

# Alpha-1 antitrypsin deficiency (AATD) Patient survey

EARCO CRC

Final report

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## **Introduction**

This survey is part of the scope of work led by EARCO (European Alpha-1 Research Collaboration), a Clinical Research Collaboration (CRC) of the European Respiratory Society. EARCO aims to facilitate multi-disciplinary collaborative research in Alpha-1 antitrypsin deficiency (AATD).

The survey asked people with AATD, and their family members and caregivers, to tell us what we should be looking at to provide answers to the challenges of care, treatment and living with AATD. The survey questions were developed by EARCO members including AATD patient representatives and the European Lung Foundation (ELF) and the survey was provided in 9 languages: English, Dutch, French, German, Italian, Polish, Portuguese, Serbian and Spanish. The survey was online for four weeks during November/December 2019 and promoted through the networks/social media of Alpha-1 Global, Alpha-1 patient organisations/groups, ELF, ERS and members of the CRC.

This report has been compiled by ELF staff based on the survey findings.

## **Short summary of survey results**

More than half of respondents were diagnosed by a respiratory specialist (56%) with the most likely cause for diagnosis being COPD (32%) followed by family testing (17%). Many were diagnosed following recurrent chest infections and pneumonia while others were diagnosed while being tested for something else. A majority of respondents were former smokers (62%).

To improve diagnosis and awareness of AATD, respondents rated: improving knowledge among General Practitioners; targeted screening programs in COPD/Asthma patients; and education for physicians as the most important areas.

The most challenging management aspects stated were decreased exercise tolerance; shortness of breath and not feeling fit or having the strength to do daily activities.

The most challenging treatment aspects were identified as: access issues to augmentation therapy; professional implications (i.e. loss of job) and access to classes to maintain fitness after rehabilitation. The most important areas to improve treatment were suggested as smoking cessation, developing other aspects of integral care and pulmonary rehabilitation.

All but two of the research areas were rated by 80% or more of respondents as 'Important' or 'Very important' with the top two most important identified as: research into the relationship between AATD and other diseases; and more evidence on the effectiveness of augmentation therapy.

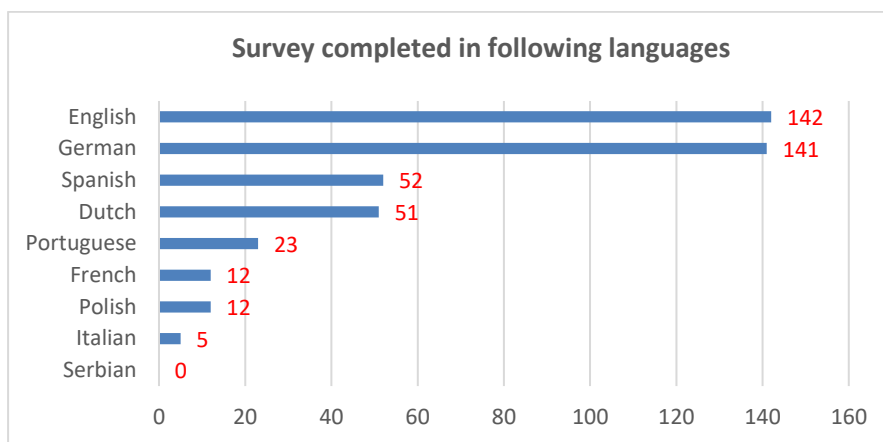
The most important areas to improve self-management and education were highlighted as: access to AATD specialised centres; access to reliable, easy-to-understand information about living with AATD and being able to recognise an exacerbation.

## About the respondents

438 respondents out of a total of 440 were included for analysis as they met the criteria of being either a person with Alpha-1 antitrypsin deficiency (AATD) or a parent, relative or care-giver of someone with AATD. The 2 excluded respondents were medical professionals.

Respondent characteristics:

- 84% a person diagnosed with AATD and 16% a parent, relative or caregiver.
- 58% female, 41% male, 1% did not state.
- The mean age of respondents was 50 years.
- See chart below for number of survey respondents by survey language:

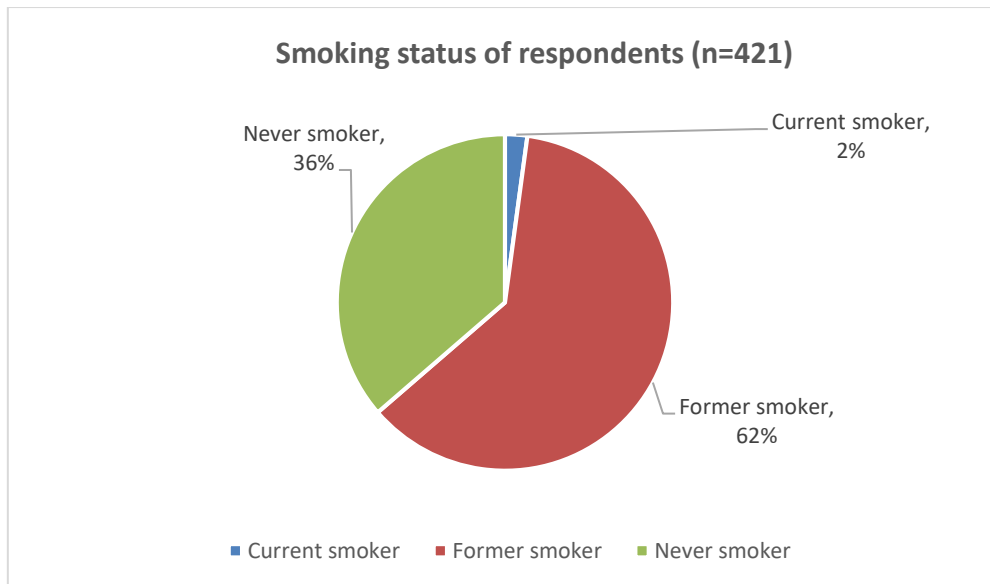


Respondents across all surveys were located in 26 countries/regions: Argentina, Australia, Austria, Belgium, Cyprus, Denmark, Ecuador, Finland, France, Germany, Ireland, Italy, Liechtenstein, Mexico, Netherlands, Norway, Poland, Portugal, Slovenia, Spain, South Africa, Sweden, Switzerland, United Kingdom, United States of America, Zimbabwe.

## Additional characteristics

### Smoking and alcohol use

- 55% of respondents drink alcohol.
- 62% of respondents were a former smoker, see chart below for full details:



### Environmental exposure

Respondents (n=421) were asked if they were/are exposed to gases, fumes or dust in their professional activities and 71% said No (29% said Yes).

### Transplants

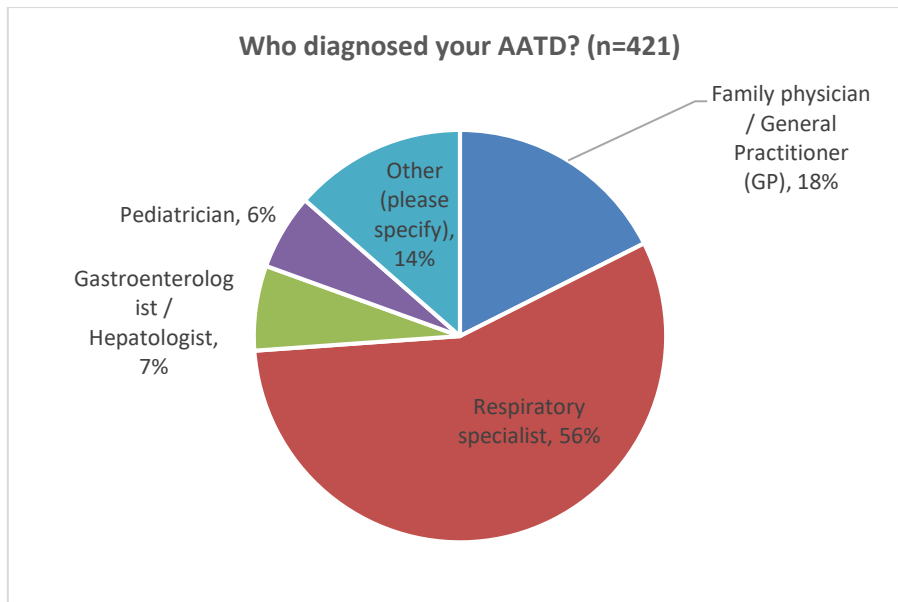
Respondents were asked if they had had a lung and/or liver transplant:

- 4% of respondents have had a lung transplant (n=421)
- 1% of respondents have had a liver transplant (n=421)

### Diagnosis

#### Who diagnosed?

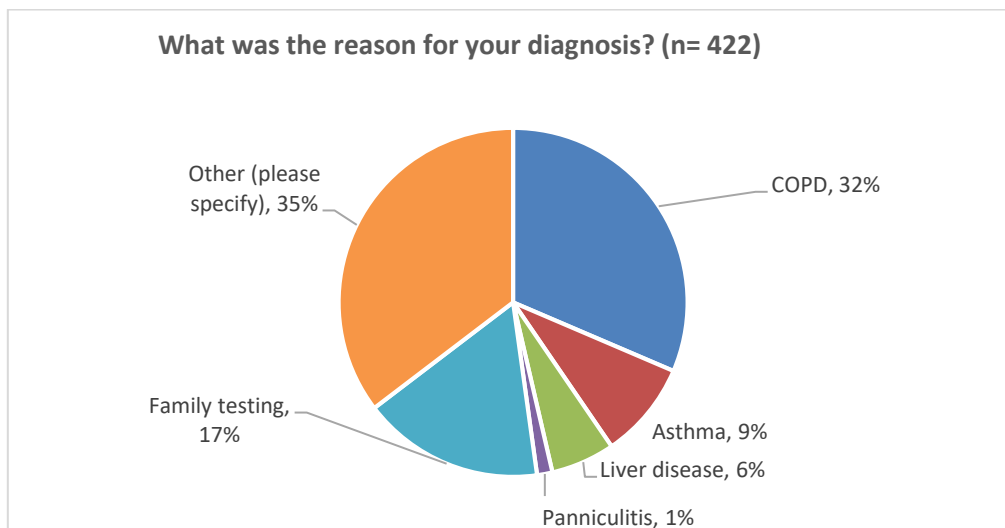
Respondents were asked which healthcare professional diagnosed AATD with the majority stated a Respiratory specialist (56%) and Family doctor (18%). See chart on next page:



Answers in the 'Other' field included: Geneticist, Immunologist, Allergist, Dermatologist, Rheumatologist, Naturopath, Endocrine, Research laboratory, Hospital, Clinic, Myself.

### Reason for diagnosis

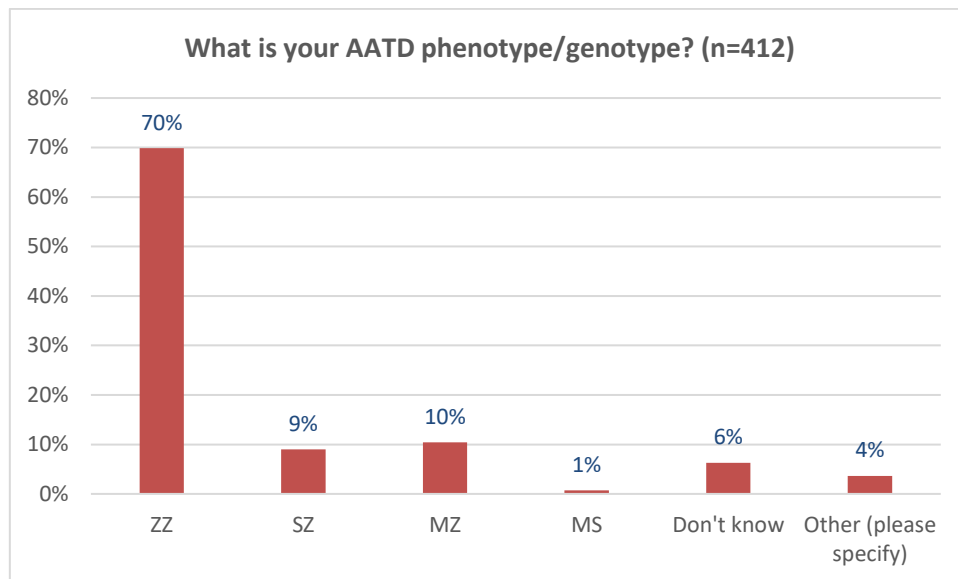
Respondents were also asked about the reason for their diagnosis and 32% stated COPD, 17% Family testing and 9% Asthma – see full results in chart below:



Other reasons given included: Breathing issues (n=14), Pneumonia (n=14), Chest infections (n=12), Liver issues (n=8), Accidental diagnosis (n=5), Weight loss (n=3), Emphysema (n=2), Swine flu (n=2), Cough (n=2) plus individuals who mentioned Fibromyalgia, Bronchiectasis, Bronchitis (n=3), Migraine, Tiredness, Low blood pressure, Inexplicable bleeds as a baby.

