



# Caring for Your Child With CDG

## Learning About CDG

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You have been given this resource because your child has been diagnosed with CDG. CDG stands for congenital disorders of glycosylation. Knowing that your child has this condition may worry you. You may feel overwhelmed right now and you may wonder what you can expect for your child.

***You do not have to manage alone.*** Your child's health care team is here to help. Just by reading this information, you have taken an important step in caring for your child.

What you can do:

- Learn everything you can about CDG and how it may affect your child's daily life. Learning about CDG can help you advocate for your child's needs and make health care decisions.
- Learn about treatment options.
- Know the symptoms of health problems that may result from CDG and what to do if your child has symptoms.
- Understand how you and your family can take an active role in your child's health care.

This resource explains how you can help your child. It includes information about the members of your child's health care team, how CDG may be treated, how to know when your child is having symptoms of CDG complications, and how you can help your child have a good quality of life.

If you have questions about CDG, talk with your child's health care team.

## Your Child's Health Care Team

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Your child's health care team includes many health care providers who work closely together to support your child, you and your family. The health care team plans care tailored to your child's specific needs. The team works together to help you manage your child's symptoms and help your child make the most of his or her abilities.



Your child's health care team may include the following:

- **Medical genetics specialists** help with diagnosis, counseling, testing family members, and finding and organizing the treatment team for your child.
- **Genetic counselors** teach you about CDG, as well as provide counseling and testing of family members.
- **A primary care pediatrician** coordinates and provides routine childhood care.
- **Nurse practitioners and special nurses** have advanced education in a nursing specialty. These nurses provide guidance for nursing staff caring for your child. They can provide support to you and your family when your child is in the hospital.
- **Neurologists** diagnose and treat seizures, balance problems and stroke-like episodes (SLEs).
- **Physical therapists (PTs)** help strengthen muscles and improve movement affected by CDG.
- **Occupational therapists (OTs)** evaluate and treat your child for problems that limit his or her ability to do functional activities, such as dressing, bathing and going to school. OTs focus on fine and gross motor skills and the ability to swallow safely. OTs help your child be as independent as possible.
- **Speech language pathologists** help with speech therapy and problems with chewing or drinking.
- **Registered dietitians** evaluate your child's nutritional needs and help decide how best to meet those needs. This includes managing symptoms, promoting healthy growth and development, and preventing complications.
- **Gastroenterologists** diagnose and treat problems with diarrhea, constipation or vomiting.
- **Endocrinologists** diagnose and treat hormone and growth problems or low blood glucose levels.
- **Cardiologists** diagnose and treat heart problems.
- **Vision specialists, called ophthalmologists,** diagnose and treat vision problems.

Depending on your child's unique health concerns, the team may also include the following:

- **Hepatologists** diagnose and treat liver problems.
- **Hematologists** diagnose and treat bleeding or clotting disorders.
- **Orthopedic specialists** diagnose and treat bone and joint problems.
- **Infectious disease physicians and immunologists** review test results and work with team members to recommend vaccinations and medications to prevent and treat infections.
- **Hearing specialists, called audiologists**, evaluate your child's hearing and assess whether your child needs a hearing aid or other type of hearing support.
- **Ear, nose and throat specialists** help your child if he or she has hearing problems.
- **Financial representatives** can meet with you and answer insurance and other financial questions.
- **Social workers** support you and your child through your health care journey. They can refer you to community resources that will help you care for your child at home and also help you make financial plans.

Remember that you, your child and your family are the most important members of the team.



# Treating CDG

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Treatment is focused on managing symptoms and preventing complications caused by the condition. Treatment depends on which sub-type your child has, which body systems are affected, age, symptoms, and other health conditions. At this time, there is no cure for CDG.

## Goals of treatment:

- Manage your child's symptoms
- Prevent complications
- Give your child the best quality of life possible

Listed here are complications your child may have and treatments that may help. This section is organized by body system.

