

## SUNY Upstate Medical University

## Authorization for the Molecular Test for Alpha-1 Antitrypsin Deficiency

**1. What is alpha-1 antitrypsin deficiency?** Alpha-1 antitrypsin is a substance normally made in the liver that inhibits the activity of a number of specific protein destroying enzymes. Alpha-1 antitrypsin deficiency can be due to either a lack of sufficient quantity of alpha-1 antitrypsin, or, if the level is sufficient, the function of the enzyme may be impaired. Alpha-1 antitrypsin deficiency is associated with such findings as chronic obstructive lung disease, emphysema, cirrhosis of the liver in children, liver cancer in adults, and asthma.

**2. What is the purpose of the test and what are its limitations?** Over 75 different genetic forms of alpha-1 antitrypsin have been identified, but only 2 are commonly associated with disease (PI S and PI Z forms). The molecular test for alpha-1 antitrypsin deficiency performed at the University Hospital will detect these two abnormalities as well as the normal gene. However, since other technologies are usually utilized to diagnosis alpha-1 antitrypsin deficiency in adults, the molecular test is intended to be used in prenatal situations where both parents are known carriers of alpha-1 antitrypsin deficiency and so are at risk of having an affected child. Since only 2 of the over 75 known mutations can be tested for, a negative test does not rule out a diagnosis of alpha-1 antitrypsin deficiency. A positive result by itself should not be used as the sole criteria for diagnosis. Rarely (less than 1% of the time), errors may occur, for example, due to sample mix ups, or due to technical errors such as rare genetic variants that mimic or mask the mutation being tested.

**3. What is required to perform the test?** A 10 ml sample of blood is necessary. This is equal to about two teaspoons. For prenatal diagnosis, 10-20 ml (2-4 teaspoons) of amniotic fluid collected by amniocentesis is necessary, and submission of parental blood samples is also required. In addition, you may be asked to provide information regarding your medical history. A correct history is critical for proper interpretation of the data.

**4. Is there a cost for the test?** This is a routine clinical laboratory test and the results from it may aid in diagnosis, so you or your health insurer will be billed for the procedure.

**5. What will happen to the DNA once the test is complete?** The only testing that will be performed on this sample is the test for alpha-1 antitrypsin deficiency. Residual DNA may be stored indefinitely (this does not constitute DNA banking) to be used use as a laboratory control in which case all identifying information will be removed.

**6. How will I obtain results from the test?** The test result will be provided to your physician who will discuss it with you. Genetic counseling may also be appropriate as follow-up. To the extent permitted by law, all of the records, findings, and results of this test are confidential and shall not be disclosed without your written consent specifically authorizing to whom such records, findings, and results are to be released.

You should consult your physician. If you have any questions about the test to be performed. You may also wish to obtain genetic counseling prior to signing this form. You may also contact the Molecular Diagnostics Laboratory at 315 464-6806.

\_\_\_\_\_  
Patient's name (**printed**)

\_\_\_\_\_  
Patient's Medical Record Number  
(for office use only)

**For the Patient:**

Please print the name, phone number, and address (if known) of all health care professionals, physicians (other than the referring physician), or other individuals/organizations (such as a health insurer) to whom you authorize the release of the alpha-1 antitrypsin deficiency test result. (Medical results cannot be sent to a patient, a patient's family member, or guardian.) Please print legibly.

Name and title	Address	Phone Number
_____	_____	_____
_____	_____	_____
_____	_____	_____
_____	_____	_____
_____	_____	_____

My signature below indicates that the above information has been explained to me and that I give consent for this alpha-1 antitrypsin deficiency testing.

Date: \_\_\_\_\_

\_\_\_\_\_  
**Signature of Patient**

\_\_\_\_\_  
Name of Parent/Guardian

\_\_\_\_\_  
**Signature** of Parent/Guardian if patient is a minor

*As referring physician/health care professional, I understand the benefits and limitations of this clinical assay. I hereby attest to the fact that I have provided the patient or patient's guardian with the information contained above in compliance with the NYS Civil Rights Act, Section 79-L, have answered any questions fully, and have obtained a signed informed consent as appropriate. I request that the above indicated genetic test be performed.*

\_\_\_\_\_  
**Printed** name of Physician/ Health Care Professional

\_\_\_\_\_  
**Signature** of Physician/Health Care Professional