



Research Priorities in Alpha-1 antitrypsin deficiency (AATD)

Which research topics might have the greatest impact on the lives of people with AATD?

Introduction

We are asking people with Alpha-1 Antitrypsin Deficiency (AATD), or their families and friends, to tell us what we should be looking at to provide answers to the challenges of care, treatment and living with AATD.

The priorities for research identified by people with AATD and their families may be different to the point of view of doctors and researchers. The aim of this survey is to find out which topics of research YOU think are most relevant or most likely to provide answers to the challenges of treating and living with AATD and will therefore have the greatest impact on quality of life for people with AATD.

This survey is part of the scope of work lead by EARCO (European Alpha-1 Research Collaboration), a Clinical Research Collaboration (CRC) of the European Respiratory Society (ERS), to facilitate multidisciplinary collaborative research in AATD (www.ersnet.org/research/earco-european-alpha-1-research-collaboration).

Your answers will influence what research is done by these research centres in the future. The questionnaire is mainly focused on the respiratory burden and lung diagnosis and treatment for AATD.

This survey will take up to 15 minutes to complete and is anonymous. If you would like to receive updates or would like to become more involved in the project, you can enter your email address at the end of the survey. If you would like to do this, you will be directed to a different surveymonkey page. This means that your email address and survey responses are separate, and anonymity is maintained.

Your participation will significantly contribute to the success of this project. This survey will close on the 8 December 2019. Thank you very much for your time and effort!

Dr. Marc Miravittles and Dr. Timm Greulich - CRC EARCO Chairs



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Instructions

This questionnaire is divided into parts:-

- Part 1 describes your demographics.
- Part 2 - in this part you will be asked which research areas you think should be prioritised:

- Symptoms and burden of the disease
- Areas of research
- Diagnosis and awareness of the disease
- Treatment burden
- Self-management and education

You are asked to rate each research priority question by order of importance (Unimportant, Not very important, Important, Very important, No Opinion) and rate the challenge you face to manage each aspect of your disease (Not an issue, Not very difficult, Difficult, Very difficult).

Although every item might be very important, we advise you to answer as objectively as possible (pointing out your priorities) so that we can generate new knowledge with direct impact on your quality of life and clinical care.

Finally, you will be asked if there are any additional research priorities that you consider necessary to include. If so, please propose them in the box provided at the end of each area of research.

This survey can be answered by patients, parents, relatives or caregivers of someone with AATD. If you are answering this survey on behalf of someone else, please provide information concerning the patient opinion.

Thank you very much for your time and effort!

Yours sincerely,

Dr. Marc Miravittles and Dr. Timm Greulich - CRC EARCO Chairs

Please do not hesitate to contact jeanette.boyd@europeanlung.org if you have any questions related to the survey.



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About you

If you are completing this on behalf of someone with AATD, please add their details in response to the demographic questions

* 1. Are you....?

- A person diagnosed with alpha-1 antitrypsin deficiency (AATD)?
- A parent, relative or caregiver of someone with AATD? (please, specify)
- Other? (please, specify)
- Please specify here if you are not a person diagnosed with AATD

2. What age are you (in years)? (if you are answering this survey on behalf of someone else, please provide the age of the person with AATD)

* 3. Are you..? (if you are answering this survey on behalf of someone else, please provide the gender of the person with AATD)

- Male
- Female
- Prefer not to say

* 4. In which country were you born?

* 5. In which country do you currently live?



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About your AATD

* 6. For how many years have you been diagnosed with AATD?

* 7. What was the reason for your diagnosis?

- COPD
- Asthma
- Liver disease
- Panniculitis
- Family testing
- Other (please specify)

* 8. Are you a ...

- Current smoker
- Former smoker
- Never smoker

9. Do you drink alcohol?

- No
- Yes

If yes, how many litres per week, on average?

* 10. In your professional activities were/are you exposed to gases, fumes or dust?

- Yes
- No

* 11. Who diagnosed your AATD?

- Family physician / General Practitioner (GP)
- Respiratory specialist
- Gastroenterologist / Hepatologist
- Pediatrician
- Other (please specify)

* 12. What is your AATD phenotype / genotype?

- ZZ
- SZ
- MZ
- MS
- Don't know
- Other (please specify)

* 13. Have you had a lung transplant?

- Yes
- No

* 14. Have you had a liver transplant?

- Yes
- No



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Part 2: Your experience of AATD

Symptoms and burden of the disease

15. What aspects of the disease do you find most challenging and/or difficult to manage (during the past 12 months)?

	Very difficult	Difficult	Not very difficult	Not an issue	N/A
Shortness of breath	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Not feeling fit or having the strength to do daily activities	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Decreased exercise tolerance	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Tiredness	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Cough	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Sputum (mucus from the lungs)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Wheezing	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Sleeping problems	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Anxiety	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Depression	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Difficulty to maintain social life	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Exacerbations (episodes of increased or change in colour of sputum, shortness of breath and/or fever that lead you to go to the doctor)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Frequent hospital admissions	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

	Very difficult	Difficult	Not very difficult	Not an issue	N/A
Having regular tests or explorations performed (e.g. lung function testing, computed tomography scan, chest x-ray, liver examinations)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Jaundice (yellowing of your skin and eyes)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Cholestasis	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Abnormal liver function tests	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Vomiting	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Swelling or pain in your belly/swollen abdomen	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Poor growth/weight loss	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Poor appetite	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Diarrhea	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Itching	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Fatigue	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Panniculitis	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Other: please give ideas not already included here



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Part 2: Your experience of AATD