## Neurological

**Poor balance and coordination, called ataxia:** Physical and occupational therapy are the most important treatments for balance and coordination problems. Muscle training and exercises may help your child's strength, flexibility, balance, motor development, and mobility.

If it is hard for your child to stand or walk, your child's physical therapist can work with you and your child to identify equipment that may help, called adaptive equipment. Adaptive equipment includes standers, walkers, canes, wheelchairs, and lift systems.

**Low muscle tone, muscle contractions and stiff muscles:** Medications that can lessen muscle tightness may be used to improve functional abilities, treat pain, and manage complications related to CDG symptoms. Your child's provider may recommend injections of botulinum toxin on a regular basis, such as every three months.

Medication your child takes by mouth may also be used, such as baclofen (Gablofen™).

**Stroke-like episodes (SLEs):** Ensuring your child gets enough liquids and stays well-hydrated can help prevent SLEs. Treatment may include physical therapy to help manage effects of the episodes.

**Seizures:** Many different types of seizures occur in CDG. Treatment may include medications, hormones, dietary supplements, and diet changes.

Fever can make seizures worse. Fever-reducing medication, such as acetaminophen, may be recommended. However, if your child has liver problems, it is important he or she is never given a higher dose than recommended.

**Slurred speech:** This is usually treated with speech therapy. Some children with CDG use a special device to communicate.

**Crossed eyes:** This is usually treated with surgery.

## Gastrointestinal

**Vomiting:** Usually, providers recommend that your child gets the right amount of calories for your child's needs, if possible. Your child's provider may recommend the use of a special formula that is pre-digested.

**Chronic diarrhea:** Treatment with mannose in MPI-CDG can prevent diarrhea.

**Protein-losing enteropathy:** A diet rich in mid-chain fatty acids (MCT) and some medications can help prevent loss of protein.

Regular albumin infusions can help keep blood protein levels in balance. These can also help prevent fluid collection, called edema, under the skin and around vital organs.

## Hepatic (liver)

**Liver problems:** Some children with liver disease are given sugar supplements. However, for most, no treatment is needed. Your child should be vaccinated for hepatitis A and B.

**Cholestasis:** This is usually treated with eating a special diet that includes fat-soluble vitamins — mainly vitamins K, D and E.

**Liver cirrhosis:** Your child's health care provider watches for the development of liver cancer, called hepatocellular carcinoma, and recommends treatment.

## Hematologic (blood)

For all the possible blood problems, your child's provider may recommend:

- Watching for signs of blood clotting, including watching test results for PT, aPTT, factors I, VIII, IX, and XI, antithrombin, protein C, and protein S.
- Making sure your child gets the right amount of fluids and activity.
- Prescribing medications to prevent blood clots, also called blood-thinners, if your child has a blood-clotting problem. These medications include unfractionated and low-molecular-weight heparin, warfarin and rivaroxaban.
- Giving your child fresh frozen plasma therapy for surgeries and in case of bleeding.
- Giving your child sugar supplements for blood clotting problems.

## Endocrine

Problems may include:

- Slow weight gain, called failure to thrive.
- Poor thyroid function that can result in growth delay and constipation.
- Problems with glucose metabolism.
- Low stress hormones associated with low blood sugar levels, called hypoglycemia.

For many of these problems, the treatment may be giving your child hormones in the form of medication he or she swallows. Treatment may also include a special diet.

**High insulin levels associated with low blood sugar levels:** This is usually treated with diazoxide.

## Immune system

A possible problem is frequent infections. Infections may be treated with:

- Medications to treat infections, such as antibiotics, antivirals and antifungals.
- Immunoglobulins given through a tiny tube, called a catheter, inserted in a vein in your child's hand or arm. This is called an IV, which stands for intravenous.
- Diet changes.
- A bone marrow transplant if a child has severe immune system failure.

