

CDG FAQs

CDG Family Network FAQ's, Updated by Jaak Jaeken, MD, Ph.D. 07/27/11 & Donna Krasnewich M.D., Ph.D. 08/24/11

How many cases of CDG are known worldwide?

We have no exact figures but the published cases figure around 600 for the N-glycosylation disorders. There are many more patients reported with a known O-glycosylation disorder (multiple exostoses, Walker-Warburg syndrome, muscle-eye brain disease, etc) but again it is very difficult to obtain exact figures. All together (known and unknown CDG) I guess that an incidence of 1 in 5000 births may be a minimum estimate. Responder: - Jaak Jaeken, MD, Ph.D. 07/27/11

How many cases of CDG are known in the United States?

In the United States, my guess is that there are close to 175 cases. In the past 16 months, I have heard about 35 affected children that I didn't know about before. Because there is no registry, this number is difficult to estimate. Many children are doing well and their physicians are taking excellent care of them, therefore, these families may not come to the attention of the organized CDG community. Responder: - Donna Krasnewich M.D., Ph.D. 08/24/11

What is a stroke-like episode?

A stroke-like episode is an acute event that very much resembles a stroke. A stroke is a sudden loss of consciousness due to an acute vascular disturbance caused by the rupture of an artery in the brain or its obstruction by a blood clot (embolism or thrombosis); we think that in CDG these episodes are due to a transient local thickening of blood. It can present in several ways: drowsiness, dullness, subcoma, coma, paralysis (on one side (hemiparesis, hemiplegia) or on both sides), loss of vision. Responder: - Jaak Jaeken, MD, Ph.D. 07/27/11

Is there a suggested therapy to prevent or help these episodes?

Medical treatment and prevention are possible but it is up to the treating physician to decide about the treatment. Responder: - Jaak Jaeken, MD, Ph.D. 07/27/11

What action should parents take during one of these episodes?

Action to be taken: measure body temperature and ask your (or another) physician to examine your child as soon as possible in order to make the diagnosis and to take appropriate measures. Responder: - Jaak Jaeken, MD, Ph.D. 07/27/11

What is the difference between a stroke-like episode and a seizure?

A seizure is the expression of an abnormal electrical activity in the brain (stroke is a vascular problem) but can resemble very much a stroke. Other words for "seizures" are "epilepsy" and "convulsions". Responder: - Jaak Jaeken, MD, Ph.D. 07/27/11

Who is likely to have a stroke-like episode and when are they likely to occur?

In the CDG field all the patients with CDG-Ia have an increased risk for thrombosis because their blood platelets have an increased tendency to stick together and to stick to the wall of blood vessels; for the other CDG-I patients this risk is probably also increased but this has not yet formally been proven also because we know only a small number of these patients. These episodes are most likely to occur on occasion of an infection (viral or bacterial); so these episodes are often accompanied by fever. Responder: - Jaak Jaeken, MD, Ph.D. 07/27/11

Are there ever any long lasting effects?

These episodes can last for hours, days or sometimes even longer. However, the positive thing is that, as a rule, they are transient. Responder: - Jaak Jaeken, MD, Ph.D. 07/27/11