

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



COLUMBIA UNIVERSITY
 MEDICAL CENTER

630 West 168th Street
 VC 15th Floor, Room 208
 New York, NY 10032
 Tel: 212-305-2118
 Fax: 212-305-3986

REQUEST FOR MOLECULAR GENETIC TESTING FOR SOD1 GENE ANALYSIS FOR AMYOTROPHIC LATERAL SCLEROSIS
(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:	
DNA SEQUENCING FOR SOD1 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"/> Genetic screening for SOD1 mutation in a patient with ALS.			
<input type="checkbox"/> Pre-symptomatic testing of in an individual with a family history of ALS.			
FAMILY HISTORY <input type="checkbox"/> YES <input type="checkbox"/> NO			
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 genetic (DNA-based) test for ALS (also known as Lou Gehrig's Disease) using direct sequencing of the Cu/Zn superoxide (*SOD1*) gene.
2. The purpose of this analysis is to test for *SOD1* gene mutation.
 - 2a. You (or the person for whom you are signing) may want genetic counseling before signing for consent.
3. A negative results (absence of identified *SOD1* mutation) largely (but never completely) excludes the chance of developing ALS due to *SOD1* mutations. A negative result does not, however, exclude the possibility of developing ALS, since less than 5% of ALS cases are related to *SOD1* mutations. A positive result of this test (presence of *SOD1* mutation) indicates a greater than 90% risk of developing ALS at some time in life. A positive result cannot predict when symptoms of ALS would begin. If the test is positive, you may wish to have further independent testing, consult your physician and/or have genetic counseling.
4. I will ask for blood samples from an affected family member, if possible. This may increase the accuracy of my result.
5. I will receive a copy of this consent form.
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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SUPEROXIDE DISMUTASE MUTATION ANALYSIS AS A SCREENING TEST FOR FAMILIAL AMYOTROPHIC LATERAL SCLEROSIS

DESCRIPTION:

Amyotrophic Lateral Sclerosis (ALS, also known as Lou Gehrig's Disease) is a rapidly progressing, invariably fatal neurodegenerative disease that affects motor neurons. Affected individuals lose control of voluntary muscles, leading to inability to move and breathe without a ventilator. ALS does not affect sensory or autonomic neurons. About 5-10% of ALS cases are familial, predominantly with an autosomal dominant pattern of inheritance. Pathogenic mutations in the Cu/Zn superoxide dismutase (*SOD1*) gene have been described in about 20% of patients with familial ALS (FALS). The genetic test is diagnostic for ALS, but is occasionally performed for predictive purposes.

REASONS FOR REFERRAL:

- Confirmation of diagnosis in affected individuals.
- Presymptomatic screening in individuals with a confirmed family history of familial ALS.

TESTING:

The five exons of *SOD1* gene are PCR amplified using specifically designed primers. The products of the amplification are then sequenced to search for mutations. **Accuracy: 99%**

PRICE: \$ 450.00 per sample

SPECIMENT REQUIREMENT:

Draw two 7.0 mL yellow-top tubes (ACD) and invert several times to mix. Two 5.0 mL lavender-top tubes (EDTA) of whole blood are also acceptable. Please ship the sample via an express courier within 24-48 hours at **ambient temperature**. Results will be available in 3-4 weeks.