*"Become a CDG expert. Familiarize yourself with the symptoms so you know what to watch for and can act quickly. Learn the medical terms so you can have meaningful conversations with medical professionals."*



## Your Child's Diet

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Most children with CDG can eat a normal diet. However, some treatments that include diet changes or supplements can help with certain problems caused by CDG. These treatments are meant to increase the concentration of sugars for sugar-chain production.

Other treatments include adding trace elements in your child's body, which are important for the normal function of enzymes involved in glycosylation. Studies have shown these to be helpful with certain sub-types of CDG:

- Galactose, which your child swallows, has improved liver, blood and hormone disease in PGM1-CDG.
- Mannose, which your child swallows, has improved liver, gastrointestinal, blood and hormone disease in PGM1-CDG. However, for some children, this therapy causes significant side effects. One is damage to the red blood cells, called hemolysis. The other is a yellow tint to the skin and eyes, called jaundice. Jaundice is caused by an excess of bilirubin, a substance created when red blood cells break down. This condition is called jaundice.
- Galactose, manganese and uridine, which your child swallows, have led to better seizure control.
- Fucose, which your child swallows, has been found to have positive effects on immune symptoms and neurologic development in SLC35C1-CDG.

### Other treatments

- If your child has low blood glucose levels, your child's provider may recommend he or she eats often. These meals need to include complex carbohydrates.
- Tube feeding to help meet nutritional needs.

### *About tube feeding*

The medical term for tube feeding is enteral nutrition. When a child cannot eat or cannot eat enough, tube feeding can provide nourishment. During tube feeding, breast milk or formula passes into the digestive tract, that is, the stomach and small intestine, through a tube in your child's nose, mouth or an opening in his or her abdomen.

Breast milk or formula can be used for tube feedings. Sometimes your child may need a supplemental or specialty formula for healing and meeting his or her nutritional goals.

Your child's health care team lets you know if your child needs these. Your child's provider may recommend continuous or nighttime tube feedings. If your child needs to be tube fed, you will be shown how to do it.

As your child grows, his or her nutritional needs will change. Meet with a metabolic dietitian to learn how to help your child meet nutritional needs.





## Health Care Concerns

### Preventing infection

Most children with CDG do not have a greater risk of infection. However, when they do have an infection, it can be harder to recover.

#### *Follow these guidelines:*

- Handwashing is the single most effective way to prevent infection. Wash your hands thoroughly with warm, soapy water. Rub them for at least 15 seconds before you rinse. Always carry a small bottle of waterless hand sanitizer for you and your children to use when soap and water are not available.
- Keep your child away from people with active, contagious disease or illness, including those with cold or flu symptoms.
- If your child is exposed to measles or chicken pox, contact your child's health care provider for information about next steps.
- Any time your child is in the hospital, keep the door of your child's room closed.
- One of the first signs of infection is a fever. Do not ignore a fever. Seek medical care for your child within one hour if he or she has a temperature of 100.4 degrees Fahrenheit (38 degrees Celsius) or higher.

### Vaccinations

Unless your child's health care provider tells you otherwise, your child should get age-appropriate vaccinations to help prevent infection and disease.

*"Know your child's health history. When there are situations that you need to act fast, doctors and nurses can act quickly to find a solution."*

## Possible Complications of CDG

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Your child with CDG has the risk of complications. Talk to your child's health care team about these possible complications and what you can do to prevent them. Here are some common complications of CDG, symptoms to watch for and what to do:

### **Possible complication: *Infection***

**Symptoms:** Temperature of 100.4 degrees Fahrenheit or higher, chills, not controlled by medication

**What to do:** Contact your health care provider immediately. Use physical cooling if needed to lower seizure risk.

### **Possible complication: *Thrombosis, which is a blood clot blocking a blood vessel***

**Symptoms:** A change in skin color or size of arms or legs

**What to do:** Seek emergency care.

### **Possible complication: *Seizures***

**Symptoms:** Repetitive shaking or stretching movements, chewing, nodding or jerking motions

**What to do:** Seek emergency care. If you have a seizure gel for rectal applications at home, use it immediately following the directions you have been given by your child's health care provider.

