

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



COLUMBIA UNIVERSITY
 MEDICAL CENTER

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

REQUEST FOR BIOCHEMICAL 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.)

PATIENT INFORMATION:		REQUESTING PHYSICIAN:	
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:
CUMC MRN (Unit number):			
INSTITUTIONAL BILLING(PREFERRED): CHARGES WILL BE BILLED TO THE SUBMITTING INSTITUTION/PHYSICIAN		ALTERNATE BILLING INFORMATION:	
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:			
TEST ORDERED (FILL IN COMPLETELY):		SPECIMEN REQUIREMENTS	
<input type="checkbox"/> GSD II Acid maltase deficiency (Pompe's disease) <input type="checkbox"/> GSD III Glycogen debrancher deficiency (Cori-Forbes' deficiency) <input type="checkbox"/> GSD IV Glycogen branching enzyme (Andersen's disease) <input type="checkbox"/> GSD V: McArdle's Disease, Myophosphorylase (PPL) deficiency <input type="checkbox"/> GSD VII: Tarui's disease, Phosphofructokinase (PFK) deficiency <input type="checkbox"/> GSD VIII: Phosphorylase b kinase (PHK) deficiency <input type="checkbox"/> GSD IX: Phosphoglycerate kinase (PGK) deficiency <input type="checkbox"/> GSD X: Phosphoglycerate mutase (PGAM) deficiency <input type="checkbox"/> GSD XI: Lactate dehydrogenase (LDH) deficiency <input type="checkbox"/> Carnitine palmitoyl transferase II (CPT II) deficiency		We require frozen muscle (approximately. 200 mg), which should be snap frozen in liquid nitrogen and shipped frozen on dry ice. Date Specimen Collected: _____ Date Specimen Sent: _____ Blood & Fluid Precautions? <input type="checkbox"/> Yes <input type="checkbox"/> No	
REASON FOR TESTING:			
<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>Note to Health Care Practitioner: 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. Biochemical analysis using enzyme function assays will measure the function 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 an abnormal glycogen or lipid metabolism that affect or could lead to exercise intolerance, muscle pain or damage, and weakness.
5. A positive test means likelihood of having defective glycogen or lipid metabolism is greater than 90%. A negative result does not 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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TESTS REQUESTED (please check appropriate boxes)

I. GLYCOLYTIC ENZYME DEFICIENCIES ASSOCIATED WITH: EXERCISE INTOLERANCE / MYALGIAS / MYOGLOBINURIA

(GSD V; McArdle's disease)	Myophosphorylase	(PPL)	<input type="checkbox"/>
(GSD VII; Tarui's disease)	Phosphofructokinase	(PFK)	<input type="checkbox"/>
(GSD VIII)	Phosphorylase b kinase	(PHK)	<input type="checkbox"/>
(GSD IX)	Phosphoglycerate kinase	(PGK)	<input type="checkbox"/>
(GSD X)	Phosphoglycerate mutase	(PGAM)	<input type="checkbox"/>
(GSD XI)	Lactate dehydrogenase	(LDH)	<input type="checkbox"/>

PGK, PGAM, LDH	3 enzyme battery	<input type="checkbox"/>
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PPL, PFK, PHK, PGK, PGAM, LDH	6 enzyme battery	<input type="checkbox"/>
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II. GLYCOLYTIC ENZYME DEFICIENCIES ASSOCIATED WITH WEAKNESS

(GSD II; Pompe's disease)	acid maltase	<input type="checkbox"/>
(GSD III; Cori-Forbes' disease)	debrancher	<input type="checkbox"/>
(GSD IV; Andersen's disease)	branching enzyme	<input type="checkbox"/>

III. DISORDERS OF LIPID METABOLISM

Carnitine palmitoyl transferase II	(CPT II)	<input type="checkbox"/>
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Charges: **Glycolytic enzymes:** **\$125 per enzyme assayed**
 CPT activity: **\$200**

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GLYCOGENOSIS/LIPIDOSIS PATIENT DATA ENTRY FORM

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

Muscle Morphology:

Positive family history: Y N NA If yes, please explain: