ALPHA 1-ANTITRYPSIN DEFICIENCY REGISTRY SCREENING FORM

Form Completion Instructions:

This form is to be used to assist in the screening process. It should be completed on all patients who are considered for the Registry. This form may be completed and blood drawn at the same time as the initial clinic visit and its associated forms. In this instance, forward initial visit forms to the Clinical Coordinating Center. If the patient is not eligible, still send the complete set of Initial Visit Forms.

QUESTION #	ITEM	INSTRUCTIONS
3	Birthdate	If the birthdate indicates that the patient is not yet 18 years old, then, the patient is not eligible for further study at this time. Complete Form #01 only. The birthdate should match the one completed on the central lab form. If not, indicate which is correct.
4, 5	Sex, Race	For these and other such questions, a check mark should be placed in the dash preceding one of the possible options. These items are required.
6a,b,c	Therapy	This is the date the patient first began augmentation therapy and the type and frequency of the therapy, at that time. 6a,b,c should be skipped if the first response is no.
7	Informed Consent	If the patient refused consent for bloodwork and/or participation in the Registry, skip to item #16. You should still complete and send this form to the Clinical Coordinating Center.
8	Kit Number	The kit number is a four (4) digit number pre-printed on each kit sent from the Central Laboratory.

ALPHA 1-ANTITRYPSIN DEFICIENCY REGISTRY SCREENING FORM

Form Completion Instructions:

QUESTION #	ITEM	INSTRUCTIONS
9	Registry ID Number	The Registry ID number is a five (5) digit code number assigned at the Clinical Center. The first two (2) digits are the Clinical Center code number. (Remember to fill in the first blank with a zero if your Clinical Center number is between 1-9. For example: If your Clinical Center number is #3, the first two digits in the Registry ID number should be 03.) The remaining three digits should be assigned sequentially, 001, 002, 003 and so on. Be careful to keep a running record of Registry ID numbers assigned to be sure no duplication occurs. If an ID is assigned and it should not have been, DO NOT re-assign the ID to anyone else.
10	Name Code	The name code should consist of the first two letters of the patient's first name and the first two letters of the patient's last name. **Example:** Mary Jones = MAJO Within each center you should have unique name codes. Do not allow more than one patient to have the same code. Use a different letter if the situation arises.
11	Alpha 1-Antitrypsin	If the patient is not on augmentation therapy, record the alpha 1-antitrypsin serum level indicated on the Central Laboratory Report form and the date when the Central Laboratory

ALPHA 1-ANTITRYPSIN DEFICIENCY REGISTRY SCREENING FORM

Form Completion Instructions:

QUESTION #	ITEM	INSTRUCTIONS
11	Alpha 1-Antitrypsin (continued)	completed the analysis. For patients already receiving augmentation therapy, therapy must be stopped long enough for Alpha 1 levels to return to pre-therapy levels. If on weekly therapy, the patient must stop therapy for four weeks, draw blood for the Central Laboratory and then patient may resume augmentation therapy. If on monthly therapy, the patient should skip one scheduled therapy. Blood should be drawn just prior to the following therapy.
12	Phenotype	The Central Laboratory will indicate in the report the phenotype of the patient. Record this phenotype in the space provided. A list of the high risk phenotypes can be found on page 4.

At this point, eligibility can be reconfirmed:

- Is the patient 18 years or older?
- Is the reported level of alpha 1-antitrypsin ≤ 11.0 micromolar?
- Is the reported phenotype identified as "high risk?" (see attached list of high risk phenotypes on page 4.)

The patient is eligible for the Registry if the answer to all three (3) of the above questions is YES.

QUESTION #	ITEM	INSTRUCTIONS
13	Family History	It is VERY important to complete this portion of the form as completely and carefully as possible. If any known blood members of the patient's family have the deficiency, list their relationship to the patient (i.e., sister, grandmother, etc). In addition, if this family member is part of the Registry,

ALPHA 1-ANTITRYPSIN DEFICIENCY REGISTRY SCREENING FORM

QUESTION #	ITEM	INSTRUCTIONS
13	Family History (continued)	either at your Clinical Center or another Clinical Center in the country, indicate their Registry ID number.
14	Referral	A check should be placed in only one of the possible options. Indicate how the referral was made.
15	Ascertainment	Identify what lead to the suspicion and diagnosis of the deficiency in this patient.

(1)Screening of blood. Actual screening of blood for the deficiency is an asymptomatic patient with no family history is very rare. This response is indicated only if the patient has no pulmonary symptoms, no liver disease, has not had any abnormal lung function test and has no known relatives with the deficiency. One situation where this might occur would be a community event where there is free blood testing offered.

(2)Pulmonary symptoms lead to testing of patient. This response is used if the patient has any pulmonary symptoms. This response takes precedence over other responses. If more than one response is true and the patient has pulmonary symptoms then this is the response that should be indicated.

(3)Identified as a family member of a known alpha 1-antitrypsin deficient patient. This response is checked if this is the only reason the patient was tested.

(4)Other reason for suspecting deficiency. (specify:_____): This response should be rarely be used and is indicated only if the patient has no pulmonary symptoms, no abnormal lung functions test, no liver disease and no known family member with the deficiency. One situation under which this response would be appropriate is if the deficiency is discovered when the patient is being worked-up for some other non-lung or non-liver health condition. The exact reason should be indicated in the space provided.

ALPHA 1-ANTITRYPSIN DEFICIENCY REGISTRY SCREENING FORM

QUESTION #	ITEM	INSTRUCTIONS
15	Ascertainment (continued)	

(5)No pulmonary symptoms present but breathing tests done and subnormal, leading to testing. This response takes precedence over having a family member with the deficiency.

(6)Liver disease lead to testing for alpha 1-antitrypsin deficiency. This response is checked only if the patient has no pulmonary symptoms and has not had any abnormal lung function tests. This also takes precedence over having a family member with the deficiency.

(7)Unverified screening of blood (Coded at the CCC)

(8)Abnormal Chest x-ray (Coded at the CCC)

(9)Unknown. This is to be used only if it is impossible to determine what lead to the patient's being tested forthe deficiency.

If the patient has been entered into the Registry, skip to the bottom of the form. Fill in the name of the person completing the form and have the principal investigator for the Registry at your center review the form and sign the form in the space provided.

If the patient refused participation in the Registry for any reason, complete items 16-18.

Make sure the FEV_1 and height measurements are recorded in the units indicated on the form. An attempt should be made to get this information. If this information is unknown or unavailable, fill in each of the dashes with a "9".

ALPHA 1-ANTITRYPSIN DEFICIENCY REGISTRY SCREENING FORM

Form Completion Instructions:

Alpha 1-Antitrypsin Alleles Associated with an Increased Risk for Emphysema*

Alleles of Known Sequence

High Risk[1]	Mild Risk[2]
Z	S
$M_{procida}$	I
$M_{heerlen}$	
$M_{mineral\ springs}$	
M_{malton}	
P_1	
$Null_{granite\ falls}$	
$Null_{bellingham}$	
$Null_{mattawa}$	
$Null_{bethesda}$	
$Null_{procida}$	
$Null_{hong\ kong}$	
$Null_{bolton}$	

Alleles Defined by Gel Electrophoresis Only

High Risk[1]
M_{duarte}
M_{like}
M_{rouen}
$Z_{ausburg}$

*For a list of codings of Phenotypes, see Notes on coding for Form 01.

[1]High risk if homozygous inheritance or if heterozygous inheritance with another high risk allele.

[2]Mild risk if heterozygous inheritance with a high risk allele; no risk if homozygous inheritance.

ALPHA 1-ANTITRYPSIN DEFICIENCY REGISTRY
Screening Form

This form should be completed for all patients identified as being a potential participant for the Registry.

1. Date Form Completed: F01Q01_fzd (fuzzed) ___/___/___ (month/day/year)
2. Clinical Center code number: clinic (censored) ___
3. Birthdate: F01Q03_fzd (fuzzed) ___/___/___ (month/day/year)
4. Sex: F01Q04 ___(1)Male ___(2)Female
5. Race: F01Q05 (1)White ___(2)Black ___(3)Oriental ___(4)Other: _____ (specify)
   → (censored)
6. Has the patient ever received alpha 1-antitrypsin augmentation therapy? F01Q06 ___(1)Yes ___(2)No
   a. Date initial augmentation therapy started: F01Q06A_fzd (fuzzed) ___/___/___ (month/day/year)
   b. Type of initial treatment: F01Q06B (1)Infusion ___(2)Other ___(9)Unknown
      Comments: never entered
   c. Initial frequency: F01Q06C (1)Weekly ___(2)Monthly ___(3)Other ___(9)Unknown
      Comments: never entered
7. Has the patient agreed to participate in the Registry (Consent Form signed)? F01Q07 ___(1)Yes ___(2)No

   IF NO TO QUESTION 7 OR IF CENTER HAS DECIDED NOT TO PURSUE PATIENT, SKIP TO QUESTION #16.

8. Serum kit number: F01Q08 (censored) ____
9. Patient Registry ID: Newid (Scrambled) _____
10. Patient Name Code (First 2 letters of first and last name): namecode (censored)

    For Questions 11 and 12:
    Report serum level/phenotype from Central Laboratory.