## Phenotype

Respondents were asked about their AATD phenotype and 70% have ZZ phenotype; 10% MZ and 9% SZ. See chart below:



## How long since diagnosis?

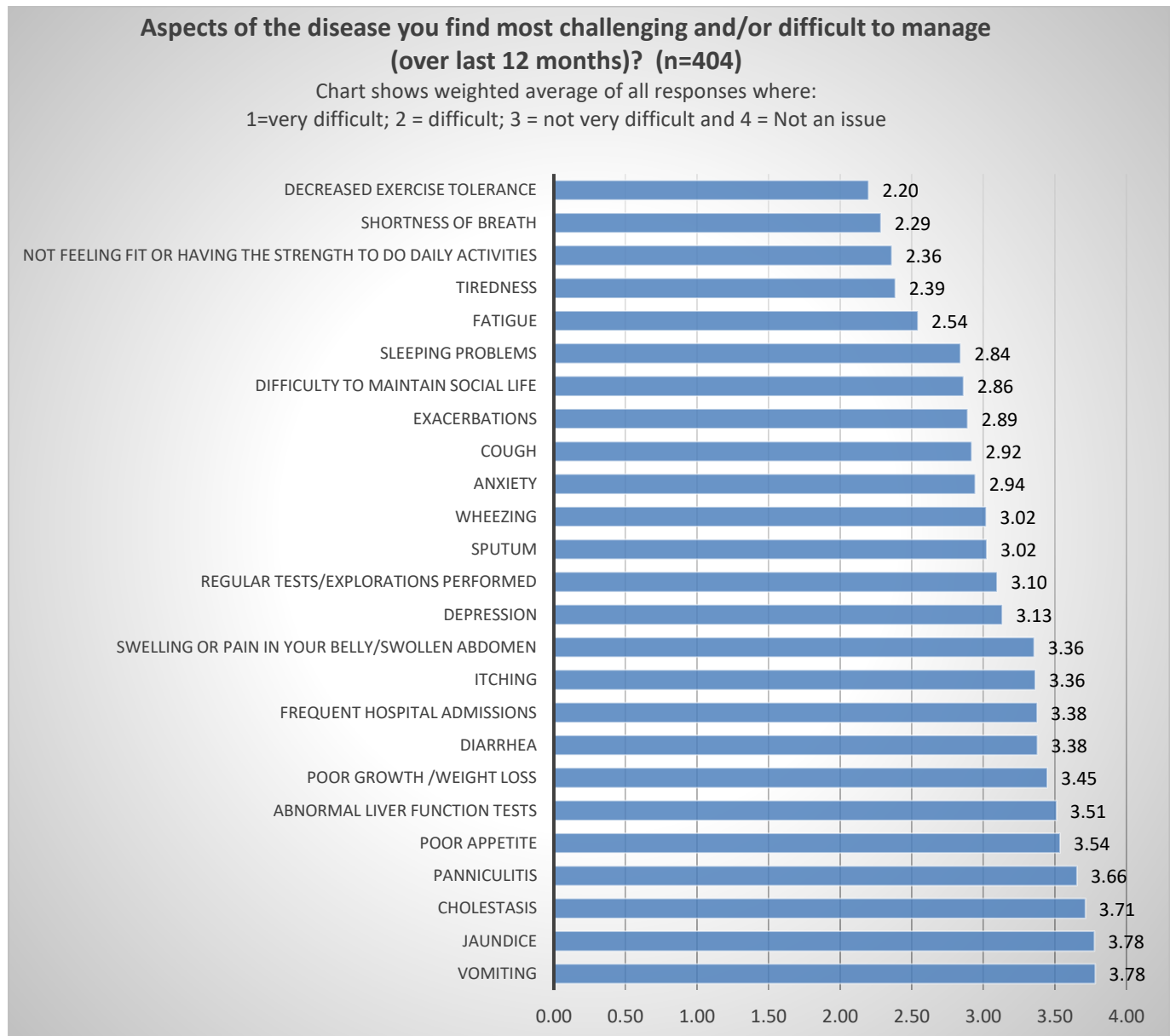
Respondents were asked how many years they had been diagnosed with AATD. The length ranged from 'less than 1 year' to 65 years with the average length since diagnosis being 12 years (median length was 9 years).

## Your experience of AATD

### Most challenging aspects to manage

Respondents were asked to rate which aspects of the disease they found most challenging and/or difficult to manage during the past 12 months. The Top 3 most challenging aspects:

1. Decreased exercise tolerance.
2. Shortness of breath.
3. Not feeling fit or having the strength to do daily activities.



*“Overwhelming difficulty in losing all aspects of normal life. No motivation as no hope of recovery”*  
(Respondent, EN survey).

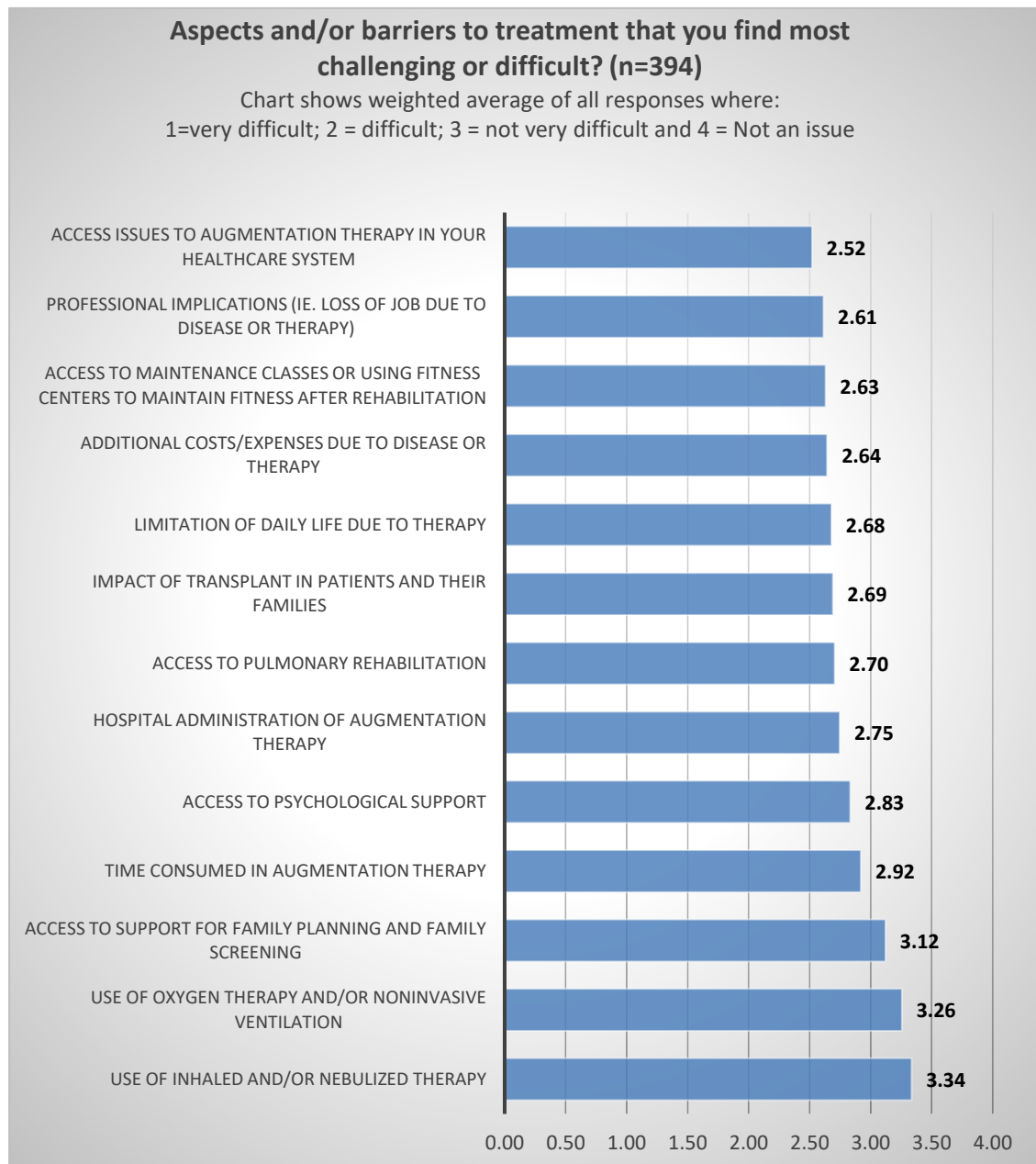
*“The fact that protein replacement therapy is being applied to my home by specialised laboratory personnel seems very important to my quality of life. That allows me to continue with my work and rehabilitation activities.”* (Respondent, ES survey (translated)).



## Most challenging aspects/barriers for treatment

Respondents were asked to rate which aspects and/or barriers for treatment do you find most challenging and difficult to manage. The Top 3 most challenging aspects were:

1. Access issues to augmentation therapy in your healthcare system.
2. Professional implications (i.e., loss of job due to disease or therapy).
3. Access to maintenance classes or using fitness centres to maintain fitness after rehabilitation



*"I struggle with oxygen therapy as I have anxiety when I wear it as people stare and comment. This makes me reclusive."* (Respondent, EN survey).

