

Laboratory of Molecular Neurogenetics  
 Department of Pathology  
 Ali Naini, Ph.D., DABCC  
 www.columbiamitodiagnosics.org



COLUMBIA UNIVERSITY  
 MEDICAL CENTER

630 West 168<sup>th</sup> Street  
 VC 15<sup>th</sup> Floor, Room 208  
 New York, NY 10032  
 Tel: 212-305-3947  
 Fax: 212-305-3986

**REQUEST FOR MOLECULAR GENETIC TESTING FOR Disorders of glycogen/ lipid metabolism**

**(Must be completely filled out; Informed consent MUST be signed by patient, parent/legal guardian or legal next of kin.)**

<b>PATIENT INFORMATION:</b>		<b>REQUESTING PHYSICIAN:</b>	
Last Name:	First Name:	Last Name:	First Name:
Date of Birth:	Gender: <input type="checkbox"/> Male <input type="checkbox"/> Female	Institution:	
Address:		Address:	
City, State, and ZIP:		City, State, and ZIP:	
Telephone:		Telephone:	Fax:
<b>CUMC MRN (Unit number):</b>			
<b>INSTITUTIONAL BILLING (PREFERRED): CHARGES WILL BE BILLED TO THE SUBMITTING INSTITUTION/PHYSICIAN</b>		<b>ALTERNATE BILLING INFORMATION:</b>	
Institution:		Bill to: <input type="checkbox"/> CREDIT CARD <input type="checkbox"/> PATIENT (SELF PAY) <input type="checkbox"/> OTHER:	
Department:		Cardholder's Name:	
Address:		Credit Card Number:	
		Card Type: <input type="checkbox"/> AMEX <input type="checkbox"/> MASTERCARD <input type="checkbox"/> VISA <input type="checkbox"/> OTHER:	
Contact:		Expiration Date:	
Telephone:                      FAX:			
<b>TEST ORDERED: PCR/RFLP ANALYSES - \$250 PER MUTATION</b>		<b>TISSUE SUBMITTED</b>	
Myophosphorylase deficiency (McArdle's Disease) <input type="checkbox"/> Codon 49 <input type="checkbox"/> Codon 204 <input type="checkbox"/> Phosphofructokinase (PFK) deficiency (Exon 5 deletion) <input type="checkbox"/> Carnitine palmitoyl transferase II (CPT II) deficiency (Ser113Leu) <input type="checkbox"/> Phosphoglycerate mutase (PGAM) deficiency (Gene Sequencing)		<input type="checkbox"/> BLOOD <input type="checkbox"/> MUSCLE <input type="checkbox"/> OTHER: _____ Date Specimen Collected: _____ Date Specimen Sent: _____ Blood & Fluid Precautions? <input type="checkbox"/> Yes <input type="checkbox"/> NO	
<b>REASON FOR TESTING:</b>			
<input type="checkbox"/> Possible diagnosis of Glycogen Metabolism Disorder <input type="checkbox"/> Pre-symptomatic testing in an individual with a positive family history of _____			
Other relevant Clinical Information:		IDC9 Code(s):	
<p><b>Note to Health Care Practitioner:</b> It is New York State Law, and Columbia University Policy that an informed consent be obtained prior to performing genetic predisposition testing and maintained in the patient's medical record. Please use the appropriate disease/gene information/informed consent sheet, ensure that the patient/legal guardian understands its contents, and obtain the person's signature.</p>			

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**INFORMED CONSENT / ADVANCE BENEFICIARY NOTICE :**

**Please read the following carefully and discuss with your ordering physician/person obtaining consent before signing consent.**

