



## Welcome to the CDG Community

You may have just learned that you or one of your family members has CDG. You may be a support person for a family affected by CDG. You may be a professional taking care of a patient with CDG. CDG can be confusing for anyone. We hope that this brochure helps you understand CDG better.

Improved understanding of CDG will help you better love and care for a person with CDG.

### **What is CDG?**

CDG stands for Congenital Disorders of Glycosylation. CDGs are a large group of ultra-rare, inherited disorders that affect the complex process in the body called glycosylation. There are currently over 160 types of CDG that have been identified. CDGs are severely under-diagnosed and misdiagnosed with only approximately 1,200 cases diagnosed with CDG globally, and only 220 cases currently reported in the United States.

## What can I expect in the future for myself or my family member diagnosed with CDG?

Every child with CDG is special and unique. Individuals with CDG have very charming personalities and are generally happy and social. CDG symptoms vary by CDG type. Each CDG type can have more or less severe symptoms and most children with CDG require a team of specialist doctors to coordinate their care.

Some children with CDG never require a hospital visit, while others may need multiple hospital visits in the first year. Some children may also have serious medical problems that can become life threatening, but life threatening episodes typically decrease as children get older.

Many individuals with CDG have disabilities. These can be improved with physical, occupational, and speech therapy. These therapies are often needed lifelong.



## What Support is Available?

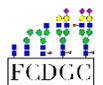
The first step each family should take after receiving a diagnosis of CDG is to join the CDG family support groups.

CDG CARE is a nonprofit organization founded by parents seeking information and support for all types of CDGs.

The CDG CARE Family Support Network is a community serving to connect patients, families and caregivers and promote the support of resources and inspiration to those affected by CDG. Together, our members stand united to advance research and resources for all CDGs.

To learn more about CDG CARE and sign up for the Family Support Network and receive the members only e-newsletter, visit [www.cdgcare.org](http://www.cdgcare.org).

On the CDG CARE website, you should also use the links under "Connect" to join the private family Facebook group, enroll in the CDG Connect global patient registry, and sign up for the community-wide quarterly e-newsletter subscription.





## Clinical Manifestations of CDGs

Most children who have CDG have neurological issues and symptoms, developmental problems, growth delays, and problems with organs not working like they should.

Congenital means that CDG is a condition that happens at or before birth. Notice that “disorders” is plural. This is because CDG is not just one disorder, but rather, a group of disorders. There are many types. Which type your child has depends on which body system is affected.

When someone has CDG, his or her body cannot properly add or attach the sugar building blocks to proteins or lipids. And because every single system in the body needs the process of glycosylation to work right, in most cases the result is that the body is not able to function normally.

Diagnosis of CDG varies with access to specialist care, as well as the severity of symptoms & potential for misdiagnosis. Genetic testing is the most common and reliable way to diagnose CDG and can also determine the type of CDG that the patient is affected with.

CDGs should be suspected in any child with multiple unexplained health concerns including those who present with: hypotonia, failure to thrive, developmental delay, liver disease, elevated liver enzymes, coagulopathy, esotropia and seizures.

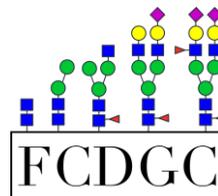
## Research & Opportunities

Most CDG types do not have specific treatments or cures currently available. However, many of the health concerns in CDG are treatable.

Over the past three years, the research landscape for CDGs has advanced considerably. We encourage you to connect with one of our Frontiers in CDG Consortium (FCDGC) regional centers for a medical consultation and to learn about natural history study research and clinical trial opportunities. To learn more about the FCDGC, visit:

[www.rarediseasesnetwork.org/fcdgc](http://www.rarediseasesnetwork.org/fcdgc)

Medical professionals, researchers, scientists, and patient advocacy groups are working tirelessly to identify new therapies and treatment options for all CDGs.



## Education & Resources

CDG CARE is pleased to offer our community online educational resources including medical professional recordings from prior year’s conference sessions, webinars targeting issues commonly faced by CDG patients and families, and short whiteboard videos providing an Overview of CDG, the FCDGC, and the process of participating in clinical trials. All of these resources are available on the CDG CARE YouTube Channel which can be easily accessed from the News & Events Tab at [www.cdgcare.org](http://www.cdgcare.org).

If a family or clinician has specific medical questions, concerns or is in need of an urgent consultation with a CDG medical professional, we encourage you to email CDG CARE at [info@cdgcare.org](mailto:info@cdgcare.org) and let us know.

When you are ready, our Community is waiting to welcome you, answer your questions, share their experiences and connect with your family so that you can be assured that you are not alone on this journey!