*"Too many visits in hospitals"* (Respondent, FR survey, translated)

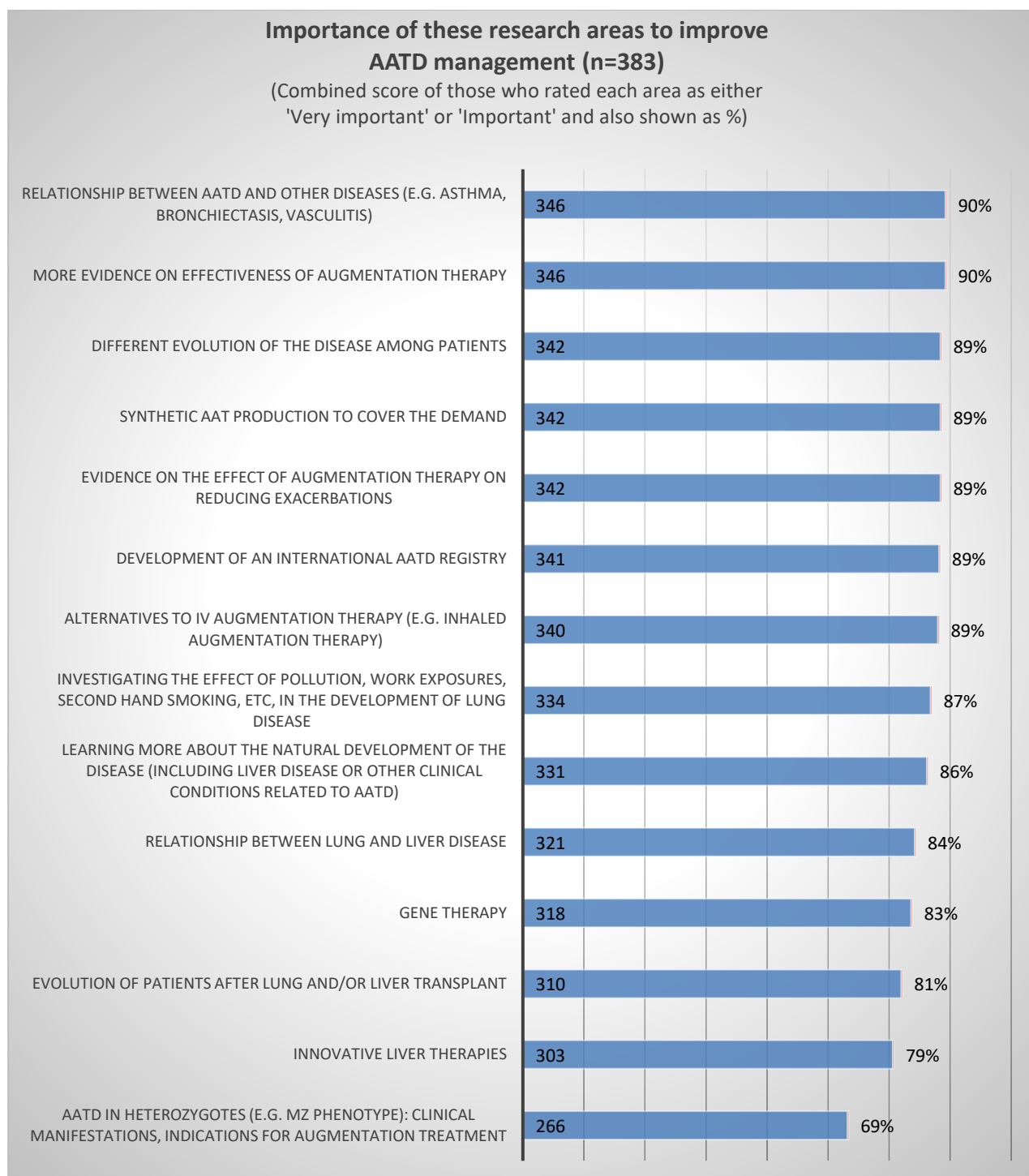
*"The fact that protein replacement therapy is being applied to my home by specialised laboratory personnel seems very important to my quality of life. That allows me to continue with my work and rehabilitation activities."* (Respondent, ES survey (translated)).

## Research prioritisation

### Improving AATD management

Respondents were asked to rate how important a list of research areas were to improve AATD management. The top most important research areas being:

1. Relationship between AATD and other diseases (90%)
2. More evidence on effectiveness of augmentation therapy (90%)
3. 5 areas of research came in at 89% - see chart below for full details:



*“The development of a drug or a method to stop lung deterioration or to cure pulmonary emphysema”* (Respondent, DE survey, translated)

*“Examination of why children with MZ are more likely to have asthma, bronchitis or pneumonia”* (Respondent, DE survey, translation)

*“Very important to start investigating heterozygotes that have very low blood AAT values and to consider them as risk patients”* (Respondent, PT survey, translation)

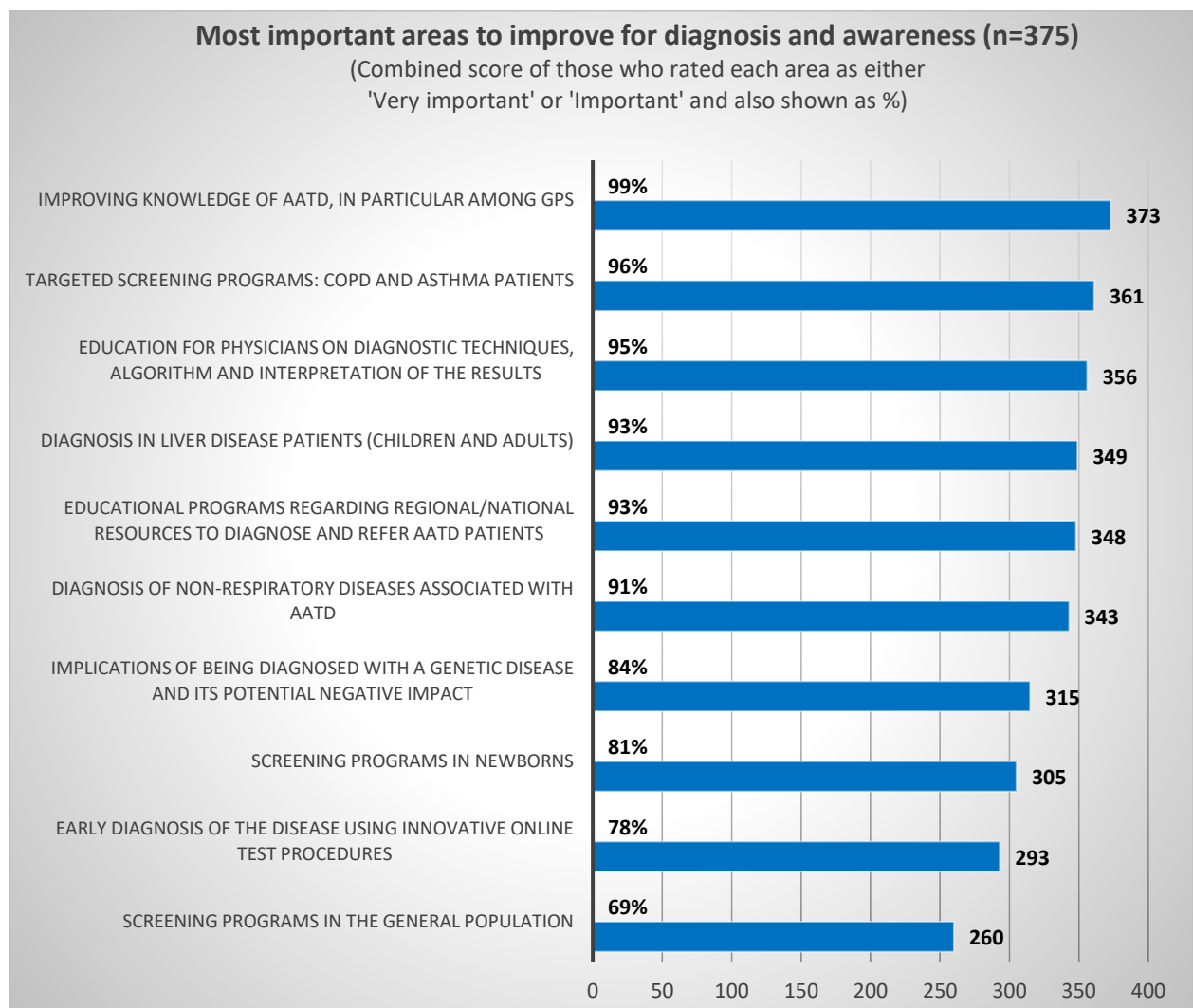
*“Diagnosing AAT in early childhood to avoid risk factors”* (Respondent, PL survey, translation)

*“In particular, investigate the reasons why one patient leads a reasonable life and another is continuously ill”* (Respondent, NL survey, translation)

### Improving diagnosis and awareness of AATD

Respondents were asked to rate a list of areas as to how important each is to improve diagnosis and awareness of AATD. All but 2 areas were rated as 80% or higher with the top 3 most important being:

1. Improving knowledge of AATD, in particular among General Practitioners (99%)
2. Targeted screening programs: COPD and Asthma patients (96%)
3. Education for physicians on diagnostic techniques, algorithm, interpretation of results (95%)



*“It is very important that general practitioners are educated because most of them know nothing about it” (Respondent, EN survey)*

*“Family members should be tested if a case of AATD has been detected” (Respondent, DE survey, translation)*

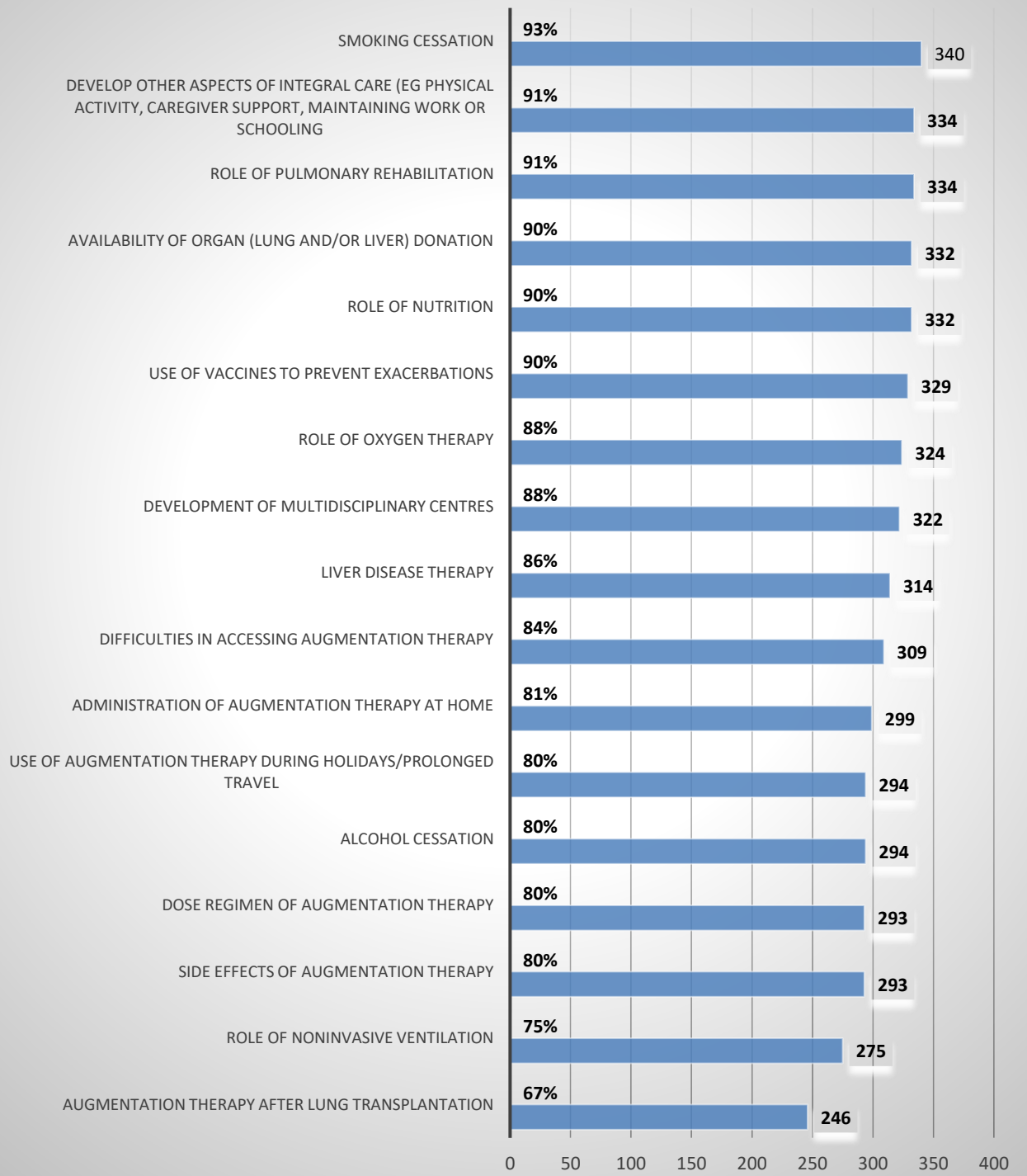
### **Improving treatment**

Respondents were asked to rate a list of areas as to how important each is to improve treatment of AATD. The results in the chart below show the combined scores of those who rated an area as ‘Very important’ and ‘Important’. The top 3 were:

