



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.