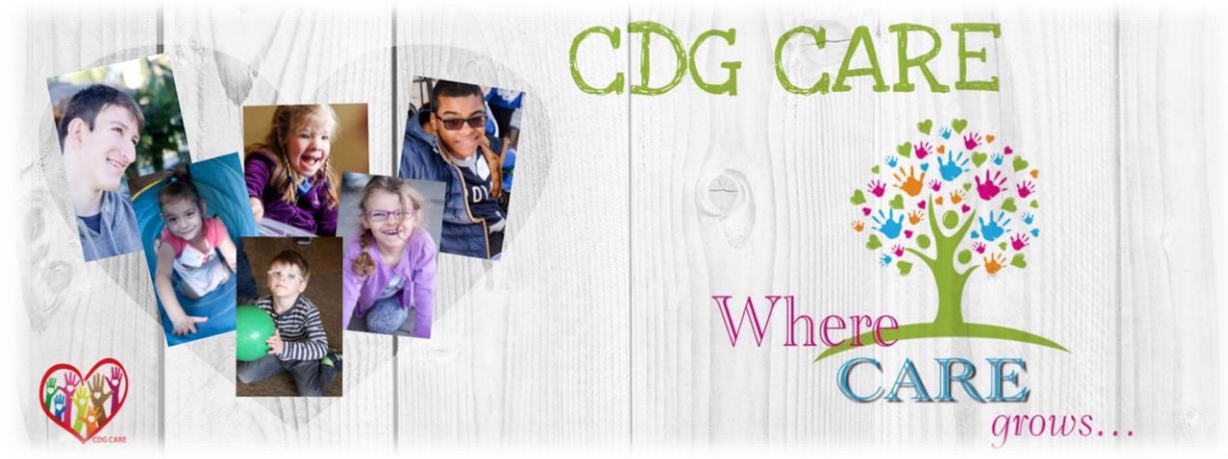


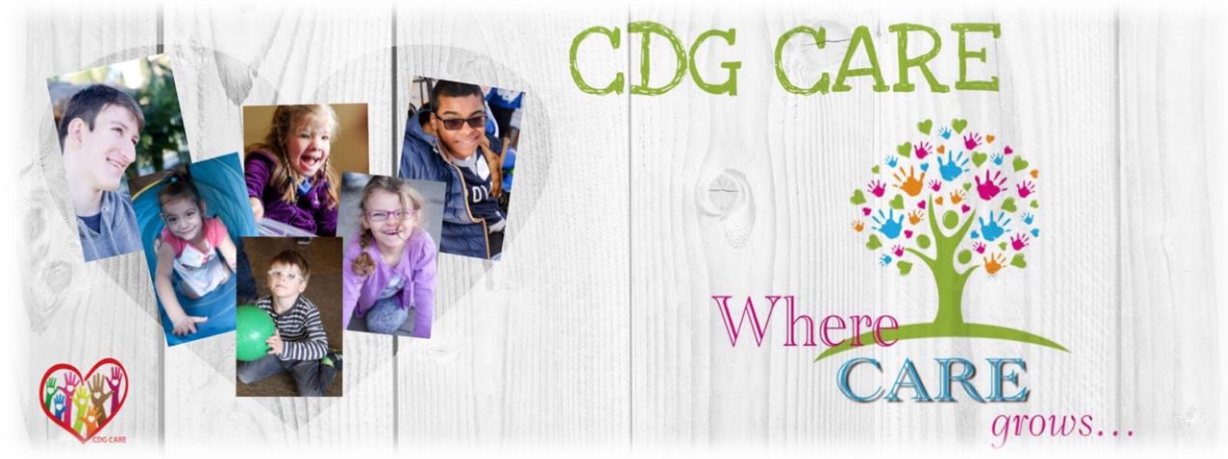
What treatments are available for CDG?

How will doctors likely treat specific medical concerns? Adapted from "CDG Summary of Features and Management"…

Growth concerns: For low weight, doctors often start by recommending a special formula with higher calorie content. Children with CDG do not typically require any dietary restrictions; they can tolerate normal amounts of sugars, fats and protein. If unable to adequately gain weight with high calorie formula, a child may require additional formula through a tube placed in the nose or directly into the stomach. This can often be removed when the weight is improved and the child is older. For additional growth concerns and issues, it is recommended that CDG children and adolescents by followed by an Endocrinologist.

Feeding problems with persistent vomiting: Many children with CDG have difficulty with feeding. It is common for parents to be concerned when the children are young because feeding can be difficult and these children may grow very slowly. Reflux and persistent vomiting are also common in many of the children. Thickening feeds, maintenance of an upright position after eating and antacids can be helpful. The involvement of a gastroenterologist and nutritionist to manage this is often necessary. Many children will require a gastrostomy tube. Should the child have a gastrostomy tube placed for nutritional support, it is important the child continue to be encouraged to eat by mouth if there is a low risk of aspiration. Continued speech and oral motor therapy is essential. This will not only smooth the transition to oral feeds but will also encourage speech when the child is developmentally ready.

Developmental delay: Developmental delays in children with CDG are typically first noticed around four months of age. At this point early intervention with occupational therapy, physical therapy and speech therapy should be started. As the child grows and the developmental gap widens between these children and their unaffected peers, parents need continued counseling and support.