1. Smoking cessation (93%)
2. Develop other aspects of integral care (e.g. physical activity, care-giver support, maintaining work or schooling, nutrition, psychological care, sex-life, daily life) - 91%
3. Role of Pulmonary rehabilitation (91%)

### How important are these areas for AATD treatment? (n=367)

Combined score of those who rated each area as either 'Very important' or 'Important' and also shown as %)

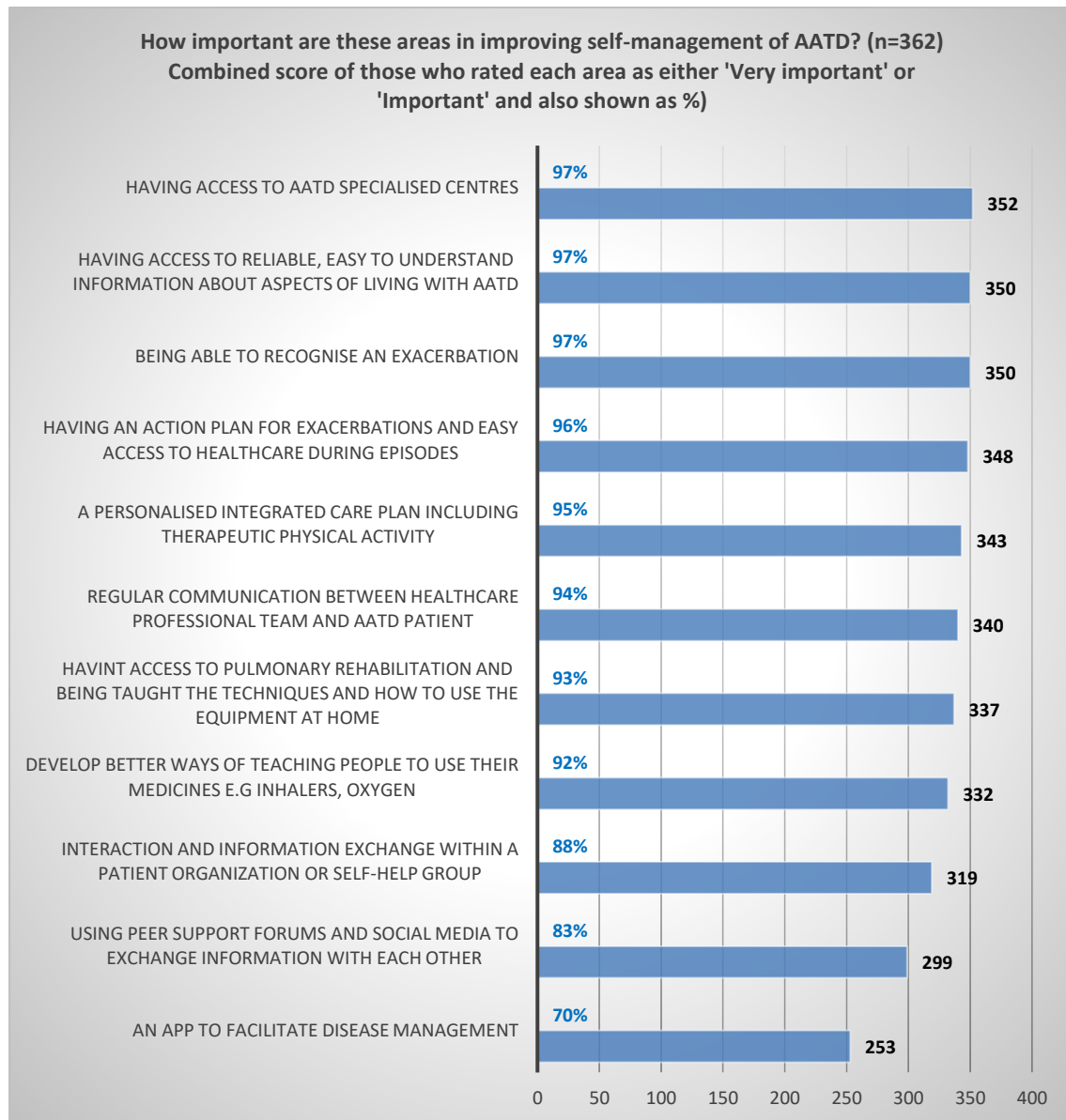


*“Being very susceptible to bacteria and viruses is therefore often lonely. Because, especially in the winter months, you are careful not to get sick among people.” (Respondent, NL Survey, translated)*

## Self-management and education

Respondents were asked to rate a list of areas as to how important each is to improve self-management of AATD. The top 3 most important being:

1. Access to AATD specialised centres (97%)
2. Access to reliable, easy to understand information about living with AATD (97%)
3. Being able to recognise an exacerbation (97%)



*"I have experienced very long wait times for pulmonary rehabilitation. Approximately 6 months for a 6-week course with no follow up. I found this to be typical through contact from social media"* (Respondent, EN survey)

*"Peer support meetings are very useful you can learn from each other. But keep it in the spring e.g. In May, June. Many of these are meetings in Sept, Oct. Then there is usually flu and then many people do not participate in order not to get sick."* (Respondent, NL survey, translated)

## General comments from survey respondents

*“Research is important but early diagnosis among new-borns is the most important thing that needs to be done” (Respondent, EN survey).*

*“DCLCO (Diffusing capacity of the lungs for carbon monoxide) not given the attention it deserves regarding treatment guidelines i.e. alphas with good FEV1 but bad DLCO can miss out on augmentation therapy and die prematurely” (Respondent, EN survey).*

*“Remote monitoring via iPhone as is now done in the US for chronic conditions would be useful, including to avoid breathing anxiety fits, unnecessary hospitalisations etc.” (Respondent, EN survey).*

*“The change of climate, and the cold make breathing difficult, living more than 2000 metres above sea level makes breathing difficult” (Respondent, ES survey, translation).*

*“Access to lung sports group in my region an impossibility (eternal waiting) or very miserable quality” (Respondent, DE survey, translation).*

*“No reference centre where we are with doctors of various specialities” (Respondent, PT survey, translation).*

*“Help patients without treatment. In countries that do not know about Alpha 1, we remain weakened and without hope of having the protein.” (Respondent, ES survey, translation).*

*“When waiting for a lung transplant, have more information regarding the progression in the list with passing time. Very scary to stay without news. Confusion between list position and donor compatibility (lung size, blood type etc)” (Respondent, FR survey, translation).*

*“A case manager who can co-ordinate and organise things” (Respondent, NL survey, translation)*

# Appendices

## Appendix 1: Data from the German language survey

A total of **141 respondents** to the German-language survey:

### Characteristics:

- 87% (n=123) a person diagnosed with AATD and 13% (n=18) a parent, relative or caregiver.
- 57% female, 43% male.
- The mean age of respondents was 51 years.
- Respondents who completed the survey were located in the following countries: Germany (n=113), Switzerland (n=16), Austria (n=6), Denmark (n=1), France (n=1), Liechtenstein (n=1), Netherlands (n=1), Norway (n=1), Sweden (n=1).

### Smoking and alcohol:

- 58% of respondents drink alcohol.
- 60% of respondents were a former smoker; 39% never-smoker and 1% current smoker.

### Environmental exposure

Respondents (n=136) were asked if they were/are exposed to gases, fumes or dust in their professional activities and 70% said No (30% said Yes).

### Transplants

Respondents (n=136) were asked if they had had a lung and/or liver transplant:

- 4% of respondents have had a lung transplant (n=6)
- 1% of respondents have had a liver transplant (n=1)

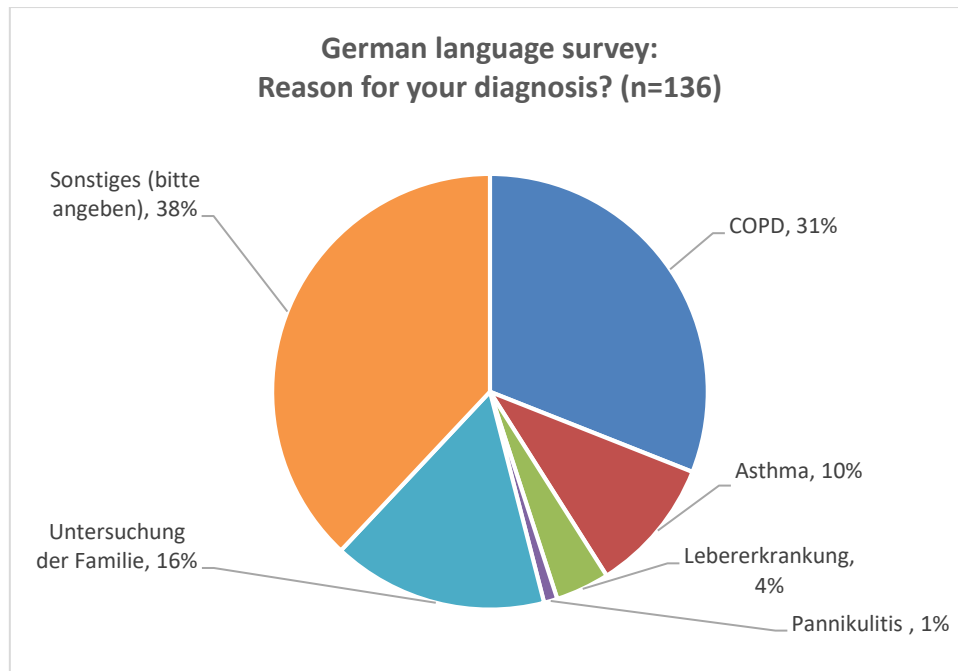
### Diagnosis

Respondents (n=136) were asked which healthcare professional diagnosed AATD:

- Respiratory specialist (e.g. pulmonologist) (60%)
- Family doctor (15%)
- Gastroenterologist/hepatologist (5%)
- Paediatrician (5%)
- Other (15%) – including Rehabilitation Doctor; Hospital; Molecular Biology Institute; Research Company; Geneticist; Naturopath.

Respondents (n=136) were also asked about the reason for their diagnosis and 31% stated COPD, 17% Family testing and 10% Asthma – see full results in chart:





**Other reasons given included:** Infections/pneumonia (n=9); shortness of breath (n=10); heavy weight loss (n=2); cough (n=1); bronchiectasis (n=1); bronchitis (n=3); diagnosis of a family member (n=5); inefficiency (n=1); allergies (n=1); body weakness (n=2); slight nosebleeds in infants (n=1); vitamin K deficiency (n=1); unclear digestive problems (n=1).

**AATD phenotype:** 75% have ZZ phenotype; 14% MZ; 6% SZ and 1% MS; 1% did not know and the rest did not state.

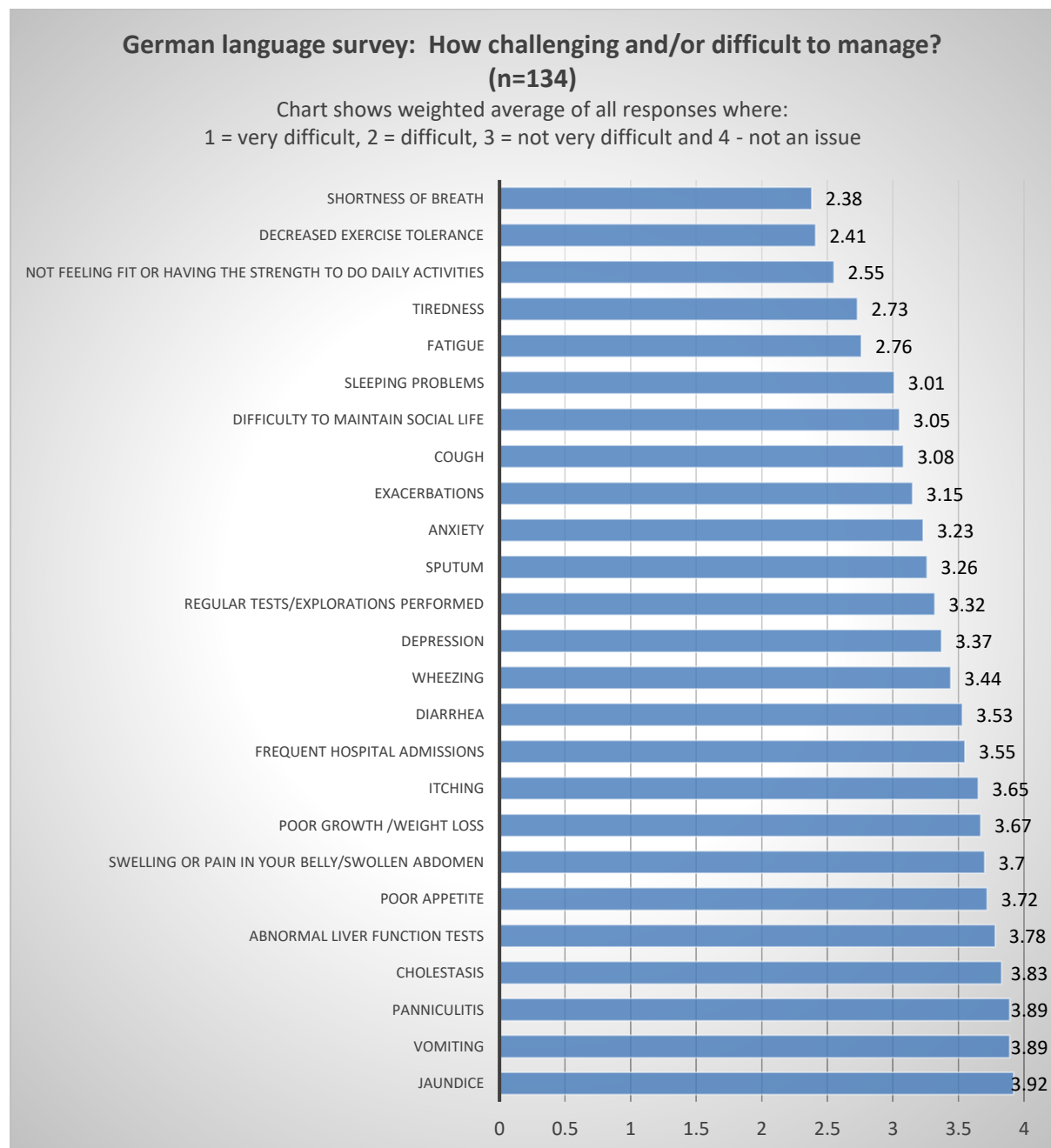
**How many years had they been diagnosed with AATD:** this ranged from '1 year' to 59 years with the mean length since diagnosis being 12 years.

## Your experience of AATD

### Most challenging aspects of AATD

Respondents were asked to rate which aspects of the disease they found most challenging and/or difficult to manage during the past 12 months. The Top 3 most challenging aspects were:

1. Shortness of breath.
2. Decreased exercise tolerance.
3. Not feeling fit or having the strength to do daily activities.



*“Angioödeme”* – Angioedema

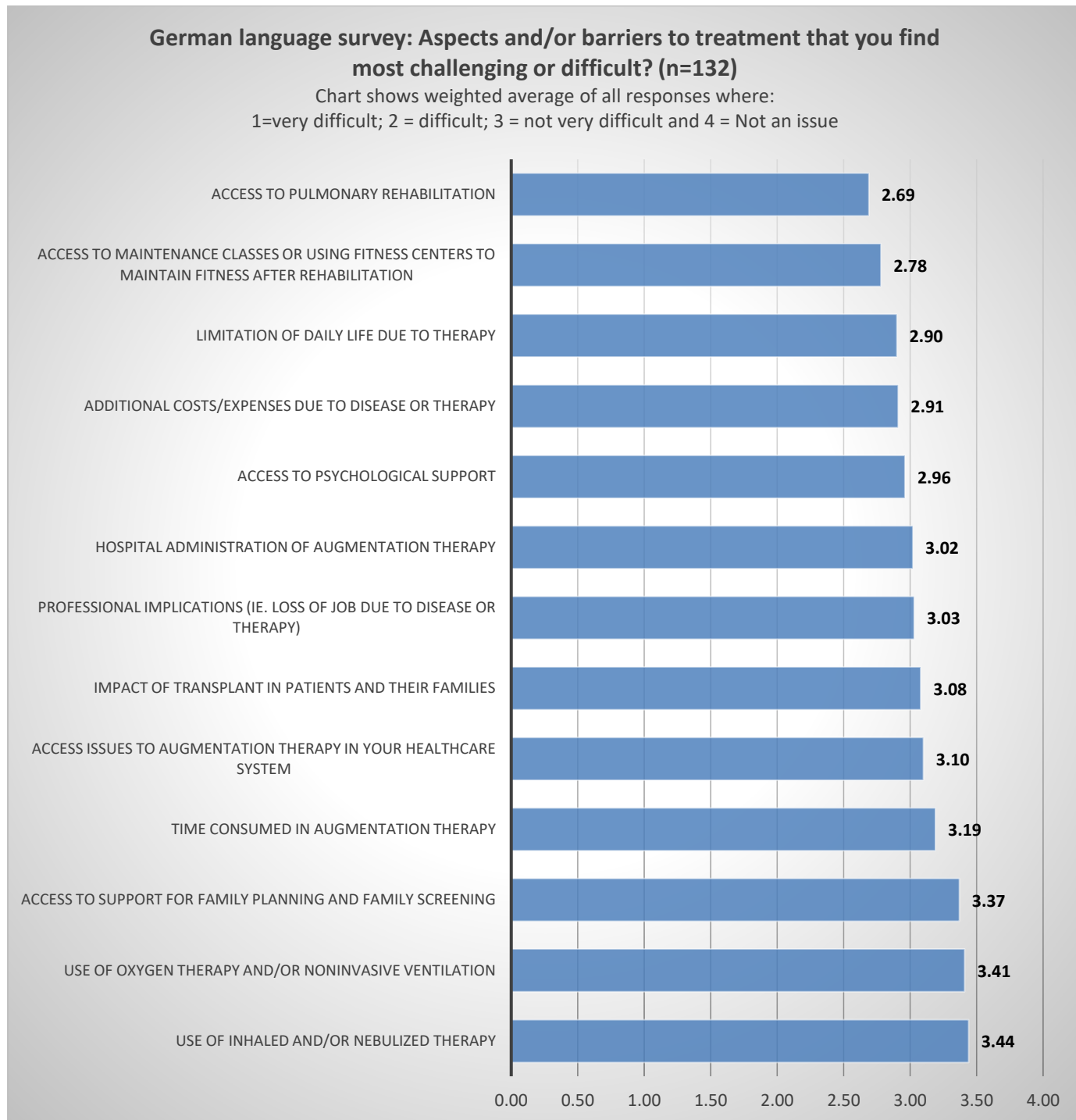
*“Verstärkte Warzenbildung”* – Increased wart formation

*“Schmerzen im Brustkorb”* - Chest pain

## Most challenging aspects/barriers for treatment

Respondents were asked to rate which aspects and/or barriers for treatment do you find most challenging and difficult to manage. The Top 3 most challenging aspects were:

1. Access to pulmonary rehabilitation.
2. Access to maintenance classes or using fitness centres to maintain fitness after rehabilitation
3. Limitation of daily life due to therapy.

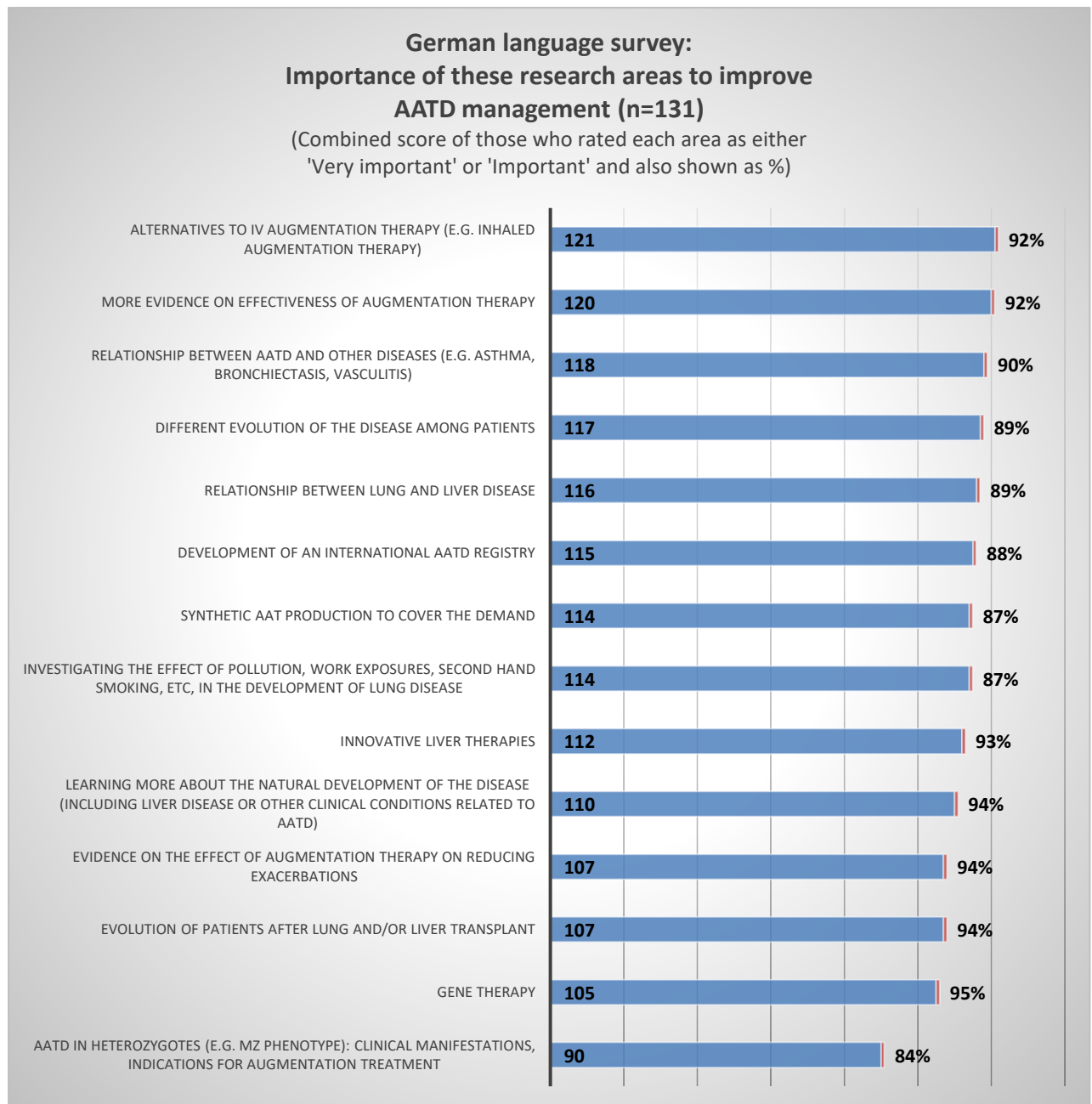


## Areas of Research

### Improving AATD management

Respondents were asked to rate how important a list of research areas was to improve AATD management. The results below show the combined scores of those who rated an area as ‘Very important’ and ‘Important’. The top most important research areas being:

1. Alternatives to IV Augmentation Therapy (92%)
2. More evidence on effectiveness of augmentation therapy (92%)
3. Relationship between AATD and other diseases (90%)