### **Possible complication: *Stroke-like episode***

**Symptoms:** One side of your child's face or body looks or acts differently than the other or your child is unable to move an arm, hand, leg, or foot.

**What to do:** Seek emergency care.

### **Possible complication: *Bleeding that does not stop within 5 minutes or loss of consciousness as a result of an accident***

**What to do:** Call 9-1-1 or seek emergency care.

# Living Well With CDG

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Having a child with CDG can bring many changes to your life and your family's life. Managing treatment and follow-up care may seem overwhelming to you.

Your child's health care team can help. Talk with them about how to manage the changes brought on by having a child with CDG. Team members can offer guidance and connect you with other support services.



## Growth and development

Children with CDG share many of the same needs as healthy children. Talk to your child's health care team regularly to evaluate the progress of your child as he or she moves through various stages of growth and development.

## Delayed milestones

New skills that children learn as they grow and develop are called milestones. These include:

- Gross motor skills, such as crawling or walking.
- Fine motor skills, such as picking up small objects.
- Speech and language.
- Social and cognitive skills, such as the ability to connect with others, reasoning and problem-solving.

Children with CDG may walk or crawl later than is typical for other children. It may take several years to know how well your child will walk or move. Your child needs regular follow-up visits with physical therapists and occupational therapists. Therapy can help improve your child's skills and mobility.

## Learning problems

Some children may have cognitive impairments or specific learning disorders. As a result, a child may need special services at school. Learning needs should be assessed before your child starts kindergarten.

## Record your child's milestones

You can help provide positive memories by recording important events, signs of progress, milestones, and funny stories.

Some ideas include:

- Establish a website to journal, communicate, and save memories and well wishes from friends and family. Ask a health care team member about using a computer if you did not bring one.
- Consider personal journaling, scrapbooking or picture-taking as a means of sharing memories with your child later in life.
- Keep a book of visiting friends and family and cards received.
- Take occasional hand-and-foot ink prints to show growth and progress throughout your child's life.
- Videotape interactions with your child and take pictures.

## Emotional issues

Having a child with health issues can exhaust you. You may feel resentful, guilty or isolated. You may have physical signs of stress, such as fatigue, headaches and muscle aches.

Other parents of children with CDG have said they have felt the same way. The important thing to know is that you do not have to manage on your own and help is available.

Social workers, therapists, chaplains, and clinical nurse specialists help you understand your feelings. They can also help you adjust your and your family's lifestyle. Over time, you may settle into a comfortable routine.

Be sure to take care of yourself. If you don't take good care of yourself, you won't be able to take good care of your child.

Tips for taking care of yourself:

- Try to get enough sleep, exercise often and eat healthy.
- Take breaks from caring for your child to do something just for you, for your partner, and for your other children.
- Accept help from friends and family. When friends and family offer help, ask them to spend time with your other children. Suggest they bring meals if you need them. Ask for help with the laundry, shopping and other household chores.

## If you have other children

Other children in the family may feel upset about their sibling's health struggles, and the extra attention he or she gets. Your other children may need one-on-one time with you. If possible, on a regular basis, arrange for another adult to care for your child with CDG while you spend time with your other children.

## Other caregivers

While being with your child is important, it is just as important to take time to care for yourself. In addition, there may be times when you are just not able to care for your child, such as when you have a health issue yourself.

Identify other people who can care for your child. These caregivers can be a parent, a sibling, a neighbor, or a friend. Consider having more than one caregiver in case someone isn't available when you need them. Caregivers should be able to get your child to and from health care appointments and provide basic care in your absence.

As you consider people who might fill this role, think about:

- Who is trustworthy, responsible and dependable?
- Who is physically capable of providing care?
- Who would be comfortable spending time in a health care environment? Who can tolerate illness, blood and unusual smells?
- Who can take time off work or school when needed?

