

ROBERT GUTHRIE BIOCHEMICAL & MOLECULAR GENETICS LABORATORY

Georgirene D. Vladutiu, PhD, Director

THE BUFFALO GENERAL HOSPITAL

KALEIDA HEALTH

100 High Street, Room A762, Buffalo, NY 14203

Telephone: (716) 859-7741 • Fax: (716) 859-7749

www.rgbgl.org

CLIA License #33D0685375 • New York State Laboratory Permit # PF12005

CONSENT FOR
DNA TESTING
Page 1 of 4

I, _____, hereby request DNA based testing for one or all of the following disorders (myoadenylate deaminase deficiency, carnitine palmitoyltransferase (CPT II) deficiency, or myophosphorylase deficiency, also known as McArdle disease). I understand that biological samples of blood, cells, or tissue, as indicated below, will be provided for this testing by me/or my relative as follows:

Name of Individual to be Tested

Date of Birth

Relationship

Please circle the **test(s)** for which you are providing consent:

- (a) Myoadenylate deaminase deficiency
- (b) CPT II deficiency
- (c) Myophosphorylase deficiency
- (d) *CPT2* gene sequence analysis

Please circle the **specimen(s)** that will be provided for testing:

- (a) Whole blood
- (b) Muscle tissue
- (c) Cultured fibroblasts
- (d) DNA extracts

1. Description of the Diseases

The purpose of this testing is to identify mutations in genes that cause metabolic muscle diseases characterized primarily by exercise intolerance.

(a) **Myoadenylate deaminase (AMPD) deficiency:** This disorder is the most common metabolic muscle disease in humans occurring in approximately 1 in 50 individuals and is characterized by muscle pain and cramps with physical exertion. Symptoms occur when the enzyme, myoadenylate deaminase, is absent in the muscle tissue of an individual who has inherited 2 mutations or alterations in the gene (genetic material known as DNA) causing this enzyme deficiency.

(b) **Myophosphorylase deficiency (McArdle Disease; Glycogen Storage Disease Type V):** This is a disorder of energy production that is characterized by pain and cramps with physical exertion. Symptoms occur when the enzyme, myophosphorylase, is absent or greatly diminished in muscle tissue of an individual who has inherited 2 mutations or alterations in the gene (genetic material known as DNA) causing this enzyme deficiency.

(c) **Carnitine Palmitoyltransferase (CPT) II Deficiency:** This is a disorder of energy production that is characterized by pain and cramps with physical exertion. Symptoms occur when the enzyme, CPT II, is absent or greatly diminished in muscle tissue of an individual who has inherited 2 mutations or alterations in the gene (genetic material known as DNA) causing this enzyme deficiency.

2. Specimens Used for Testing

DNA will be extracted from you/your child/relative from **blood** collection (5 to 7 cc or 1-2 teaspoons) taken from your vein, or from **muscle biopsy** tissue if already available from previous biochemical studies, or from **cultured skin fibroblasts** (if already available from earlier testing in another laboratory), or from any source of **DNA extracts** prepared by another laboratory and released to our laboratory with your permission. There is no risk to you/your child/relative of blood collection with the exception of a possible small bruise at the site of blood draw. There is no risk associated with the collection of cheek cells. All other specimens listed above would be pre-existing specimens with no associated additional risk from this testing. You may also be asked to provide information regarding the medical history that led you/your doctor to recommend this testing.

3. Description and Principle of the Testing

Testing for the diagnosis of one or more of the disorders above involves the use of specialized procedures that will analyze your DNA to detect the presence of one or more specific causative mutations. Unless specifically indicated, this test is not part of a research protocol and is being used for clinical purposes. The testing is very accurate and sensitive for the diagnosis of each disorder with detection in 80 to 95% of cases in which the specified mutation is present depending on the disorder being evaluated. The test report will state the level accuracy with which each disorder has been ruled out or diagnosed and whether or not further testing will be necessary to confirm or refute the diagnosis.

4. The Meaning of the Test Results

(a) **If one mutation is present** in your DNA for one or more of the disorders listed above:

- You may be a carrier for the disorder(s) and you may have symptoms as a carrier under certain conditions.
- Or you may be a carrier without symptoms.
- Or you may have a second mutation in the designated gene that was not detected by the testing and you, therefore, may require further biochemical or molecular testing for a diagnosis.

(b) **If two mutations are present** in your DNA for one or more of the disorders listed above, then this finding will constitute a diagnosis of the disorder(s).

(c) **If no mutations are present** in your DNA it does not mean that the risk of carrying or developing the disorder is no longer present. It only means that the mutations searched for were not found and others may be present and remain unidentified. You may need further biochemical or molecular testing for a diagnosis.

5. Cost of Testing

You will be responsible for payment of fees related to this testing. Because this is relatively new testing, it is possible that your health insurance company will not reimburse you for the related costs.

6. Confidentiality

Because DNA testing and the interpretation of the results are complex, the information obtained from this test will be provided to your physician who will then inform you of the results either directly or through a genetic counselor. It will be your physician's obligation to provide you with any genetic counseling pertaining to the results and potential consequences of these test results. To the extent required by law, all of the records, findings, and results of this testing are considered and shall remain confidential and shall not be disclosed to anyone without your written consent. In addition, the results can only be released by law to (a) insurance companies and other third party payers, such as Medicaid if necessary for the payment of services to you and (b) to any person to whom a court orders disclosure under limited circumstances set forth by law.

7. Use of Your DNA Specimen After Testing is Completed

Once your test results are completed, additional testing may or may not be performed on your specimen depending on which option you choose below. Please select one of the following options:

- (a) **My biological sample will be destroyed within 60 days of testing.**

Initials

OR

- (b) **An aliquot of my biological sample may be stored as long as deemed necessary for general research purposes with the retention of my name on the sample.**

I understand that the Robert Guthrie Biochemical & Molecular Genetics Laboratory is not performing a DNA banking service by retaining my specimen. I consent to future contact for any and all purposes related to further diagnostic studies performed including the provision of general information about the findings; information about specific testing of my sample that may benefit me or my family members in relation to my choices regarding preventive or clinical care; and I understand there is no additional risk to me for consenting to future contact and complete confidentiality will continue to be maintained. The potential benefits of further clinical studies of my sample may provide additional information that impacts favorably on my future health care.

Initials

- (c) **An aliquot of my biological sample may be stored as long as deemed necessary for additional research studies, however, all identifiers, including my name, must be removed before initiating the research studies.** I understand that information about specific testing of my sample will not be available because the sample will not be individually identifiable.

Initials

I further understand that participation in DNA-based testing is completely voluntary and I have had the opportunity to have all of my associated questions answered by my physician or a genetic counselor. I also may withdraw my consent in writing for any and all testing at this time.

Signature: _____

Name (Print): _____

Witnessed by: _____

Date: _____

Physician's/Counselor's Statement: I attest to the fact that I have explained DNA-based testing to this individual. I have addressed the limitation of the testing as outlined above, and have answered any questions posed by the individual regarding the testing. I request that the above indicated genetic testing be performed.

Signature: _____ Name (print): _____

Date: _____

Please **Print** the name, address and phone number of all physicians to whom you authorize the release of test results. Any further disclosure of test results to person or organizations not listed below will require further informed consent at a later date.

Name of Licensed Physician	Address	Telephone Number
_____	_____	_____
_____	_____	_____
_____	_____	_____