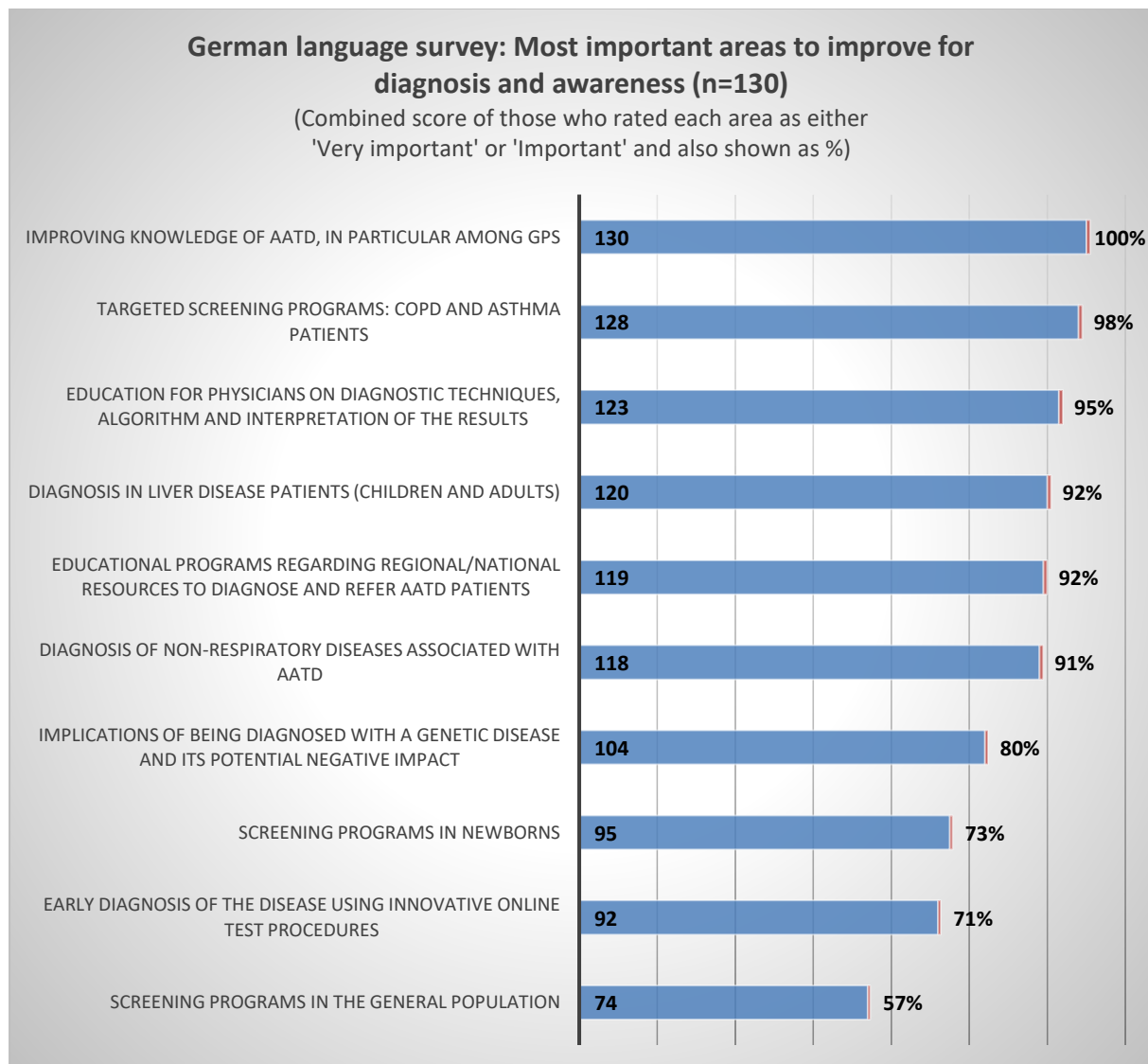


*“Mehr Austausch unter Aphas” – More exchange among alphas*

## Improving diagnosis and awareness of AATD

Respondents were asked to rate a list of areas as to how important each is to improve diagnosis and awareness of AATD. The top 3 rated as most important being:

1. Improving knowledge of AATD, in particular among General Practitioners (100%)
2. Targeted screening programs: COPD and Asthma patients (98%)
3. Education for physicians on diagnostic techniques, algorithm, interpretation of results (95%)



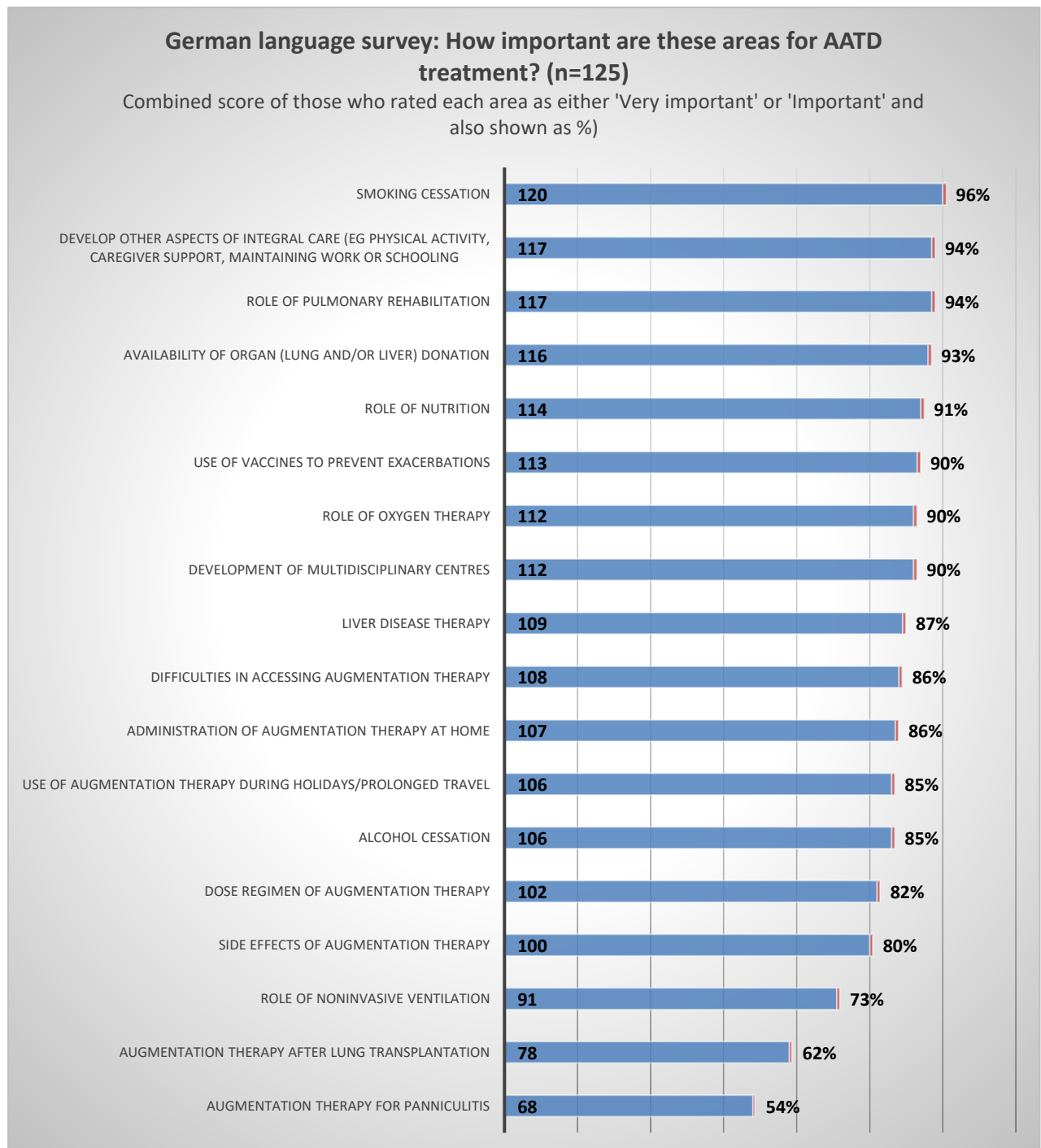
*“Allgemeine Informationen zur Krankheit. Musste meiner Behindertstelle aufklären um was es sich für eine Krankheit es ist”* - General information about the disease.

*“Das jeder Lungenarzt bei Patienten mit Emphysem einen Alphatest automatisch macht”* - Every pulmonologist in patients with emphysema automatically does an alpha test

## Improving treatment

Respondents were asked to rate a list of areas as to how important each is to improve treatment of AATD. The results in the chart below show the combined scores of those who rated an area as 'Very important' and 'Important'. The top 3 rated areas were:

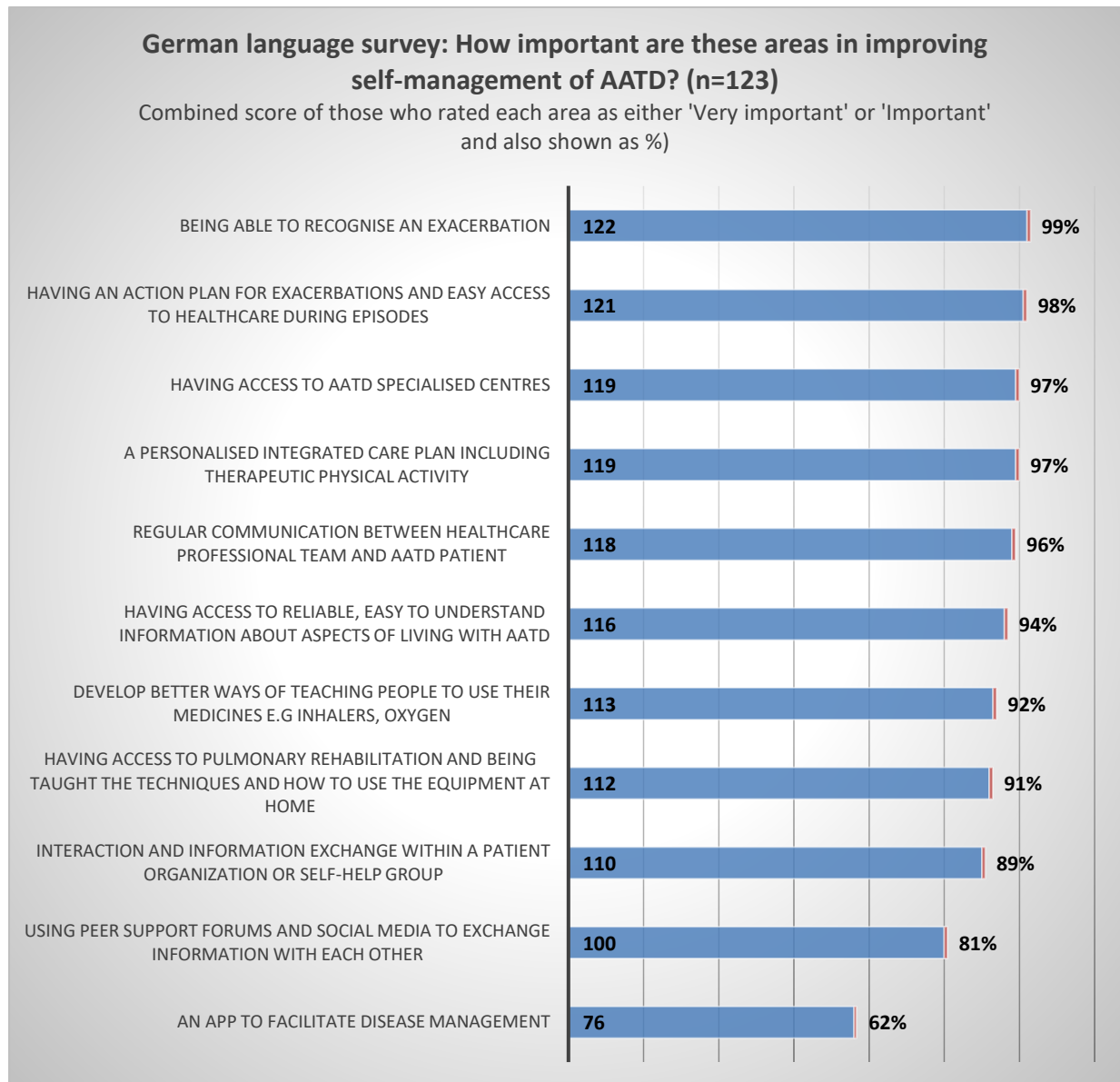
1. Smoking cessation (96%)
2. Develop other aspects of integral care (e.g. physical activity, care-giver support, maintaining work or schooling, nutrition, psychological care, sex-life, daily life) - 94%
3. Role of Pulmonary rehabilitation (94%)