1. DNA analysis using direct sequencing and/or other methods to detect genetic abnormalities of the enzymes responsible for normal glycogen metabolism.
2. The purpose of this analysis is to test for genetic disorders of glycogen metabolism, which normally supplies energy through sugar (glucose) metabolism.
- 2a. You (or the person for whom you are signing) may want genetic counseling before signing for consent.
3. This is a test for genetic susceptibility ("genetic predisposition"). The risk of having the disorder may be altered by family history and/or other factors. If the test is positive for the disorder or for an increased risk of the disorder, you may wish to have further independent testing, consult your physician or have genetic counseling.
4. The condition being tested is abnormal glycogen or lipid metabolism, which can lead to exercise intolerance, muscle pain or damage, and weakness.
5. A positive test increases the likelihood of defective glycogen metabolism. When a mutation is found in **both copies** of the gene, there is **a greater than 90% risk** of glycogenoses. When only one abnormal gene copy is detected, the likelihood of disease is low, as the person is probably a carrier. A negative result (no mutations detected) largely, but does not entirely, exclude the possibility of disease, as other mutations or tissues other than those tested could be affected.
6. The results of the above test become a part of the patient's medical record, and may be made available to individuals/organizations with legal access to the patient's medical record, on a strict "need-to-know" basis, including, but not limited to the physicians and nursing staff directly involved in the patient's care, the patient's current and future insurance carriers, and others specifically authorized by the patient/authorized representative to gain access to the patient's medical records.
7. No additional tests will be performed on this sample, without specific, signed authorization by the patient. After 60 days, unless consent is given the sample will be destroyed – please see below.
8. Medicare/Insurance Carriers may not pay for the test, in which case, the patient/responsible party will be billed for the test.

**Requesting Physician or Licensed Nurse Practitioner:**

**Name:** \_\_\_\_\_ **Title:** \_\_\_\_\_

**Name of person obtaining consent:** \_\_\_\_\_ **Signature:** \_\_\_\_\_  
**Date:** \_\_\_\_\_

I have read and fully understood the above, and give my consent for this testing.

**Patient name:** \_\_\_\_\_  
**Patient signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_

**If consent is given by parent or legally authorized representative:**  
**Name:** \_\_\_\_\_ **Relationship:** \_\_\_\_\_  
**Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_

**Consent for sample retention:**

**I consent to the retention of this blood for: (check and initial on appropriate lines)**

\_\_\_\_\_ My specimen may be used for routine laboratory use only. After 60 days, unless consent is given the sample will be destroyed.

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## GLYCOGENOSIS/LIPIDOSIS PATIENT DATA ENTRY FORM

### McArdle's Disease, PFK, PGAM, and CPT II Deficiencies

Patient Name: \_\_\_\_\_ Date: \_\_\_\_\_

#### **Clinical Information:**

Clinical diagnosis: \_\_\_\_\_ Age of onset: \_\_\_\_\_ Ethnic background: \_\_\_\_\_

Clinical features (circle appropriate responses; Y = yes; N = no; NA = information not available)

#### **Symptoms**

Exercise intolerance	Y	N	NA	
Cramps after exercise	Y	N	NA	
Myoglobinuria	Y	N	NA	If Yes, how many episodes? _____
Proximal limb weakness	Y	N	NA	
Neuropathy	Y	N	NA	If Yes, Neuropathy type: _____

#### **Signs**

Floppy baby	Y	N	NA
Developmental delay	Y	N	NA
Seizures	Y	N	NA
Headache	Y	N	NA
Ophthalmoplegia	Y	N	NA
Ptosis	Y	N	NA
Retinopathy	Y	N	NA
Asthenia	Y	N	NA
Congestive heart failure	Y	N	NA
Nausea/vomiting	Y	N	NA
Resp. insufficiency	Y	N	NA
Hypothyroidism	Y	N	NA
Diabetes mellitus	Y	N	NA
Other:	_____		

#### **LABORATORY STUDIES:**

Resting serum creatine kinase                      Normal                      Abnormal                      NA  
 If elevated, state CK level outside episodes of myoglobinuria: \_\_\_\_\_

Ischemic Exercise Test                      Normal                      Abnormal                      NA  
 If abnormal, is there no rise of lactate (i.e. flat response)? \_\_\_\_ abnormally low rise of lactate? \_\_\_\_  
 Times above baseline: \_\_\_\_\_

Resting serum lactate level                      Normal                      Abnormal                      NA  
 If elevated, level: \_\_\_\_\_

ECG                      Normal                      Abnormal                      NA

EMG/NCS                      Normal                      Abnormal                      NA