

Fast Facts: Genetics

For families who have children with special needs

Family Voices Colorado

*What we do to help
families who have
children with
special needs:*

- Statewide Information & Referral
- Navigation Through the Various Systems for Parents and Professionals
- Advocacy & Support
- Training & Education
- Family Leadership Development
- Parent & Family Engagement
- Legislative Advocacy
- System & Policy Change
- Home-Based Services
- Data Collection
- Health Benefits Counseling & Advocacy

Family Voices Colorado

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www.familyvoicesco.org

Colorado Resource Directory

www.coloradoresourcedirectory.net

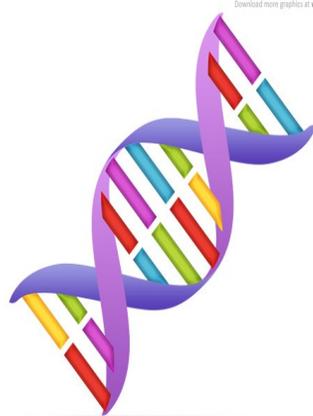
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What is Genetics?

Genetics is the study of heredity. Heredity is a biological process where a parent passes certain genes onto their children or offspring. Every child inherits genes from both of their biological parents and these genes in turn express specific traits. Some of these traits may be physical for example hair and eye color and skin color etc. On the other hand some genes may also carry the risk of certain diseases and disorders that may pass on from parents to their offspring.

Genes in the cell

The genetic information lies within the cell nucleus of each living cell in the body. The information can be considered to be retained in a book for example. Part of this book with the genetic information comes from the father while the other part comes from the mother.



Chromosomes

The genes lie within the chromosomes. Humans have 23 pairs of these small thread-like structures in the nucleus of their cells. 23 or half of the total 46 comes from the mother while the other 23 comes from the father.

Genetic Testing

Genetic tests are done by analyzing small samples of blood or body tissues. They determine whether you, your partner, or your baby carry genes for certain inherited disorders. Genetic testing has developed enough so that doctors can often pinpoint missing or defective genes. The type of genetic test needed to make a specific diagnosis depends on the particular illness that a doctor suspects. Many different types of body fluids and tissues can be used in genetic testing. For deoxyribonucleic acid (DNA) screening, only a very tiny bit of blood, skin, bone, or other tissue is needed.

Genetic Testing During Pregnancy

For genetic testing before birth, pregnant women may decide to undergo amniocentesis or chorionic villus sampling. There is also a blood test available to women to screen for some disorders. If this screening test finds a possible problem, amniocentesis or chorionic villus sampling may be recommended.



Family Voices

Colorado

- *Navigation*
- *Support*
- *Information*
- *Advocacy*

Programs currently funded through FVC

- Information, Referral and Training for Families and Professionals
- Family to Family Health Information Network
- Medical Home Initiative

Helpful knowledge... know about your **Single Entry Point (SEP) Agency:**

Go to HCPF to find the case management agency that will help your family with long term care needs: including personal care, medical transportation, home access modifications, and respite care.

www.familyvoicesco.org

Testing during pregnancy– continued..

Amniocentesis is a test usually performed between weeks 15 and 20 of a woman's pregnancy. The doctor inserts a hollow needle into the woman's abdomen to remove a small amount of amniotic fluid from around the developing fetus. This fluid can be tested to check for genetic problems and to determine the sex of the child. When there's risk of premature birth, amniocentesis may be done to see how far the baby's lungs have matured. Amniocentesis carries a slight risk of inducing a miscarriage.

Chorionic villus sampling (CVS) is usually performed between the 10th and 12th weeks of pregnancy. The doctor removes a small piece of the placenta to check for genetic problems in the fetus. Because chorionic villus sampling is an invasive test, there's a small risk that it can induce a miscarriage.

Common Genetic Disorders

Down Syndrome

Down syndrome is a developmental disorder, caused by additional copy of chromosome. Patients suffering from this syndrome have a flat face and broad nose and their appearance is very distinctive. Many of children with Down syndrome are mentally challenged and they are exposed to higher risk of respiratory infections, hearing and visual problems, leukemia and heart related conditions. Life expectancy of these patients is usually reduced by 50 years. Corrective surgery and physical and speech therapy may help these patients. Scientists have discovered that mothers older than 35 years of age more often gave birth to children with Down syndrome. Ultrasound may detect this syndrome in an unborn child.