## Self-management and education

Respondents were asked to rate a list of areas as to how important each is to improve self-management of AATD. The top 3 most important being:

1. Being able to recognise an exacerbation (99%)
2. Having an action plan for exacerbations and easy access to healthcare during episodes (98%)
3. Access to AATD specialised centres (97%) and A personalised integrated care plan (97%)



*“Mehr Lungensportgruppen es gibt viel zu wenige”* – More Pulmonary Sports there are far too few

## **Appendix 2: Data from the English language survey only**

A total of **142 respondents** to the English-language survey:

### **Characteristics:**

- 88% (n=125) a person diagnosed with AATD and 12% (n=17) a parent, relative or caregiver.
- 65% female, 34% male, 1% prefer not to say.
- The mean age of respondents was 52 years.
- Respondents who completed the survey were located in the following countries: United Kingdom (n=94), USA (n=12), Sweden (n=10), Denmark (n=7), Norway (n=6), Ireland (n=4), Germany (n=2), Finland (n=2), Australia (n=1), Slovenia (n=1), Cyprus (n=1), Canada (n=1), Belgium (n=1)

### **Smoking and alcohol:**

- 58% of respondents drink alcohol.
- 63% of respondents were a former smoker; 33% never-smoker and 4% current smoker.

### **Environmental exposure**

Respondents (n=136) were asked if they were/are exposed to gases, fumes or dust in their professional activities and 68% said No (32% said Yes).

### **Transplants**

Respondents (n=136) were asked if they had had a lung and/or liver transplant:

- 2% of respondents have had a lung transplant (n=3)
- 1% of respondents have had a liver transplant (n=1)

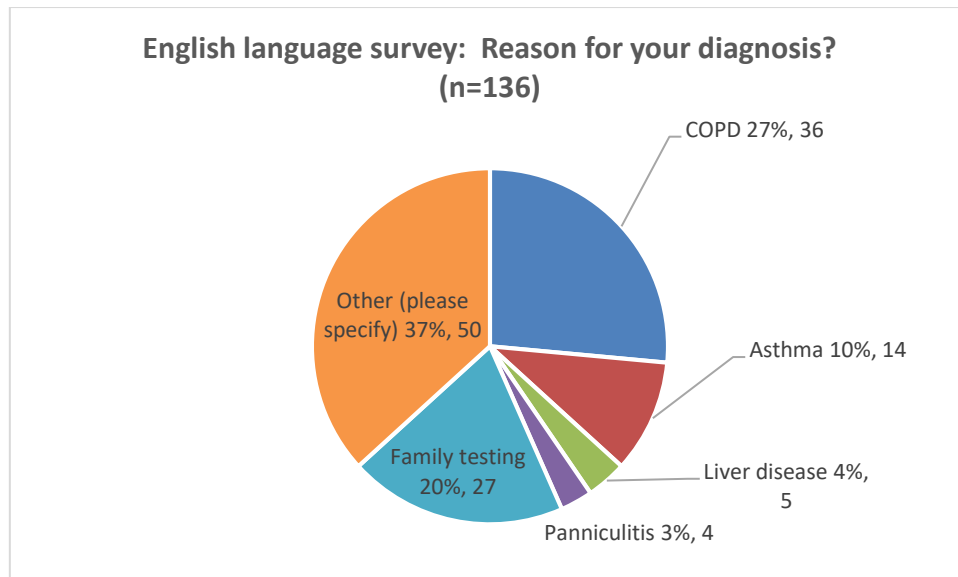
### **Diagnosis**

Respondents (n=136) were asked which healthcare professional diagnosed AATD:

- Respiratory specialist (e.g. pulmonologist) (44%)
- Family doctor (32%)
- Gastroenterologist/hepatologist (7%)
- Paediatrician (4%)
- Other (13%) – including Hospital; Geneticist; Dermatologist; Immunologist; Rheumatologist.

Respondents (n=136) were also asked about the reason for their diagnosis and 26% stated COPD, 20% Family testing and 10% Asthma – see full results in chart on next page:





**Other reasons given included:** Infections/pneumonia (often recurring) (n=14); when looking for/treating something else (n=6); shortness of breath (n=5); diagnosis of a family member (n=3); tiredness(n=3); weight loss (n=2); bronchiectasis (n=2); bronchitis (n=2); swine flu (n=2); aspergillosis (n=1); emphysema (n=1); abnormal liver function test (n=1); baby colic (n=1); jaundice (n=2); cough (n=1); ulcerative colitis (n=1).

**AATD phenotype:** 63% have ZZ phenotype; 7% MZ; 13% SZ and 1% MS; 11% did not know and 4% described as: Pi Z (n=1); Z null (n=2); Snull (n=1) and Ss (n=1).

**How many years had they been diagnosed with AATD:** this ranged from 'less than 1 year' to 45 years with the mean length since diagnosis being 11 years.

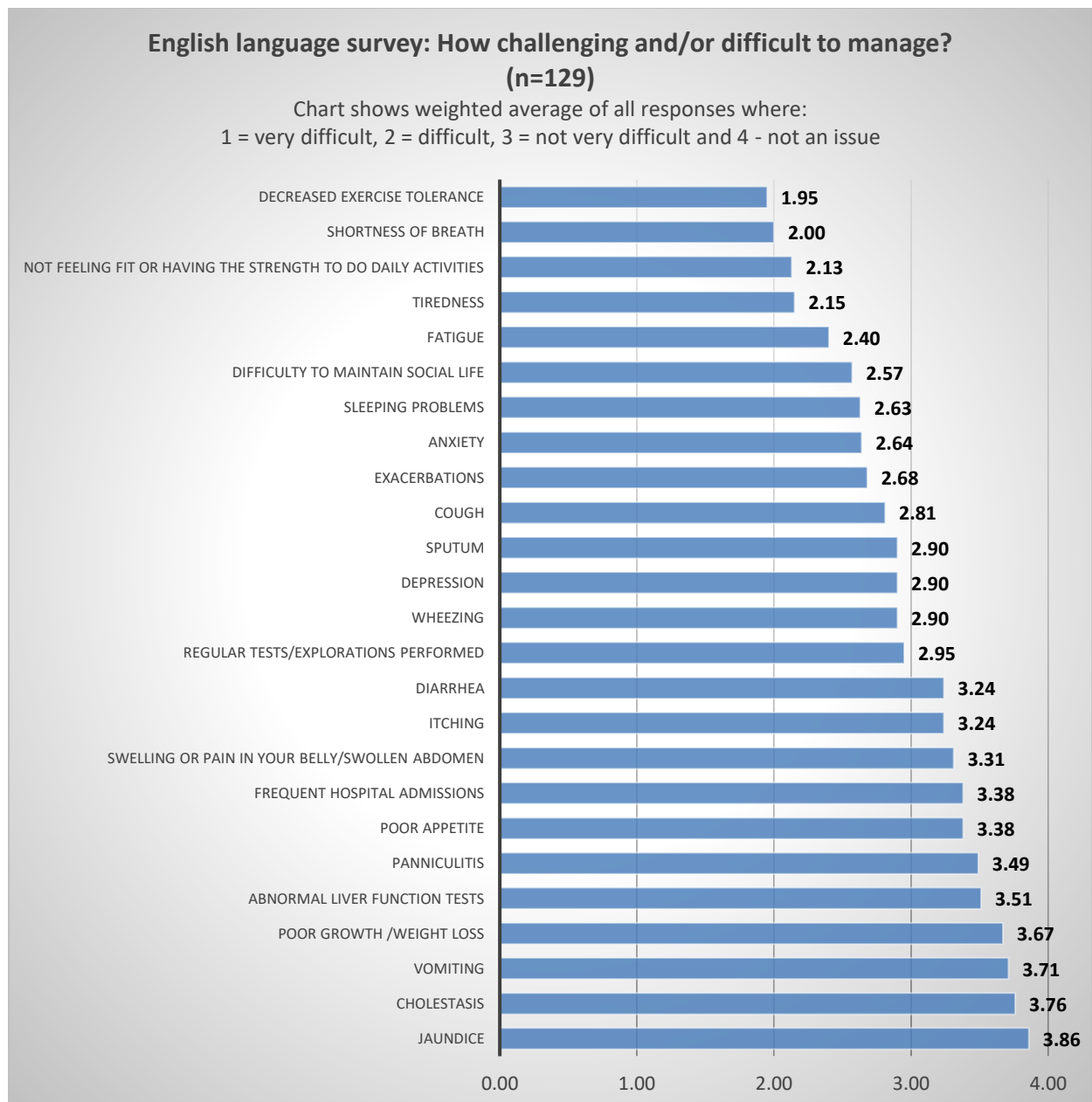
## Your experience of AATD

### Most challenging aspects of AATD

Respondents were asked to rate which aspects of the disease they found most challenging and/or difficult to manage during the past 12 months.

The Top 3 most challenging aspects were:

1. Decreased exercise tolerance.
2. Shortness of breath.
3. Not feeling fit or having the strength to do daily activities.



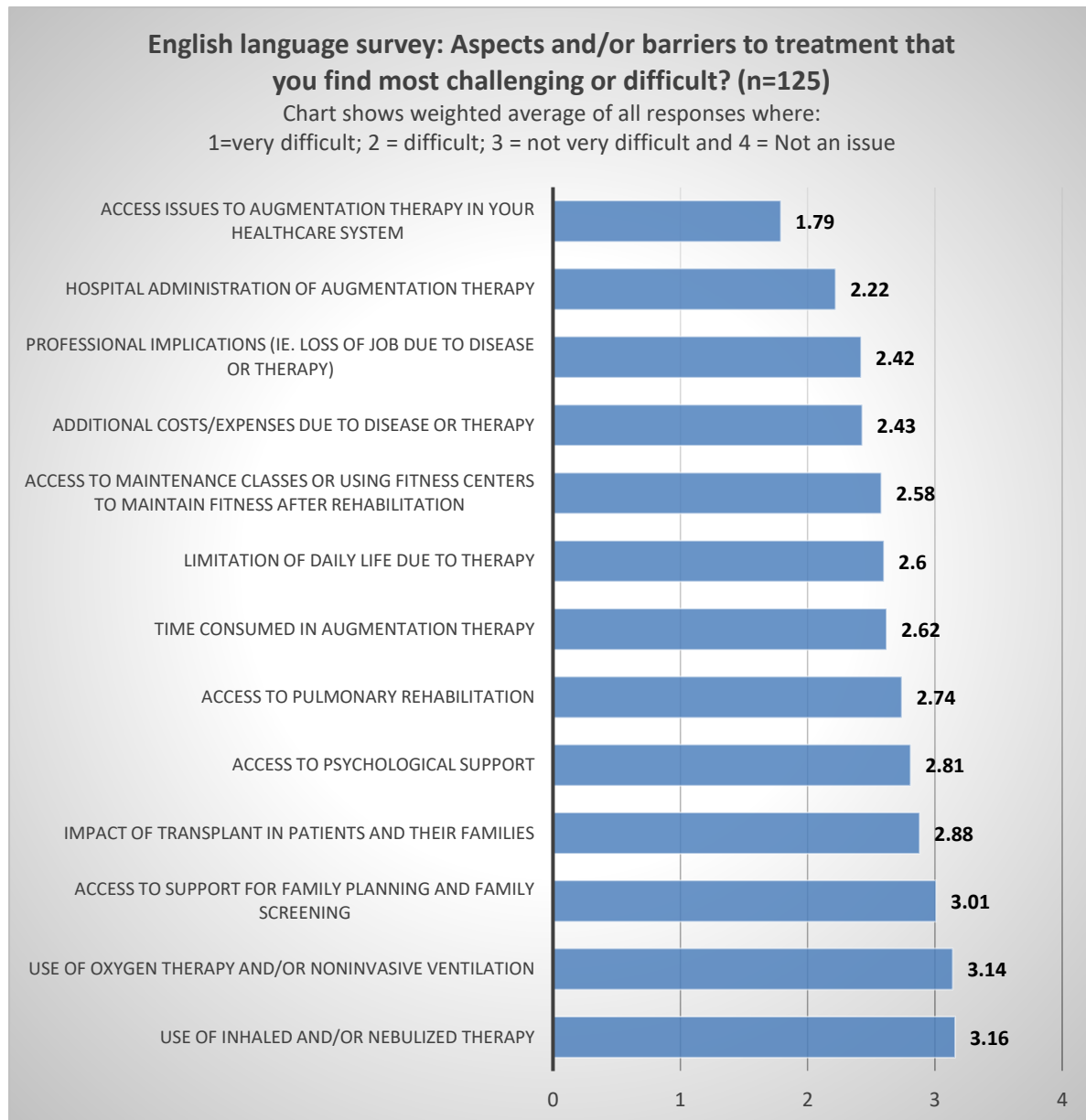
*“Overwhelming difficulty in losing all aspects of normal life. No motivation as no hope of recovery.”*