11. Serum alpha 1-antitrypsin level (micromolar): F01Q11 __.__
    a. Date test confirmed: F01Q11A_fzd (fuzzed) ___/___/___ (month/day/year)
12. Phenotyping from Central Laboratory: F01Q12 ___

    (Question 12 was coded by the CCC. See notes on coding.)
    a. Date test confirmed: F01Q12A_fzd (fuzzed) ___/___/___ (month/day/year)

White/Yellow: Clinical Coordinating Center, Pink: Clinical Center

PWO 1861

Patient Registry ID: ___ ___ ___ ___ ___
Birthdate: ___ ___ / ___ ___ / ___ ___
month day year

13. Any blood relatives who also have alpha 1-antitrypsin deficiency: F01Q13 ___(1)Yes ___(2)No ___(9)Unknown

If YES, Specify: unreliable

a. Relationship: F01Q13A1
Registry Pt ID: F01Q13A2 ___ ___
b. Relationship: F01Q13B1
Registry Pt ID: F01Q13B2 ___ ___
c. Relationship: F01Q13C1
Registry Pt ID: F01Q13C2 ___ ___

Unreliable

d. Relationship: F01Q13D1
Registry Pt ID: F01Q13D2 ___ ___
e. Relationship: F01Q13E1
Registry Pt ID: F01Q13E2 ___ ___
f. Relationship: F01Q13F1
Registry Pt ID: F01Q13F2 ___ ___

14. How was patient referred to Clinical Center: F01Q14

___(1)Outside Doctor
___(2)Clinical Center Doctor
___(3)Advertisement
___(4)Lung Organization
___(5)Chart Review
___(6)Family Member
___(7)Other (specify): never entered
___(9)Unknown

15. What led to suspicion and diagnosis of alpha 1-antitrypsin deficiency in this patient: F01Q15

___(1)Screening of blood.
___(2)Pulmonary symptoms led to testing of patient.
___(3)Identified as a family member of a known alpha 1-antitrypsin deficient patient.
___(4)Other reason for suspecting deficiency (specify): never entered
___(5)No pulmonary symptoms present but breathing tests done and subnormal, leading to testing.
___(6)Liver disease led to testing for alpha 1-antitrypsin deficiency.
___(7)Unverified screening of blood (CCC use only)
___(8)Abnormal chest x-ray (CCC use only) ___(9)Unknown.

IF PATIENT REFUSED TO PARTICIPATE IN THE REGISTRY, ANSWER QUESTIONS 16-18:

16. Reason for non-participation: F01Q16 (never entered)

17. Date of most recent spirometry test: F01Q17 fzd (never entered) ___ ___ / ___ ___
month day year

a. FEV_1 (L) (BTPS) from most recent spirometry: F01Q17A (never entered)

18. Patient height (cm): F01Q18 (never entered) ___ ___ ___

Form Completed By (Name): never entered

Physician Signature: never entered

White/Yellow: Clinical Coordinating Center, Pink: Clinical Center

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Notes on Coding:

Question 12. Phenotype codes from the Central Laboratory

Lising of codes used:

(1) Z
(2) SNULL
(3) SZ
(4) M1M1
(5) M1Z
(6) M3Z
(7) IZ
(8) M2Mmalton
(9) M1S
(10) NULL+LOWLEVEL
(11) PlowellNULL
(12) M3M3
(13) NULLbellingham
(14) ZMheerlen
(15) NULLgranitefalls
(16) ZNULLgranitefalls
(17) M1NULL
(18) MheerlenNULLclayton
(19) MheerlenNULL
(20) SMheerlen
(21) M1NULLheerlen
(22) M1M3
(23) NZ
(24) ZMmalton
(25) PENDING
(26) M1M2
(27) M2S
(28) M2Z
(29) M2M2
(30) FZ or IZ
(31) GeneScreen Z
(32) GeneScreen Non-Z
(33) PlowellZ
(34) MheerlanNullbolton
(35) M1NULLbolton
(36) FZ
(37) M1Xchristchurch
(38) NULLclayton
(39) M1Nullwest
(40) M1I

Note: Codes 1, 3, 8, 11, 13-16, 18, 20, 24, 25, 31, 33, and 34 were represented among the 1129 eligible Registry subjects

Notes on Coding, continued

Additional variables coded by the CCC:

Variable	Coding
FAMID	Unique numeric code assigned by the CCC. Only assigned to registry subjects who are members of a family cluster. Registry subjects with the same FAMID value are related by blood.
PROBAND	Code to identify family relationships within family clusters, i.e., among those who share the same FAMID. Note: the family member selected to be the "proband" (code=1) is arbitrarily chosen and is not a proband in the sense of being an "index case". Relationships of other family members are coded as their relation to the proband. Codes used: 1 = proband (arbitrarily chosen member of the family cluster) 2 = sibling of proband 3 = parent of proband 4 = child of proband 5 = uncle/aunt of proband 6 = niece/nephew of proband 7 = cousin of proband 8 = grandparent of proband 9 = grandchild of proband 10= identical twin of proband 11 = fraternal twin of proband