



Instructions: Complete all information below. **Send paperwork with the specimen, or return by fax to MCL Biochemical Genetics Laboratory at 507-538-4158.** For questions or additional assistance, call 800-533-1710 and ask for the on-call Biochemical Genetics Counselor.

Patient Information

Patient Name <i>(Last, First, Middle)</i>		Birth Date <i>(mm-dd-yyyy)</i>
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 Provider Information

Referring Provider Name <i>(Last, First)</i>	Phone	Email	Fax*
Genetic Counselor Name <i>(Last, First)</i>	Phone	Email	Fax*

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

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 507-538-4158.

List relevant clinical information and the results of any applicable testing (screening and diagnostic):

Age at presentation: _____ Molecular test results: _____

Current medications and diet:

CDG presentations are variable. Below are possible findings.

Skin	<ul style="list-style-type: none"> Abnormal fat distribution Aplasia cutis congenita 	<ul style="list-style-type: none"> Cutis laxa Hypoplastic nails 	<ul style="list-style-type: none"> Inverted nipples Scaly skin or rashes
Hematologic	<ul style="list-style-type: none"> Bleeding episodes 	<ul style="list-style-type: none"> Coagulation defects 	<ul style="list-style-type: none"> Thrombosis
Heart	<ul style="list-style-type: none"> Cardiomyopathy Hypertension 	<ul style="list-style-type: none"> Long QT syndrome Patent ductus arteriosus 	<ul style="list-style-type: none"> Septal defects Structural heart defects
Eyes	<ul style="list-style-type: none"> Cataracts Nystagmus 	<ul style="list-style-type: none"> Optic atrophy Ptosis 	<ul style="list-style-type: none"> Retinitis pigmentosa Strabismus Visual impairment
Endocrine	<ul style="list-style-type: none"> Delayed puberty 	<ul style="list-style-type: none"> Hormonal Dysregulation 	<ul style="list-style-type: none"> Infertility Virilization
Intestinal	<ul style="list-style-type: none"> Constipation Diarrhea 	<ul style="list-style-type: none"> Failure to thrive Feeding difficulties 	<ul style="list-style-type: none"> Hirschsprung Protein-losing enteropathy Vomiting
Genito-urinary	<ul style="list-style-type: none"> Abnormal renal ultrasound Cryptorchidism 	<ul style="list-style-type: none"> Hemolytic uremic syndrome Hydrocele 	<ul style="list-style-type: none"> Hydronephrosis Hypospadias Inguinal hernia Multicystic kidneys
Brain	<ul style="list-style-type: none"> Ataxia Autism Behavior issues Brain malformations Cerebellar hypoplasia 	<ul style="list-style-type: none"> Cortical atrophy Developmental delay Dysarthria Dyskinesia Epileptic encephalopathy 	<ul style="list-style-type: none"> Hypotonia Intellectual impairment Pain Paresthesia Seizures Speech delay Stroke-like episodes Tremor
Immunologic	<ul style="list-style-type: none"> Autoimmune disorders 	<ul style="list-style-type: none"> Recurrent infections 	<ul style="list-style-type: none"> Splenectomy Unexplained fever episodes
Liver	<ul style="list-style-type: none"> Cholestasis 	<ul style="list-style-type: none"> Cirrhosis 	<ul style="list-style-type: none"> Hepatomegaly Hepatopathy
Skeletal	<ul style="list-style-type: none"> Abnormal teeth Bony overgrowth 	<ul style="list-style-type: none"> Contractures Kyphoscoliosis 	<ul style="list-style-type: none"> Pes cavus Pes planus Osteopenia/Osteoporosis
Lab Findings	<ul style="list-style-type: none"> Decreased protein C & S, AT-III, Factor VIII, Factor XI Elevated transaminases 	<ul style="list-style-type: none"> Hypogammaglobulinemia Hypoalbuminemia 	<ul style="list-style-type: none"> Hypoglycemia Low triglycerides Manganese