Be sure to explain what is needed so they have a clear understanding of your child's needs.

Talk to your child's health care team if you have trouble finding others who can care for your child. Having a good support system is important for both you and your child.

## Daycare and school

It can be helpful for you and your child to spend time with other children, including those who don't have CDG as well as those who do. Going to daycare and later, to school, can offer those opportunities.

Talk to your child's health care provider about early intervention programs (EIPs) and special education classes. All states are required by law to offer early intervention programs for children from birth to 3 years old. Some states also offer special education classes for children between the ages of 3 and 5 years old. Ask your child's health care provider about special services that may be available.

## **Keeping your child safe**

You need to set up a safe environment for your child when you are at home and when you are away from home.

Preventing injuries and harm is not that different for children with CDG than for children without CDG. Each child is different, and the general recommendations to keep children safe should be adapted to fit your child's skills and abilities.

To help keep your child safe:

- Know and learn about the unique concerns and dangers for your child.
- Once you have ideas about keeping your child safe, make a safety plan and share it with your child, your family, and other adults who might be able to help if needed.
- Remember that your child's needs for safety will change over time.

Think about the following things when you make a safety plan for your child:

### ***Moving around***

Children who have limited ability to move or make decisions might not realize that something is unsafe or they might have trouble getting away.

- Carefully evaluate the areas where your child spends time. Make sure the areas are safe.
- Check your child's clothing and toys. Are they suitable for his or her abilities, not just age and size? For example, clothing and toys that are meant for older children might have strings that are not safe for a child who cannot easily untangle themselves, or toys might have small parts that are not safe for children who are still putting toys in their mouths.

### ***Car safety***

In addition to choosing a car seat that is age-, weight-, and height-appropriate, consider whether your child has difficulties sitting up or sitting still in the seat. Your child may need something different than a traditional car seat, as he or she gets older, depending on his or her ability to sit independently.

Talk to your child's health care provider or a child passenger safety specialist about the best type of car seat or booster seat and the proper seat position for your child.

### ***Safety equipment***

Examples of special safety equipment include hand rails and safety bars that can be put into homes to help a child who has difficulty moving around or a child who is at risk for falling.

### ***Communicating about safety***

Children who have problems communicating might have limited ability to learn about safety and danger. Try to find different ways to teach your children about safety such as:

- Showing them what to do.
- Using pretend play to rehearse.
- Practicing on a regular basis.



## *Traveling*

Your child may have a higher risk for developing blood clots. When people travel by plane, the risk of blood clots is higher due to long periods of time spent not moving.

If you plan to travel, encourage your child to move at least once an hour. Make sure your child drinks or gets enough fluids to stay well-hydrated.

Before your trip, ask your child's health care provider:

- How to massage your child's arms, hands, legs, and feet to encourage circulation and prevent blood clots.
- About whether your child should wear elastic travel socks.

## *Family planning*

People who have children with CDG have a higher risk of having another child with CDG. Talk with a genetic counselor about your specific risk.

## Financial Concerns

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Understanding what expenses are involved related to having a child with CDG can help you make decisions as you plan ahead.

One of the first things you can do to help get and stay organized is get a three-ring binder. Use the binder to:

- Record your child's medical identifier number(s), insurance company name and policy and group numbers, pharmacies you use, phone numbers for the billing office of the medical facilities your child receives care from, and social workers' names and phone numbers.
- Store your receipts.
- Estimate expenses.
- Take notes about the conversations you have related to financial aspects.