Treatment Burden

16. What aspects and/or barriers for treatment do you find most challenging and difficult to manage?

	Very difficult	Difficult	Not very difficult	Not an issue	N/A
Access issues to augmentation therapy in your healthcare system	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Hospital administration of augmentation therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Time consumed in augmentation therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Access to pulmonary rehabilitation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Access to maintenance classes or using fitness centers to maintain fitness after rehabilitation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Use of inhaled and/or nebulized therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Use of oxygen therapy and/or noninvasive ventilation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Access to psychological support	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Access to support for family planning and family screening	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Professional implications (ie, loss of job due to disease or therapy)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Additional costs/expenses due to disease or therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Limitation of daily life due to therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Impact of transplant in patients and their families	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Other: please give ideas not already included here



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Part 2: Research Prioritisation

Areas of Research

17. The priorities for research identified by people with AATD and their families may be different from the point of view of doctors and researchers. It is important we understand what patients' research priorities are.

How important do you think the following research areas are to improve AATD management?

	Very important	Important	Not very important	Unimportant	No opinion
Development of an international AATD registry	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
More evidence on effectiveness of augmentation therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Alternatives to IV augmentation therapy (e.g. inhaled augmentation therapy)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Evidence on the effect of augmentation therapy on reducing exacerbations	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Synthetic AAT production to cover the demand	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Gene therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Innovative liver therapies	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
AATD in heterozygotes (e.g. MZ phenotype): clinical manifestations, indications for augmentation treatment	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Very important Important Not very important Unimportant No opinion

Learning more about the natural development of the disease (including liver disease or other clinical conditions related to AATD)

Different evolution of the disease among patients

Evolution of patients after lung and/or liver transplant

Relationship between lung and liver disease

Relationship between AATD and other diseases (e.g. asthma, bronchiectasis, vasculitis)

Investigating the effect of pollution, work exposures, second hand smoking, etc, in the development of lung disease

Other: please give ideas not already included here



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Part 2: Research Prioritisation

Diagnosis and awareness of the disease

18. How important do you think it is to improve the following areas for diagnosis and awareness of AATD?

	Very important	Important	Not very important	Unimportant	No opinion
Screening programs in newborns (neonatal screening)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Screening programs in the general population	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Targeted screening programs: COPD and asthma patients	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Early diagnosis of the disease using innovative online test procedures	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Diagnosis in liver disease patients (children and adults)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Improving knowledge of AATD, in particular among General Practitioners	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Education for physicians on diagnostic techniques, algorithm and interpretation of the results	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Diagnosis of non-respiratory diseases associated with AATD (e.g. panniculitis, vasculitis, liver disease)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Very important

Important

Not very important

Unimportant

No opinion

Educational programs regarding regional/national resources to diagnose and refer AATD patients

Implications of being diagnosed with a genetic disease (ethical and economic) and its potential negative impact

Other: please give ideas not already included here



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Part 2: Research Prioritisation

Treatment

19. How important do you think it is to improve the following areas for AATD treatment?

	Very important	Important	Not very important	Unimportant	No opinion
Dose regimen of augmentation therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Augmentation therapy after lung transplantation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Augmentation therapy for panniculitis	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Difficulties in accessing augmentation therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Administration of augmentation therapy at home	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Use of augmentation therapy during holidays/prolonged travel	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Side effects of augmentation therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Availability of organ (lung and/or liver) donation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Development of multidisciplinary centers	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Develop other aspects of integral care (eg physical activity, care-giver support, maintaining work or schooling, nutrition, psychological care, sex-life, daily- life)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Smoking cessation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

	Very important	Important	Not very important	Unimportant	No opinion
Alcohol cessation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Role of nutrition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Role of pulmonary rehabilitation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Role of oxygen therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Role of noninvasive ventilation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Use of vaccines to prevent exacerbations	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Liver disease therapy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Other: please give ideas not already included here



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Which research topics might have the greatest impact on the lives of people with AATD?

Part 2: Research Prioritisation

Self-management and education

20. Education, technology and self-awareness, known as self-management, can help each person gain greater control over their disease and to improve daily quality of life.

How important do you think the following areas are in improving self-management of AATD?

	Very important	Important	Not very important	Unimportant	No opinion
Being able to recognize an exacerbation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Having an action plan for exacerbations and easy access to healthcare during episodes	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
An app to facilitate disease management and treatment (e.g. activity diary)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Regular communication between healthcare professional team and each individual with AATD	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Interaction and information exchange within a patient organization or self-help group	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Using peer support forums and social media to exchange information with others	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Having access to reliable, easy to understand information about different aspects of living with AATD	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Very important Important Not very important Unimportant No opinion

Develop better ways of teaching people to use their medicines (e.g. inhalers, oxygen)

A personalized integrated care plan including therapeutic physical activity

Having access to pulmonary rehabilitation and being taught the techniques and how to use the equipment at home

Having access to AATD specialized centres

Other: please give ideas not already included here



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Thank you for completing the AATD patient survey.
Your contribution is greatly appreciated.

If you would like to receive the results of the survey and updates on the project please follow this link to enter your email address: <https://www.surveymonkey.co.uk/r/EARCO-ATTD>

Please note that the European Lung Foundation will only contact you in relation to this survey and to send you updates about this project. We will not share your email address with any third parties. Your email address will be stored on our secure servers and we will retain your email only for as long as is necessary to provide you with the service stated above.

**Please contact Jeanette Boyd at the European Lung Foundation if you have any queries:
jeanette.boyd@europeanlung.org**