

What Is a Genetic Condition?

A genetic condition or disorder is a health problem caused by a change in a person's genes. Genes are parts inside our cells that tell the body how to grow and work and are passed down from one generation to the next. Genetic conditions can be passed down from one or both parents, happen randomly on their own, or change because of things in the environment like chemicals or smoke. Genetic disorders can cause birth defects and variations in how we look, long-term diseases, developmental problems, and issues with seeing and hearing. Many genetic disorders cannot be cured, but symptoms can be managed through treatments like medicine, surgery, diet, and therapy. The right treatment depends on the specific condition.

Symptoms vary widely. Signs include delays in development, changes in behavior, feeding or digestive problems, slow growth, nerve or brain issues, and vision or hearing loss.

Frequently Asked Questions (FAQ)

How common are genetic conditions?

Gene changes are common, but genetic disorders that affect daily life are not. Some of more well-known genetic conditions include Down syndrome, Fragile X syndrome, congenital heart defects, spina bifida, cystic fibrosis, sickle cell disease, and Tay-Sachs disease. Some gene changes can cause rare disorders, which are conditions that affect fewer than 200,000 people in the United States.

What should I do if I suspect my child has a genetic condition?

Talk to a medical provider. They can examine your child and refer you to a specialist for genetic testing. Early care is important, so do not wait to make an appointment. Diagnosing rare conditions can take time.

What kind of help is there for genetic conditions?

Support and treatment depend on your child's needs and diagnosis. Options may include:

- **A care team:** The team may consist of specialists, therapists, nutrition support, mental health providers, your regular doctor, and others.
- **Medical care:** Medication, surgery, and specialized treatments can help with a wide range of needs.
- **Home and community-based services:** New York State's Office for People with Developmental Disabilities (OPWDD) offers children and youth services that support them throughout their entire lives.

- **Assistive technology and equipment:** Certain devices and tools can help with communication, learning, or daily activities.

What other conditions are common in people with genetic disorders?

Children with genetic conditions often have complex medical needs. Be open with medical providers about all symptoms and concerns.

Talking with a Medical Provider

When talking with medical providers or specialists, you might want to:

- **Bring notes:** Keep track of any symptoms you see at home. Using a notebook or phone app can help.
- **Ask questions:** Find out which specialists your child should see and how often. Discuss what testing might be needed.
- **Advocate for your child:** Not all providers understand genetic disorders and rare conditions. It's OK to ask questions and get a second opinion.

What You Can Do at Home

Getting help can take time, and there are often waitlists. In the meantime, it may help to:

- **Manage stress:** It's normal to feel sad, overwhelmed, and frustrated. Try to make time for fun activities for your child and yourself. Ask friends and family for help when needed.
- **Seek support:** Many genetic conditions have local or national support groups, including groups for rare conditions. Talking with other families can be very helpful.

Learning about genetic conditions and getting help is a journey. You're not alone.

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About This Series

This series of handouts on health conditions was written by clinicians in Weill Cornell Medicine's Pediatric Mental Health Integration Program, partners in the ACT for Youth CYSHCN Program Center of Excellence at Cornell University.