Some facts about expenses:

- Many insurance programs and companies require you to get approval before having some medical procedures, called preapproval.
- Not all medical costs are covered by insurance.
- Ongoing costs include regular medical appointments as well as the cost of regular trips for health care. These may include travel, parking and lodging expenses.
- Your child may need to take certain medications. These medications can be expensive and may not be completely covered by insurance.
- You are ultimately responsible for all charges related to medical care not covered by insurance. Most health care organizations submit all charges to your insurance company. Any unpaid balance is your responsibility to pay. These balances are usually the result of your insurance co-pay, coinsurance and deductibles.

If your child is covered by Medicaid, you may be able to receive financial assistance for meal and lodging expenses when you travel. Call your county's social services department and ask to speak to a county financial worker to find out more. Ask if you need preapproval to qualify.



### Steps to take:

- Talk to a financial coordinator or social worker about resources to help you meet the costs of your child's care.
- If you work, find out from your company's human resources department if you have disability benefits. Some employers provide short- and long-term disability, which covers some of your salary when you can't work.
- Set up as many automatic services in your life as possible, such as online bill payment.
- Meet with an accountant. Some expenses may be tax deductible. Find out what sort of expenses qualify and what you need to do to get credits or deductions.
- Consider private fundraising as a way to help meet expenses. Some organizations provide help in planning benefits and other fundraising activities.
- If you receive financial help from Medicaid or Social Security Disability, your coverage may be affected by your employment status. When you are ready to return to work, check with the agencies about how your coverage will be affected.

If you have questions or concerns, talk with a social worker and a financial representative from the health care facility where your child receives care.

## Follow-Up Care

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Children with CDG need long-term care by a medical care team. How often you bring your child in for follow-up care depends on your child's needs, age, symptoms, and abilities. Your child will probably have appointments with a primary care provider several times a year. You likely will bring your child once a year for appointments and tests with specialists.

It is very important you take your child to all regularly scheduled follow-up appointments. These visits are a way for your child's health care providers to monitor your child's health and development.



Bring to your child's follow-up appointments:

- A list of your child's medications, including dosages.
- A list of problems or challenges your child has been having as well as your own questions and concerns.

Over time, your child may have many appointments with the care team. During follow-up appointments, a health care provider examines your child. The provider may order tests. Use this time to talk to your child's provider about problems he or she is having and about questions you have.

After each appointment, your health care team recommends when the next appointment should be. Ask your child's provider whether follow-up appointments are scheduled by a team member or whether you need to do that.



## Resources

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Consider joining a support group, which may help you in many ways. It can be helpful to talk with others who understand the challenges — and rewards — of having a child with CDG.

Remember that the internet can be a wonderful tool for finding information on almost any topic. Unfortunately, not all health information on the internet is reliable. Be sure to evaluate the health information you find on the internet. Ask your child's health care provider where you can find good information.

For more information, ask your child's provider to suggest websites that have reliable information. Your health care provider cannot guarantee the accuracy of the information on websites. However, your child's provider can direct you to some that are likely to be reliable.

*"Share your story with others. Be the one to talk about it first, answer the difficult questions and smile at all the concerning stares. The more you talk about it, the more you make your vulnerability known, and the easier talking about your child's condition is going to be for you."*

## Final Thoughts

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### **Rely on your team of care professionals.**

You'll need to make important decisions about your child's treatment. A team, coordinated by your health care provider, may include social workers, teachers and therapists, who are all familiar with CDG and able to help explain the resources in your area. Ask if the team includes a case manager or service coordinator who can help access financial services and government programs in your community or county.

### **Find other families of children with CDG.**

Other families may have helpful advice. Some communities have support groups for parents and siblings of children with disabilities or who have chronic illnesses.

### **Keep records of visits with health care and service providers.**

Your child may have visits, evaluations and meetings with many people involved in his or her care. Keep an organized file of these meetings to help you decide about treatment options and to help you monitor progress.

### **Stay current on new technologies and therapies.**

Researchers continue to explore new approaches to help children with CDG.

Your child is special in so many wonderful ways. Yes, there will be challenging times ahead, but there will also be many joyful moments. And remember, at all times, your child's health care team is here to help you.



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