

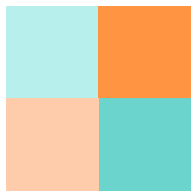
MAKING SENSE OF YOUR GENES

A GUIDE TO GENETIC COUNSELING



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WHAT IS "GENETICS"?

Genetics is the study of genes and inheritance.

- ✓ *Genes* are the instructions that tell the body how to grow and develop.
- ✓ Genes are found in structures called *chromosomes*.
- ✓ A person gets half of their genes or genetic information from their mother (the egg), and half of their genetic information from their father (the sperm). This means that they have two copies of each chromosome, and two copies of each gene, in each *cell* of their body.
- ✓ Each part of the body is made of many cells working together, like building blocks.
- ✓ The human *genome* is all the instructions in a cell to make a person.

Another way to think of this is:

A gene is like one recipe in a book of recipes.
A chromosome is like one book of recipes.
A genome is like a complete set of recipe books.



Gene

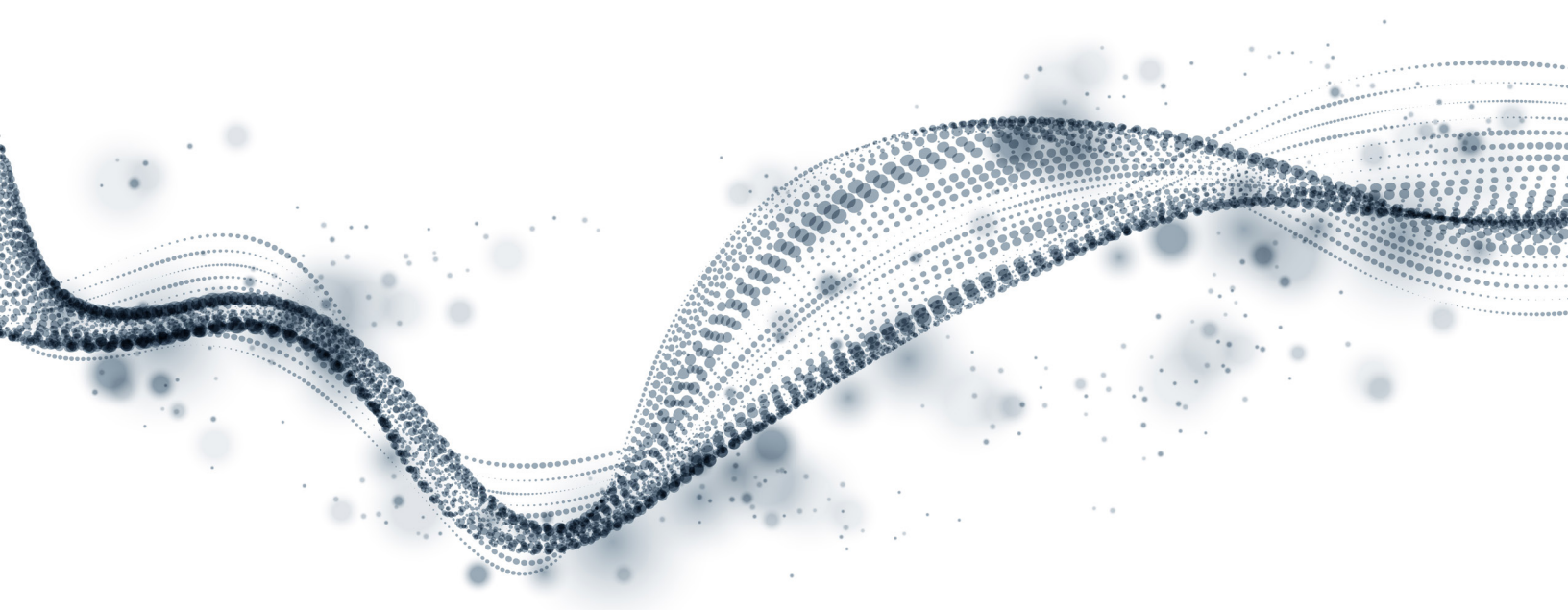


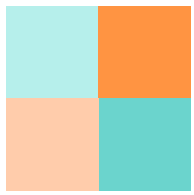
Chromosome



Genome

- ✓ *Inheritance* is how genes or genetic information can be passed from a parent to a child or from one generation to the next. We do not control the genes we inherit or that are inherited by our children.
- ✓ A person can have a *genetic condition* if there is a change in the way a gene works (also known as a mutation), a change in the structure of a chromosome (extra or missing pieces of chromosomes), or a change in the number of chromosomes (entire extra or missing chromosomes). How the genetic condition affects a person depends on what instructions for the body are changed.
- ✓ Using the recipe book example, a genetic condition can be thought of as a recipe with a change in ingredients or instructions, or a book of recipes missing several recipes, or an extra or missing book of recipes in the complete set of books.
- ✓ Some genetic conditions happen because a genetic change is inherited from one or both parents. Some conditions happen just by chance for the first time in that person. Other conditions may be due to both genetic changes and environmental factors.
- ✓ *Genetic testing* looks closely at a person's genes or chromosomes for changes that cause genetic conditions. The *results* or information learned from genetic testing are specific to each test.
