



**EMORY GENETICS LABORATORY CDG (Congenital Disorders of Glycosylation)**

Please send the completed form with the sample  
Or fax to (404)778-8559

Clinical Data Information Sheet  
Patient Information Questionnaire

Patient Name: \_\_\_\_\_ DOB: \_\_\_\_\_

Referring Provider: \_\_\_\_\_ Phone: \_\_\_\_\_

Age at initial presentation: \_\_\_\_\_

**Presenting symptoms (check):**

**Neurology**

Psychomotor Retardation	Yes ___	No ___	Unknown ___
Epilepsia	Yes ___	No ___	Unknown ___
Stroke-like episodes	Yes ___	No ___	Unknown ___
Cerebellar atrophy	Yes ___	No ___	Unknown ___
Cerebral atrophy	Yes ___	No ___	Unknown ___
Microcephaly	Yes ___	No ___	Unknown ___
Ataxia	Yes ___	No ___	Unknown ___
Tetraspastic Paresis	Yes ___	No ___	Unknown ___
Muscular hypotonia	Yes ___	No ___	Unknown ___

**Eye**

Strabism	Yes ___	No ___	Unknown ___
Retinitis pigmentosa	Yes ___	No ___	Unknown ___
Cataracts	Yes ___	No ___	Unknown ___
Optical atrophy	Yes ___	No ___	Unknown ___
Coloboma	Yes ___	No ___	Unknown ___

**Skeletal/Growth**

Failure to thrive	Yes ___	No ___	Unknown ___
Growth retardation	Yes ___	No ___	Unknown ___
Short limbs	Yes ___	No ___	Unknown ___
Joint contracture	Yes ___	No ___	Unknown ___
Delayed puberty	Yes ___	No ___	Unknown ___

**Skin**

Inverted nipples	Yes ___	No ___	Unknown ___
Abnormal fat pads	Yes ___	No ___	Unknown ___
Cutis laxa/wrinkly skin	Yes ___	No ___	Unknown ___
Ichthyosis	Yes ___	No ___	Unknown ___

**Dysmorphology**

Dysmorphic facial features	Yes ___ (describe below)	No ___	Unknown ___
Genital anomalies	Yes ___	No ___	Unknown ___

**Coagulation**

Thrombosis'	Yes ___	No ___	Unknown ___
Bleeding tendency	Yes ___	No ___	Unknown ___
Decreased AT III activity	Yes ___	No ___	Unknown ___

Decreased clotting factor XI activity	Yes ___	No ___	Unknown ___
Decreased protein C	Yes ___	No ___	Unknown ___
<b>Renal</b>			
Congenital nephritic syndrome	Yes ___	No ___	Unknown ___
<b>Gastrointestinal</b>			
Chronic diarrhea	Yes ___	No ___	Unknown ___
<b>Heart</b>			
Cardiomyopathy	Yes ___	No ___	Unknown ___
Cardial effusion	Yes ___	No ___	Unknown ___
Hydrops fetalis	Yes ___	No ___	Unknown ___
<b>Additional information:</b>			
<b>Laboratory results</b>			
Carbohydrate deficient transferrin/Laboratory (or attach result): _____			
Lysosomal enzyme test/Laboratory (or attach result): _____			
Urinary oligosaccharide/Laboratory (or attach result): _____			
Liver enzymes/Laboratory (or attach result): _____			
FSH, LH, prolactin/ Laboratory (or attach result): _____			
Leukocytosis/ Laboratory (or attach result): _____			
Bombay blood group/ Laboratory (or attach result): _____			
<b>Other relevant results</b> (molecular studies, research results; include laboratory name & copy of results):			
<b>Family History:</b>			
	Significant (indicate below)	Not significant	