

ROBERT GUTHRIE BIOCHEMICAL & MOLECULAR GENETICS LABORATORY

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CONSENT FOR COENZYME
Q10 TESTING IN MUSCLE
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I, _____, hereby request testing for Coenzyme Q10 deficiency in my muscle biopsy.
I understand that biological samples of muscle tissue will be provided for this testing by me or my relative as follows:

Name of Individual to be Tested	Date of Birth	Relationship
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(1) Description of Coenzyme Q10 Deficiency

The purpose of this testing is to identify a deficiency in a substance known as Coenzyme Q10. Decreased levels of Coenzyme Q10 in muscle have been reported in patients with gait disturbances (that is, in problems with walking), and in individuals with seizures. Some affected individuals may also have muscle weakness, exercise intolerance, recurrent coca cola colored urine, or development delay.

Coenzyme Q10 deficiency may be a serious inherited disorder considered as a primary deficiency, or may occur secondary to other disorders. It may also be an acquired condition caused by the use of certain medications such as lipid-lowering drugs.

Primary coenzyme Q10 deficiency is rare and is characterized by significant variations in clinical symptoms varying from central nervous system symptoms to widespread multi-system disease (that is, affecting many organs and tissues), such as cardiovascular disease and hypertension.

(2) Specimens Used for Testing

Coenzyme Q10 will be analyzed from your muscle biopsy tissue. The muscle specimen is usually obtained for multiple tests of which Coenzyme Q10 is only one, therefore, no associated additional risk to you exists 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

Coenzyme Q10, also known as Ubiquinone, is a vitamin-like substance present in all cells of the body and is a powerful antioxidant essential for cellular health and energy production (ATP). Coenzyme Q10 deficiency may be caused by insufficient dietary Coenzyme Q10, impairment in Coenzyme Q10 biosynthesis, excessive utilization of Coenzyme Q10 by the body, or any combination of the three. Deficiencies also have been reported in association with reduced activities of other enzymes in the mitochondrion, a tiny organelle found in body cells that is considered to be the "factory" for energy production.

(4) The Meaning of the Test Results

Abnormal: If the Coenzyme Q10 activity falls below the normal reference range, the results will be considered abnormal and reported as a deficiency in Coenzyme Q10. This finding may be clinically significant. Your physician will discuss the consequences of this finding and possible associated therapy. A limitation of this finding may be that the distinction between primary and secondary causes of Coenzyme Q10 deficiency may not be determined by this testing.

Normal: If the Coenzyme Q10 activity falls within the normal reference range, the results will be reported as normal.

(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

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 an genetic counseling pertaining to the results and potential consequences of these test results, such as therapy. 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 Muscle Specimen After Testing is Completed

Once your test results are completed, the fate of any remaining muscle tissue must be determined. Please select and initial 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 muscle tissue 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

I further understand that participation in Coenzyme Q10 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 Coenzyme Q10 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

_____	_____	_____
_____	_____	_____
_____	_____	_____