



**Instructions:** Complete all information below. **Send paperwork with the specimen, or return by fax to Mayo Clinic Laboratories, Attn: Biochemical Genetics Laboratory at 507-284-1759.** For questions or additional assistance, call 800-533-1710 and ask for the on-call Biochemical Genetics Counselor. **International clients call +1-507-266-5700 or email [mliint@mayo.edu](mailto:mliint@mayo.edu)**

### Patient Information

Patient Name (Last, First Middle)		Birth Date (mm-dd-yyyy)	
Sex Assigned at Birth <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Unknown <input type="checkbox"/> Choose not to disclose		Legal/Administrative Sex <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Nonbinary	

### Referring Healthcare Professional Information

Requesting Healthcare Professional Name (Last, First)	Phone	Email	Fax*
Genetic Counselor Name (Last, First)	Phone	Email	Fax*

\*Fax number given must be from a fax machine that complies with applicable HIPAA regulations.

### Reason for Testing

 Do not use this form for prenatal testing.

<input type="checkbox"/> Screening: _____	<input type="checkbox"/> Investigate molecular results (provide): _____
<input type="checkbox"/> Monitor treatment: _____	<input type="checkbox"/> Known diagnosis (provide): _____

### Clinical Information

 Complete below or fax most recent clinic note to 507-284-1759.

CDG presentations vary widely. Indicate the patient's clinical presentation by selecting the boxes below.

Age at presentation: \_\_\_\_\_ Molecular test results: \_\_\_\_\_

List any additional clinical findings, current medications, and diet:

\_\_\_\_\_

\_\_\_\_\_

<b>Skin</b>	<input type="checkbox"/> Abnormal fat distribution	<input type="checkbox"/> Cutis laxa	<input type="checkbox"/> Inverted nipples
	<input type="checkbox"/> Aplasia cutis congenita	<input type="checkbox"/> Hypoplastic nails	<input type="checkbox"/> Scaly skin or rashes
<b>Hematologic</b>	<input type="checkbox"/> Bleeding episodes	<input type="checkbox"/> Coagulation defects	<input type="checkbox"/> Thrombosis
<b>Heart</b>	<input type="checkbox"/> Cardiomyopathy	<input type="checkbox"/> Long QT syndrome	<input type="checkbox"/> Septal defects
	<input type="checkbox"/> Hypertension	<input type="checkbox"/> Patent ductus arteriosus	<input type="checkbox"/> Structural heart defects
<b>Eyes</b>	<input type="checkbox"/> Cataracts	<input type="checkbox"/> Optic atrophy	<input type="checkbox"/> Retinitis pigmentosa
	<input type="checkbox"/> Nystagmus	<input type="checkbox"/> Ptosis	<input type="checkbox"/> Strabismus
<b>Endocrine</b>	<input type="checkbox"/> Delayed puberty	<input type="checkbox"/> Hormonal dysregulation	<input type="checkbox"/> Infertility
			<input type="checkbox"/> Virilization
<b>Intestinal</b>	<input type="checkbox"/> Constipation	<input type="checkbox"/> Failure to thrive	<input type="checkbox"/> Hirschsprung
	<input type="checkbox"/> Diarrhea	<input type="checkbox"/> Feeding difficulties	<input type="checkbox"/> Protein-losing enteropathy
<b>Genito-urinary</b>	<input type="checkbox"/> Cryptorchidism	<input type="checkbox"/> Hydrocele	<input type="checkbox"/> Hypospadias
	<input type="checkbox"/> Hemolytic uremic syndrome	<input type="checkbox"/> Hydronephrosis	<input type="checkbox"/> Inguinal hernia
<b>Brain</b>	<input type="checkbox"/> Ataxia	<input type="checkbox"/> Cortical atrophy	<input type="checkbox"/> Hypotonia
	<input type="checkbox"/> Autism	<input type="checkbox"/> Developmental delay	<input type="checkbox"/> Intellectual impairment
	<input type="checkbox"/> Behavior issues	<input type="checkbox"/> Dysarthria	<input type="checkbox"/> Pain
	<input type="checkbox"/> Brain malformations	<input type="checkbox"/> Dyskinesia	<input type="checkbox"/> Paresthesia
	<input type="checkbox"/> Cerebellar hypoplasia	<input type="checkbox"/> Epileptic encephalopathy	<input type="checkbox"/> Seizures
<b>Immunologic</b>	<input type="checkbox"/> Autoimmune disorders	<input type="checkbox"/> Recurrent infections	<input type="checkbox"/> Splenectomy
			<input type="checkbox"/> Unexplained fever episodes
<b>Liver</b>	<input type="checkbox"/> Cholestasis	<input type="checkbox"/> Cirrhosis	<input type="checkbox"/> Hepatomegaly
			<input type="checkbox"/> Hepatopathy
<b>Skeletal</b>	<input type="checkbox"/> Abnormal teeth	<input type="checkbox"/> Contractures	<input type="checkbox"/> Pes cavus
	<input type="checkbox"/> Bony overgrowth	<input type="checkbox"/> Kyphoscoliosis	<input type="checkbox"/> Pes planus
<b>Lab Findings</b>	<input type="checkbox"/> Decreased protein C & S, AT-III, Factor VIII, Factor XI	<input type="checkbox"/> Elevated transaminases	<input type="checkbox"/> Hypogammaglobulinemia
		<input type="checkbox"/> Hypoalbuminemia	<input type="checkbox"/> Hypoglycemia
			<input type="checkbox"/> Low triglycerides
			<input type="checkbox"/> Manganese