Mayo Clinic Congenital Disorders of Glycosylation (CDG) Initial Clinic Preparation List

The CDG Clinic staff is happy you made an appointment at Mayo Clinic. Below is a checklist to help you prepare prior to your visit to maximize the benefits the clinic can offer. You can also reach out to CDG CARE at www.cdgcare.com or email frontiersincdg@cdgcare.org for more information.

This is an example of how your clinic appointment might be structured (scheduling times may be different):
8:30-12:00 - Blood draw and sample collection for laboratory studies, ECG (echocardiogram), abdominal ultrasound, fibroscan, ophthalmology, endocrine, physical therapy evaluation, etc, depending on the clinical features
1:00 - Family history and family tree analysis
1:30 - Clinical appointment with the CDG specialist, biochemical geneticist (physical exam, neurologic exam, Nijmegen CDG Score assessment, Goal Attainment Score assessment), including disease counseling
2:30 - Discussion of clinical trials, research opportunities
3:00 - Dietary evaluation by metabolic nutritionist
3:30 - Assessment by Pediatric Neurology (not in all cases, only if requested by referring physician or by the CDG specialist)

Things to prepare before the appointment

a) Medical records and information necessary before the first CDG clinic visit:
   ✓ Genetic test results (genetic variants found in blood) or biochemical test results (transferrin glycoform or CDG enzyme analysis in blood) that suggest you or your child has CDG
b) Medical records and information recommended before the first CDG clinic appointment
   ✓ Brain imaging results (MRI, CT), on CD if possible
   ✓ Developmental assessment
   ✓ Laboratory results (TSH, free T4, coagulation studies, transaminases, blood glucose, CBC)
   ✓ ECG (echocardiogram), liver ultrasound or fibroscan results
   ✓ Ophthalmology and hearing exam results
c) The clinic coordinators will send surveys to be filled out prior to the appointment. These include the PROMIS patient reported outcome survey and the 3-days food intake survey. Please fill in the surveys and bring it to the appointment, or send them electronically through the patient portal.
d) MAKE A LIST OF QUESTIONS YOU HAVE (diagnosis, symptoms related to CDG, treatment, clinical trials etc.)

Useful tips

a) Bring along a caretaker, if possible, to allow minimally interrupted discussions.
b) Bring some toys, books and snacks for the long day

The CDG Clinic staff looks forward to seeing you at Mayo Clinic.