- ✓ Using the recipe book example, genetic testing can be thought of as reading through the recipe to find the change in ingredients or instructions, looking through the recipe book for the missing recipes, or finding the missing or extra book in the complete set of books.





WHAT IS A GENETIC COUNSELOR?

A genetic counselor is a healthcare expert trained in genetics and counseling. A genetic counselor helps individuals or families understand information about genes, inheritance, and how a condition might be linked to a genetic change based on personal or family history. A genetic counselor also provides information about different genetic testing options. The goal of genetic counseling is to educate and support individuals wanting more information about their personal or family genetic information.

Genetic counselors work in different healthcare settings, including:



Preconception/
prenatal (before/
during pregnancy)



Many others



Cancer



Adult



Pediatric

Genetic counselors see patients in hospitals, private clinics, or through telehealth (video conferencing). Referrals to genetic counselors often come from doctors or nurses, but sometimes individuals can make appointments with a genetic counselor directly. It is common to be referred to a genetic counselor before or after having genetic testing or to discuss personal or family health history.

What to expect during a genetic counseling visit

Each genetic counseling session is unique. How long it lasts and what is discussed will depend on the reason for genetic counseling. Usually, a visit includes:

- ✔ Talking about questions, concerns, and goals of the person or family members
- ✔ Asking about personal and family medical history
- ✔ Going over options for genetic testing
- ✔ Giving information about helpful resources and support

Talking about questions, concerns, and goals

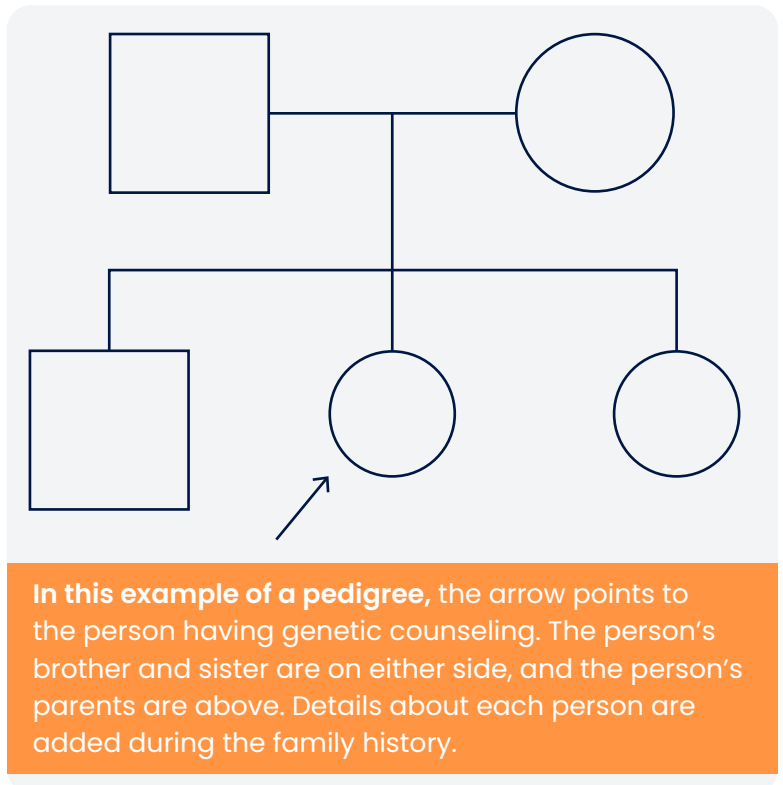
During a visit, a genetic counselor asks questions to learn what information feels most important to the individual or family members. This helps a genetic counselor to make a session fit each person's different needs. An appointment may cover a lot of information. Often there are questions that come up after a visit, so it can be helpful to discuss options for follow-up phone calls or an appointment with the genetic counselor.