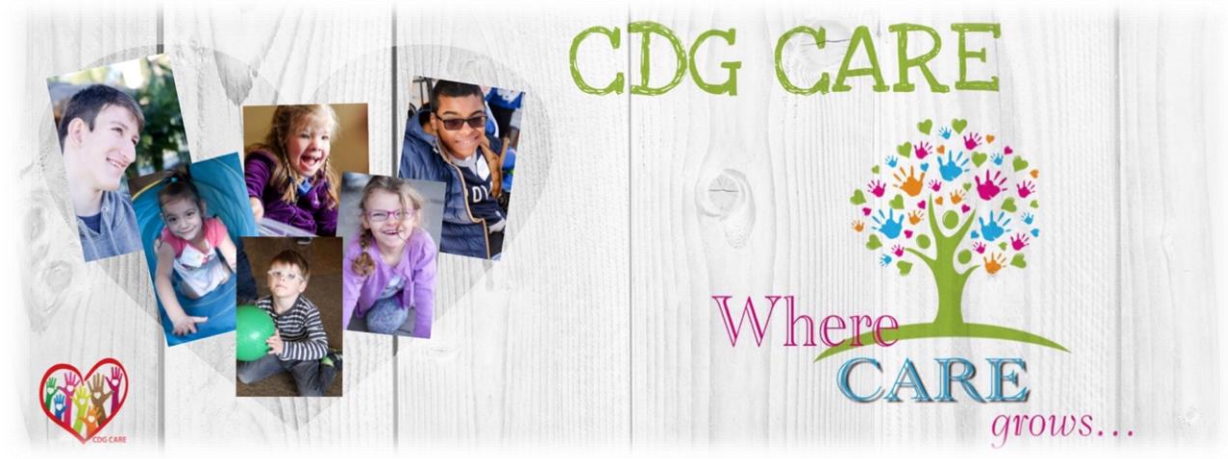
Abnormal liver function: In many of the types of CDG liver function tests (AST and ALT) are elevated in the first year of life. The AST and ALT may peak in the 1000-1500 range before it begins to return to normal. Typically, the ALT and AST return to normal by age 3-5 in children with PMM2-CDG and remain normal throughout the remainder of their lives. Other types of CDGs seem to follow a similar pattern although MAGT1-CDG patients may have persistent elevations of ALT even as adults.

Abnormal Bleeding/Clotting: Many patients with CDG have clotting factor deficiencies that can make it easier to bleed or deficiencies in other blood proteins such as ATIII, Protein C or Protein S that make it easier to develop blood clots. These usually balance each other out however they can change over time and with illness, injury or surgery, so consultation with a hematologist to monitor this over time is important. Infusion of fresh frozen plasma corrects the factor deficiency and clinical bleeding when indicated. Sometimes blood thinners such as Lovenox are used to prevent abnormal clotting.

Strabismus: Aggressive intervention by a pediatric ophthalmologist early in life is important to preserve vision in these children. Some children just require patching and glasses, but others with esotropia (crossed eyes) may require corrective surgery. Many patients with CDGs develop a retinitis pigmentosa that can cause night vision problems.

Pericardial effusion or Cardiomyopathy: Some CDG patients will be born with or develop pericardial effusions or cardiomyopathy, which can be life threatening. Most pericardial effusions do not cause any medical issues and resolve early in life. An initial echocardiogram is warranted with follow-up by a cardiologist every few years.

Hypothyroidism: Children with CDG who have elevated TSH and low free T4 should be treated with thyroid hormone. Assessment by a pediatric endocrinologist may be useful in some circumstances.



Seizures: Seizures can be variable in CDG. Some types of CDG have very difficult to control seizures while other types of CDG only rarely have seizures that are easily controlled with medication. If a child with CDG has seizures the involvement of a neurologist is essential.

Stroke-like episodes: Transient loss of neurologic function or a stroke-like episode may occur as early as 4 years of age in a child with CDG but most occur later. These may occur more frequently with head trauma (falls), dehydration or fever, although a formal study has never been done. Some of the children have seizures at the same time. Supportive therapy for the children as they recover, including good hydration by IV if necessary, and physical therapy during the recovery period is important. Full recovery may only take be a week but may extend to several months in some cases.

Additional management issues of adults with CDG include:

Orthopedic issues: Thorax shortening, scoliosis/kyphosis – Appropriate orthopedic and physical medicine management, with well supported wheel chairs, appropriate transfer devices for the home, and continued physical therapy is important. Some children and adults have had surgical treatment of their spinal curvature with variable success. Osteopenia/Osteoporosis are very common in CDGs and are usually treated with Vitamin D3. Other medications for osteoporosis do not always work in patients with CDGs and Estrogen supplements can sometimes be associated with blood clots so we do not like to use hormone replacement unless it is absolutely necessary.

Retinitis pigmentosa: With age, some individuals with CDG develop a pigment at the back of their eye that can affect their vision, in particular night vision. An eye specialist should monitor the progression of vision impairment.

Independent living issues: Young adults with CDG and their parents need to have issues of independent living addressed as they grow older. Aggressive education throughout the school years in functional life skills and even vocational training will support the transition to the years after schooling is completed. Independence in self-care and the tasks of daily living should be encouraged as much as is physically possible. Support and provision of resources to parents of a disabled adult is an important part of the management of the care of these patients.