Sickle Cell Anemia

Red blood cells become rigid in this type of anemia. They normally transport oxygen from the lungs to other parts of the body, but because of the rigidity, there is less oxygen transported to important organs in the body. Heart, kidneys, liver, spleen and lungs get less oxygen than they need so patients experience anemia, jaundice and growth problems. In most cases, patients suffering from this genetic disorder live 30 to 40 years. The most common treatment options are blood transfusions, plenty of water and avoidance of exercise and stress.

Cystic fibrosis

Cystic Fibrosis- mostly affects the lungs and digestive system. It results from a fault in a particular gene. As a result, the mucus produced by the lungs and intestines to be thick and sticky. Both parents must carry the faulty cystic fibrosis gene for the disease to be passed to their child. Cystic fibrosis is usually detected in newborn babies through a neonatal screening test, known as the heel prick test. This free test involves pricking the heel of the baby to gain a tiny blood sample and detects up to 95% of babies with cystic fibro-

sis. If a baby has a positive heel prick test, it should then have a sweat test at about 6 weeks old to see if it either has the disorder, or is a healthy carrier of the faulty gene. The lives of people with cystic fibrosis are usually shortened by the disorder, but they can lead happy and productive lives well into middle age. While cystic fibrosis cannot be cured, physiotherapy and many other treatments are available to improve quality of life and reduce complications.

Duchenne muscular dystrophy (DMD)

The DMD gene helps produce a protein called dystrophin, which is important for muscle strength, support and repair. People with Duchenne muscular dystrophy don't produce the normal form of dystrophin, which means their muscles are more easily damaged and don't work properly. The genetic mutation of the DMD gene is either inherited from parents or caused by a genetic change in the child. There are many types of muscular dystrophy, with Duchenne muscular dystrophy being the most common in children. Becker muscular dystrophy is very similar to Duchenne muscular dystrophy. It has the same underlying cause but is usually less severe, presenting with slightly different symptoms. The first thing parents usually notice is that their child isn't reaching their milestones. They might also notice that their child falls over often, is clumsy and walks on their toes.

Later, the child with Duchenne muscular dystrophy might develop:

- muscle weakness that affects their [posture](#), [walking](#) and running
- reduced joint movement due to shortening of their muscles
- problems with their [heart muscle](#), affecting heart function
- [difficulty breathing](#) as their muscle weakness worsens

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Genetic Counseling

What is Genetic Counseling?

Genetic counseling gives you information about how genetic conditions might affect you or your family. The genetic counselor or other healthcare professional will collect your personal and family health history. They can use this information to determine how likely it is that you or your family member has a genetic condition. Based on this information, the genetic counselor can help you decide whether a genetic test might be right for you or your relative.

Reasons for Genetic Counseling

Based on your personal and family health history, your doctor can refer you for genetic counseling. There are different stages in your life when you might be referred for genetic counseling:

Planning for Pregnancy: Genetic counseling before you become pregnant can address concerns about factors that might affect your baby during infancy or childhood or your ability to become pregnant, including

- Genetic conditions that run in your family or your partner's family
- History of infertility, multiple miscarriages, or stillbirth
- Previous pregnancy or child affected by a birth defect or genetic condition
- Assisted Reproductive Technology (ART) options

During Pregnancy: Genetic counseling while you are pregnant can address certain tests that may be done during your pregnancy, any detected problems, or conditions that might affect your baby during infancy or childhood, including

- History of infertility, multiple miscarriages, or stillbirth
- Previous pregnancy or child affected by a birth defect or genetic condition
- Abnormal test results, such as a blood test, ultrasound, CVS, or amniocentesis
- Maternal infections, such as CMV, and other exposures such as medications, drugs, chemicals, and x-rays

Genetic screening that is recommended for all pregnant women, which includes cystic fibrosis, sickle cell disease, and any conditions that run in your family or your partner's family

Caring for Children: Genetic counseling can address concerns if your child is showing signs and symptoms of a disorder that might be genetic, including

- Abnormal newborn screening results
- Birth defects
- Intellectual disability or developmental disabilities
- Autism spectrum disorders (ASD)
- Vision or hearing problems

Following your genetic counseling session, you might decide to have genetic testing. Genetic counseling after testing can help you better understand your test results and treatment options, help you deal with emotional concerns, and refer you to other healthcare providers and advocacy and support groups.



Resources

Mountain States Regional Genetic Network (MSRGN) www.mountainstatesgenetics.org

9111 Jollyville Road, Suite 280
Austin, TX 78759
General Telephone: 512-279-3910 or 512 279-3902
Fax: 512-279-3911

Children's Hospital Colorado— Genetics Department

www.childrenscolorado.org/doctors-and-departments/departments/genetics
303-724-2370