Asking about personal and family medical history

A genetic counselor will often draw a person's *pedigree*, a picture with symbols representing different family members, when asking about personal and family health history. This history includes a person's brothers or sisters, children, parents, aunts, uncles, cousins, and grandparents. A pedigree is a way of looking at a family for patterns to see if a condition may be inherited.

A genetic counselor may ask if there is personal or family history of various health- or learning-related conditions. Some terms may be unfamiliar so it is important to ask for clarification if needed. Here are some of the things that may be asked about, along with other names that are sometimes used:

- ✔ Developmental challenges
 - ⦿ May also be called *intellectual, cognitive, or developmental disabilities*
 - ⦿ Examples: someone who has experienced lifelong difficulties in physical, learning, language, or behavior development. Autism spectrum disorders may also be included
- ✔ Physical differences at birth
 - ⦿ May be called a *birth defect or congenital disorder*



- ⦿ Examples: a cleft or opening in the lip, a hole in the heart needing surgery
- ✓ Long-term health conditions
 - ⦿ May be called *chronic health problems*
 - ⦿ Examples: diabetes, heart disease
- ✓ Mental or behavioral health
 - ⦿ May be called *mental health disorders*
 - ⦿ Examples: bipolar disorder, schizophrenia
- ✓ Genetic conditions
 - ⦿ May be called a *genetic syndrome or genetic abnormality*, and can include *chromosome conditions*
 - ⦿ Examples: sickle cell anemia, Huntington's disease, Down syndrome
- ✓ Death of a baby close to delivery or shortly after birth
 - ⦿ May be referred to as *stillbirth or neonatal death*
- ✓ Pregnancy loss or miscarriage
- ✓ Cancer

The genetic counselor will also ask questions specific to the reason for the visit.

When possible, it's best to gather this information from family members before the genetic counseling visit. It's also okay to not have all the details during the appointment; the information can be provided to the genetic counselor later.

Going over options for genetic testing

Genetic testing may come up during a genetic counseling visit, depending on the situation. This discussion usually covers:

- ✓ Results of genetic testing if already done
- ✓ What genetic testing options are available
- ✓ Reasons why someone might choose or not choose to do testing
- ✓ How the test is done, such as using blood or saliva
- ✓ When the test can be done, which could be on the same day or later if more time is needed to decide
- ✓ Communication and meaning of results
- ✓ The possible emotional impact of test results for the person and family
- ✓ What steps can be taken based on the test results

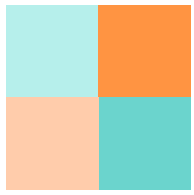
Whether or not to do genetic testing is a very personal choice based on a person's or family's unique needs. Genetic counselors support each person's choice to decide what is best for them. Some people might choose to do a genetic test because the results may guide medical care or treatment. Others may decide not to do a genetic test because the information at that time is not as helpful to them.

Common questions from patients if genetic testing is done or being considered:

- ✔ Will this test give a clear answer about what is happening in a person or their family?
 - ⦿ A genetic counselor can explain what a test can and cannot tell about a condition or its impact on a person. A test may rule out or diagnose a condition, provide chances for a condition, or give unclear results needing more tests. Unexpected information, like non-paternity, might also come up. Testing might be repeated in the future as technology and understanding of genetic conditions improve.
- ✔ How much does the test cost?
 - ⦿ Costs and insurance coverage of testing depend on the situation. A genetic counselor might have some helpful information, but sometimes the person needs to contact their insurance company for more details.
- ✔ When and how will the results be communicated?
 - ⦿ A genetic counselor can give an idea of when results will be ready. Results might come through a patient portal, email, over the phone, or at a follow-up appointment. It is possible that a person will receive their results as soon as they are ready, which may be at the same time as their genetic counselor. It is important to talk about what to expect and plan how to discuss the results before taking any further steps.
- ✔ Could genetic testing lead to genetic discrimination for a person or their family?
 - ⦿ The Genetic Information Nondiscrimination Act (GINA) is a U.S. federal law that protects against genetic discrimination at work and with health insurance. A link to more information about GINA can be found in the resources section or be discussed with a medical provider.

