Current treatment for CDGs involves treating the child’s specific symptoms. There is no medicine available to treat the problem that causes the symptoms of CDG, except for patients with CDG Ib (who take a medicine called mannose) and for some patients with CDG IIc (who take a medicine called fucose). Children with CDG are followed regularly by a geneticist and other specialists to treat their specific symptoms.

If you are concerned your child may have CDG, your health care provider can refer your child for a genetic consultation. The geneticist will examine your child and discuss his or her development. The geneticist or the genetic counselor will also discuss your family history in detail, and ask about any health conditions in members of your extended family. If this information is suggestive for CDG, the geneticist may talk with you about ordering testing for CDG. The geneticist may also recommend other genetic testing to determine if there is another genetic explanation for your child’s condition.

For more information on testing, including prices, CPT codes, and turn-around-times, please ask your doctor or contact Emory Genetics Lab.

The CDG Family Network: www.cdgs.com

National Organization for Rare Diseases (NORD): www.rarediseases.org

CDG Overview for Parents from the UCL Institute of Child Health: http://www.ich.ucl.ac.uk/website/gosh/clinicalservices/Metabolic_medicine/Custom%20Menu_02/CLIMB_CDG_parents_assay.doc

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Emory Genetics Laboratory (EGL) is a comprehensive clinical genetics testing laboratory specializing in molecular cytogenetics, rare disease testing, and newborn screening confirmatory testing.
What are Congenital Disorders of Glycosylation?

Congenital disorders of glycosylation (CDGs) are a group of genetic disorders that can cause a wide variety of symptoms. CDGs are caused by changes in the production and structure of sugar “tags” that are added to proteins and lipids (fats) in cells. The sugar tags tell the body where the proteins and lipids should be located. The process of creating and adding these sugar tags is called “glycosylation”.

If the sugar tags are missing or put together incorrectly, the proteins and lipids may not end up in the correct place. This misplacement can lead to the symptoms seen in individuals with CDG.

For example, if the misplacement occurs in cells in the brain, the individual may have seizures or developmental delay. If the misplacement occurs in cells in the gastrointestinal tract, the individual may have vomiting and diarrhea or trouble putting on weight. If the misplacement occurs in cells in the muscles, the individual may have low muscle tone, or be “floppy”. If the misplacement occurs in more than one organ at the same time, the individual may have multiple different symptoms. Any organ in the body could be affected.

Some of the symptoms become more obvious as the child grows, while some become less obvious when the child is older. Most commonly, CDG disorders begin in infancy and are associated with minor differences in facial, skin, and other body features.

Classification

The most common CDGs are classified as types I and II (CDG I, CDG II) based on the specific problem with the sugar tag. Different disorders in each type are defined by a small letter code (a, b, c, etc.) because each of them is caused by mistakes in a different gene.

Type I CDGs are caused by mistakes in genes that create the sugar tags or in genes that attach the sugar tags to proteins. Type II CDGs are caused by mistakes in genes that re-shape the sugar tags after they are added to protein. Currently, more than 30 different causes of CDG have been described. CDG type Ia is the most common form of CDG; over 700 patients with CDG type Ia have been reported.

Testing for CDG

There are two types of testing that may be used to diagnose CDG, biochemical testing and molecular testing. Most individuals with CDG can be diagnosed by biochemical blood tests that analyze the sugar tags on proteins in the blood. These tests can tell the doctor if the sugar tags are missing or put together incorrectly. These tests cannot detect every CDG, however, so the doctor may also need other blood or urine tests as well.

Once CDG is diagnosed by biochemical testing, or if biochemical testing does not give a clear result, further testing is required to determine the type or cause of CDG. Genetic (DNA) testing on a blood sample is available for most, but not all, of the known forms of CDG. If the genetic cause of CDG can be determined, family members can be tested to see if they are at risk for having (additional) children with CDG. Prenatal genetic testing would also be available if desired.

If genetic testing for CDG is negative, meaning that a genetic cause was not found, the child may still have CDG based on the biochemical test results. The negative genetic results mean that the child does not have a form of CDG caused by a gene that has been identified. New genes involved in CDG continue to be discovered. Additional genetic testing in the future may be able to identify a genetic cause for the CDG in a child who tests negative now.

Sometimes it is difficult to interpret genetic test results. These unclear results are called “variants of unknown significance”. In these cases, testing of the child’s parents and other family members may be recommended in order to try to understand the child’s results.