

Laboratory of Molecular Neurogenetics
 Department of Neurology
 Ali Naini, Ph.D., DABCC
 www.lomn.org



COLUMBIA UNIVERSITY
 MEDICAL CENTER

1150 Saint Nicholas Avenue
 Russ Berrie, Room 318
 New York, NY 10032
 Tel: 212-305-3947
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REQUEST FOR MOLECULAR GENETIC TESTING FOR: EARLY-ONSET TORSION DYSTONIA (DYT1)

(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:		SPECIMEN REQUIREMENTS	
DYT1 GENE MUTATION		Whole blood: Two 5.0 - 7.0 mL ACD (yellow top) or EDTA (lavender top) delivered within 24-48 hours at room temperature	
		Date and time collected: _____	
REASON FOR TESTING:			
<input type="checkbox"/> Molecular confirmation of early-onset torsion dystonia			
<input type="checkbox"/> Pre-symptomatic testing for early-onset torsion dystonia in an individual with a positive family history			
Race/ethnicity (OPTIONAL) _____			
Other relevant Clinical Information:		IDC9 Code(s):	
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.			

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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. This is a test for an abnormality (mutation(s)) in the *DYT1* gene for Early-Onset Torsion Dystonia using direct DNA sequencing.
2. The purpose of this analysis is to test for small deletion in gene for *DYT1*.
- 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 *DYT1* mutation, which can lead to Early-Onset Torsion Dystonia, a neurologic muscle disorder of involuntary and sometimes painful muscle contractions.
5. . If the *DYT1* gene mutation is detected in a person with disease symptoms, the likelihood of diagnosis is nearly 100%. A positive result in an individual without symptoms means a 30-40% risk of the disorder. Rare variations in the DNA of individuals can cause uncertainty in predicting carrier status or diagnosis. Therefore, the test is not 100% accurate, and the results will be reported as a probability.
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 line)

_____ 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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DNA-BASED ANALYSIS FOR EARLY-ONSET TORSION DYSTONIA

BACKGROUND: Early-onset dystonia represents the most severe and most common form of hereditary dystonia. The gene (DYT 1) responsible for the disease has been localized to the long arm of chromosome 9q34. Symptoms usually begin in an arm or leg at approximately 12 years (this can range between 4 and 44 years) and spread to other limbs in about 5 years (Bressman et al, 1994). The disease is dominantly inherited with 30-40% penetrance. Almost all cases of early-onset dystonia have a unique 3-bp deletion that appears to have arisen independently in different ethnic populations (Ozelius et al, 1997).

INDICATIONS FOR TESTING:

- Confirmation of clinical diagnosis of Torsion Dystonia
- Presymptomatic testing for Torsion Dystonia in individual with positive family history.

METHODOLOGY: Direct Mutation Analysis: DNA is extracted from nucleated cells in the blood. Specific region of DYT 1 gene is amplified, using polymerase chain reaction (PCR). Amplified DNA is cut with a restriction enzyme (BseR1) and run on agarose gel. Those patients who have the mutation within DYT 1 gene will have altered enzyme cutting sites which can be directly identified.

PRICE: \$225 per specimen

ACCURACY: >99% detection

SPECIMEN REQUIREMENTS: Draw two 5.0 mL lavender-top (EDTA) tubes of whole blood and invert several times to mix. Two 7.0 mL yellow-top (ACD) of whole blood is also acceptable. Forward within 24-48 hours via overnight carrier at ambient temperature. Requisition and signed consent forms must accompany specimens.

REFERENCES:

1. Bressman SB et al. Dystonia in Ashkenazi Jews: Clinical characterization of a founder mutation. *Ann Neurol* 36, 77 1-777 (1994).
2. Ozelius LJ et al. The early-onset torsion dystonia gene (DYT1) encodes an ATP-binding protein. *Nature Genet* 17, 40-48 (1997).