Giving information about helpful resources and support

A genetic counselor can help a family navigate the results of a genetic test or a diagnosis of a genetic condition, or the challenges of not getting a diagnosis. Some families need emotional support, some families are seeking helpful resources, and some are looking for both. Each person or family is different, and their needs may change over time. Useful information from a genetic counselor can include suggestions for websites, books, or articles that have trustworthy and accurate information, as well as support groups or networks related to a specific genetic condition. A genetic counselor can also provide guidance about social, ethical, and legal matters related to genetic test results. Depending on the condition, genetic counselors may be aware of opportunities to connect with other families locally or get involved in research on the condition.



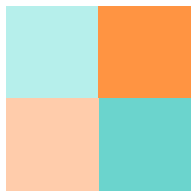
PRENATAL/PRECONCEPTION GENETIC COUNSELING

Anytime someone is pregnant, there is a chance their baby could have a genetic condition, physical difference, or developmental challenge. A prenatal genetic counselor helps explain these possibilities based on factors like a person's age, family history, or results of tests or ultrasounds during the pregnancy. People are often referred to a prenatal genetic counselor if any of these factors increases the chance of having a baby with a condition. It is also common to see a prenatal genetic counselor just because someone is pregnant, to go over all testing options available when expecting a baby. Genetic counselors also meet with those facing challenges getting pregnant or planning for future pregnancy (preconception).

What to expect during a prenatal/preconception genetic counseling visit

A genetic counseling visit may be done the same day as an ultrasound if a person is pregnant or may be scheduled on a separate day. Sometimes the visit involves meeting with other pregnancy specialists, such as a perinatologist or Maternal Fetal Medicine doctor.

During a prenatal/preconception genetic counseling session, questions include the age of the person who is pregnant or considering pregnancy, their partner's age, number of previous pregnancies, pregnancy losses, medications taken, illness during pregnancy, and results of prior ultrasounds or genetic testing. Based on the available information, a genetic counselor will review available tests for various conditions and what the results could mean. Testing can occur before, during, or after pregnancy, involving the person who is pregnant, their partner, or the pregnancy itself. Prenatal genetic counselors offer support and guidance to help individuals identify what information is most useful to them about their baby. This can include whether to do genetic testing or gain a better understanding of a known or suspected diagnosis during pregnancy.



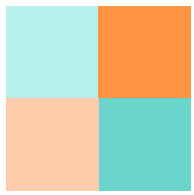
GENERAL, PEDIATRIC, AND ADULT GENETIC COUNSELING

Genetic counselors in general genetic clinics see children or adults with known or suspected genetic conditions. A genetic counselor often works with other members of a healthcare team, including a specialist known as a medical geneticist. A referral may be made to a general genetics clinic if a baby has abnormal newborn screening results or if there's suspicion of a genetic condition based on physical or developmental changes. Some clinics focus on children (pediatric genetics clinics), others on adults (adult genetics clinics), and some on specific conditions like skeletal or muscular disorders (specialty genetic clinics).

What to expect during a genetic counseling visit at a general genetics clinic

Visits to a genetics clinic often involve meeting with several members of the team. An appointment may include a full physical exam by the medical geneticist in addition to seeing the genetic counselor.

During a visit at a general genetics clinic, questions may include health concerns for the child or adult, family history of similar conditions, and results of prior tests or evaluations. Additional testing may be recommended and can involve looking at genetic changes or substances in the blood or other exams, like X-rays. The genetic counselor and medical geneticist can figure out the best tests to find a diagnosis in each situation. They can also provide information about what to expect with a condition and help families connect with other specialists. Sometimes testing does not find an answer and more visits are needed. Whether a diagnosis is known, suspected, or has yet to be found, a genetic counselor works with the family to meet their needs for information and support.



