run in the Family* toolkit, visit [www.geneticalliance.org/ccfhh](http://www.geneticalliance.org/ccfhh)

This guide is available as a PDF file for free download at [www.geneticalliance.org/publications](http://www.geneticalliance.org/publications)



---

THIS GUIDE WAS PRODUCED AS A PARTNERSHIP BETWEEN THE JOHNS HOPKINS/NATIONAL HUMAN GENOME RESEARCH INSTITUTE, GENETIC ALLIANCE, AND THE NATIONAL SOCIETY OF GENETIC COUNSELORS.

THE JOHNS HOPKINS UNIVERSITY/NATIONAL HUMAN GENOME RESEARCH INSTITUTE GENETIC COUNSELING TRAINING PROGRAM

*Genetic Counseling class of 2008*

Melanie Baxter

Julie S. Cohen

Gillian Hooker

Mindy Simonson

Megan Truitt

Holly Peay, *Assistant Director*

GENETIC ALLIANCE

Sharon Terry, *Executive Editor*

James O'Leary, *Project Director*

Amelia Chappelle, *Project Director*

Heather Ferguson, *Associate Staff*

Rebecca Sample, *Intern*

Hilary Andreff, *Intern*

NATIONAL SOCIETY OF GENETIC COUNSELORS

Angela Trepanier, *Advisor and Reviewer  
President, 2008*

---

INDEPENDENT ADVISORS AND REVIEWERS

Lisa Schoyer  
*Costello Syndrome Family Network*

Michele Manion  
*The Primary Ciliary Dyskinesia Outreach Foundation*

Deborah A. Jacobs  
*Lowe Syndrome Association*

Kristy Colvin  
*International Mosaic  
Down Syndrome Association*

Kimberly Crawford  
*National Tay Sachs & Allied Diseases Association*

Sandra Prucka, MS CGC  
*University of Alabama-Birmingham*

Shonee Lesh, MS CGC  
*Rocky Mountain Cancer Centers*

Robin Troxell, MS CGC  
*University of Missouri-Columbia*

Kira Apse, ScM, CGC  
*Autism Consortium  
Harvard Medical School*

Deborah Axtell, MS CGC  
*Howard County General Hospital*

national society  
of genetic  
counselors, inc.



401 N. Michigan Ave. Chicago, IL 60611  
Phone: 312.321.6834 Fax: 312.673.6972 [nsgc@nsgc.org](mailto:nsgc@nsgc.org)



**Genetic Alliance**

4301 Connecticut Ave. NW, Suite 404, Washington, DC 20008-2369  
Phone: 202.966.5557 Fax: 202.966.8553 [info@geneticalliance.org](mailto:info@geneticalliance.org)

ISBN 978-0-9821622-0-0

Published 2008

Creative Commons Attribution-No Derivative Works 3.0 United States License