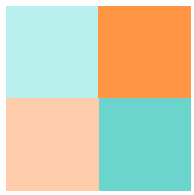
CANCER GENETIC COUNSELING

Cancer genetic counselors meet with individuals with personal or family histories of cancer. While most cancers occur randomly, some happen because of inherited gene changes, known as *hereditary cancer*. If someone inherits a gene change linked to cancer, they have a higher chance for certain cancer or cancers. A referral to a cancer genetic counselor is made when someone's history indicates a higher risk of hereditary cancer. Reasons for referral include early cancer diagnoses, multiple cancers in one person, or a rare cancer diagnosis. Cancer genetic counselors then assess how likely it is that an inherited gene change is involved in the risk of developing cancer and if genetic testing may be helpful.

What to expect during a cancer genetic counseling visit

Anyone with a history of cancer themselves or in their family can meet with a cancer genetic counselor. A cancer genetic counselor may see someone as part of a visit with other members of a cancer care team or at a separate appointment. Questions asked during a cancer genetic counseling visit include the type and location of cancer, age at diagnosis, and previous test results. Having this information about anyone with a history of cancer is helpful.

If the choice is made to have genetic testing, a genetic counselor will explain what different results mean based on personal and family history. If someone has a history of cancer, a genetic counselor can talk about options for genetic testing and why it can be helpful for decisions about treating and managing cancer. If there are family members with cancer, a genetic counselor can look at the family history and decide if genetic testing is appropriate. If a genetic change is found in someone with a family history of cancer, this information can help understand the risk of getting cancer and what steps can be taken for screening, early detection, and prevention. Such information can be important to share with other family members who might have inherited the same change.



OTHER GENETIC COUNSELING SPECIALTIES

Genetic counselors work in many other specialties, including:

- ✓ Cardiovascular (heart)
- ✓ Neurological (brain)
- ✓ Connective tissue (skin, joints, and blood vessels)
- ✓ Differences or variations in sex development (hormones and reproductive organs/genitals)
- ✓ Psychiatric (mental health)

Genetic counselors also meet with people who have had “direct-to-consumer” genetic testing (DTC-GT). These tests are sold directly to a person, without involving a healthcare professional. They can give information about some genetic information such as ancestry, how someone responds to certain medicines (known as *pharmacogenomics*), or whether they carry certain genetic conditions. Meeting with a genetic counselor is recommended to go over DTC-GT results. This information could be important for a person or their family, and additional testing may be needed.

There are many other settings where a genetic counselor can be involved in education about genes, inheritance, and testing options. Anyone interested in speaking to a genetic counselor can ask their healthcare provider or visit one of the below websites for more information.



RESOURCES

For more information about genetic counseling or to find a genetic counselor, please visit the National Society of Genetic Counselor's patient-friendly website:

<https://www.aboutgeneticcounselors.com/>.

To search for genetics clinics by location and/or specialty area, please visit the American College of Medical Genetics and Genomics search tool:

<https://clinics.acmg.net/site/index>.

For a helpful guide on collecting family healthy history, please visit the National Society of Genetic Counselor's family history fact sheet:

[https://www.aboutgeneticcounselors.com/Portals/0/NSGENETIC COUNSELOR%20Family%20History%20Fact%20Sheet%20Feb%202021.pdf](https://www.aboutgeneticcounselors.com/Portals/0/NSGENETIC%20COUNSELOR%20Family%20History%20Fact%20Sheet%20Feb%202021.pdf)

For more information about the Genetic Information Non-Discrimination Act, please visit the National Human Genome Research Institute's resource:

www.genome.gov/sites/default/files/media/files/2020-09/GINA_patient_resource.pdf.

For more information about genetic services, please visit the National Genetics Education and Family Support Center website:

<https://nationalfamilycenter.org/>



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Family Center Workgroup on Inclusion, National Genetics Education and Family Support Center

Family Center Workgroup on Inclusion:

Molly Martzke

Bailey Perez

Cindy Weber

Mita Bhattacharya

Deepa Srinivasavaradan

Jeannette Mejias

Linda Hampton Starnes

Sarita Edwards

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