*“Intolerance to poor air quality (I live in London)”*

## Most challenging aspects/barriers for treatment

Respondents were asked to rate which aspects and/or barriers for treatment do you find most challenging and difficult to manage. The Top 3 most challenging aspects were:

1. Access issues to augmentation therapy in your healthcare system.
2. Hospital administration of augmentation therapy.
3. Professional implications (i.e. loss of job).



*“Knowledge and treatment of alpha-1 antitrypsin deficiency in Sweden is almost non-existence for us patients.”*

*“We don't get augmentation therapy in the UK that's the whole point. There is something that can't prolong a symptomatic sufferer's life, but we are not allowed it. Disgusting also being mZ even the specialists deny you any support uttered devastated my world as u have a young child.”*

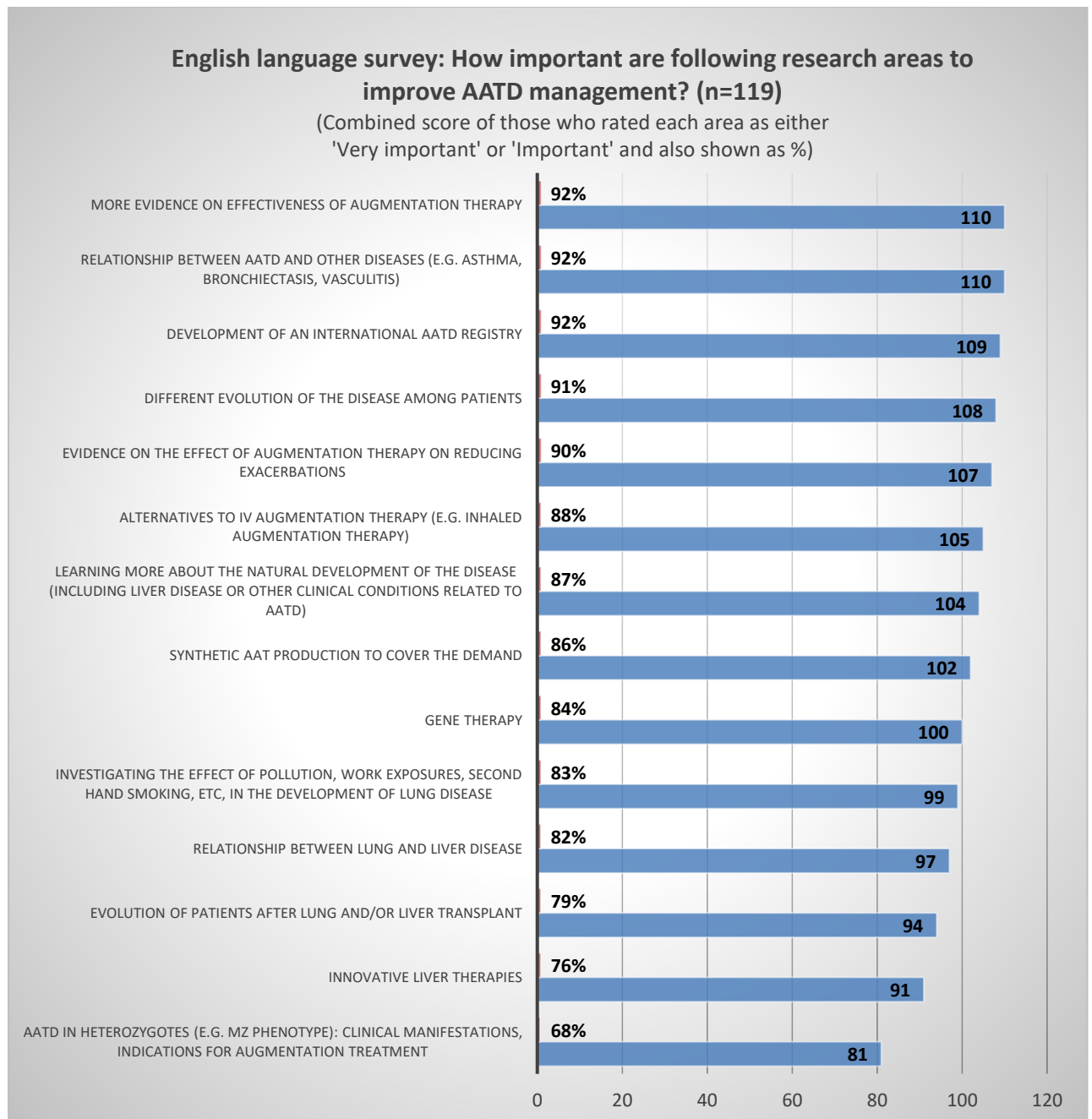
*“It is time consuming to make sure I do enough exercise to keep my lungs healthy, and expensive to eat well. Also expensive to pay for gym membership”*

## Areas of Research

### Improving AATD management

Respondents were asked to rate how important a list of research areas was to improve AATD management. The results below show the combined scores of those who rated an area as 'Very important' and 'Important'. The top most important research areas being:

1. More evidence on effectiveness of augmentation therapy (92%)
2. Relationship between AATD and other diseases (92%)
3. Development of an international AATD registry (92%)



*“The benefits of oral treatment versus inhaled treatment”.*

*“Progression of the disease in non-smokers and those in an environment free from smoke/dust /gasses etc.”*

*“Further research into AATD role in the skin. Research into which types/whether all augmentation therapy brands work specifically on panniculitis”.*

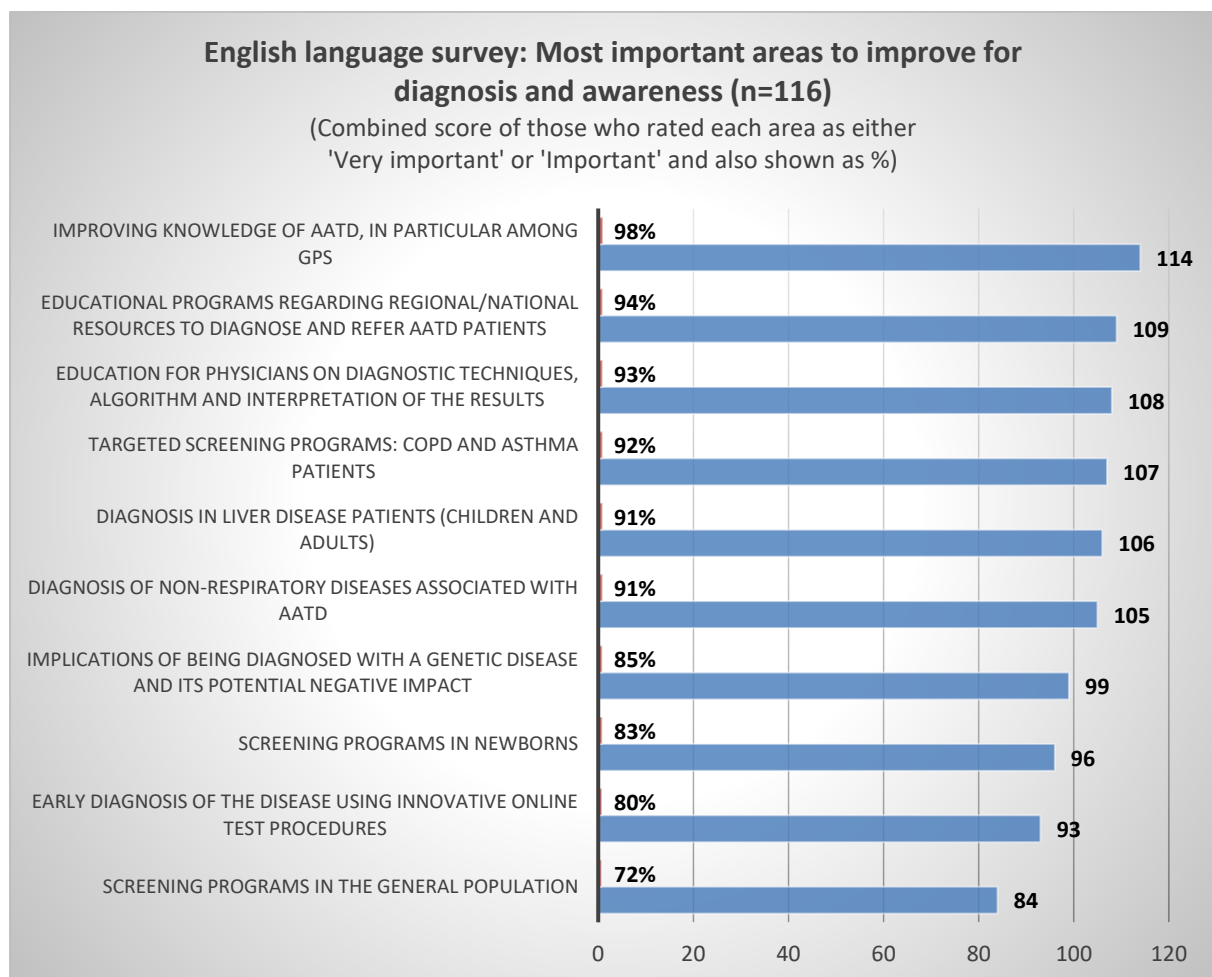
*“Make treatments available in the UK for all MZ’s heterozygous with lung & liver affected”.*

*“I have had AAT therapy whilst living in Spain and retained the same lung function. I now live in the UK with no access to this and have significant decline in lung function.”*

### Improving diagnosis and awareness of AATD

Respondents were asked to rate a list of areas as to how important each is to improve diagnosis and awareness of AATD. The results in the chart below show the combined scores of those who rated an area as ‘Very important’ and ‘Important’. All but 1 area was rated as 80% or higher with the top 3 most important being:

1. Improving knowledge of AATD, in particular among General Practitioners (98%)
2. Educational programs regarding regional/national resources to diagnose and refer AATD patients (94%)
3. Education for physicians on diagnostic techniques, algorithm, interpretation of results (93%)



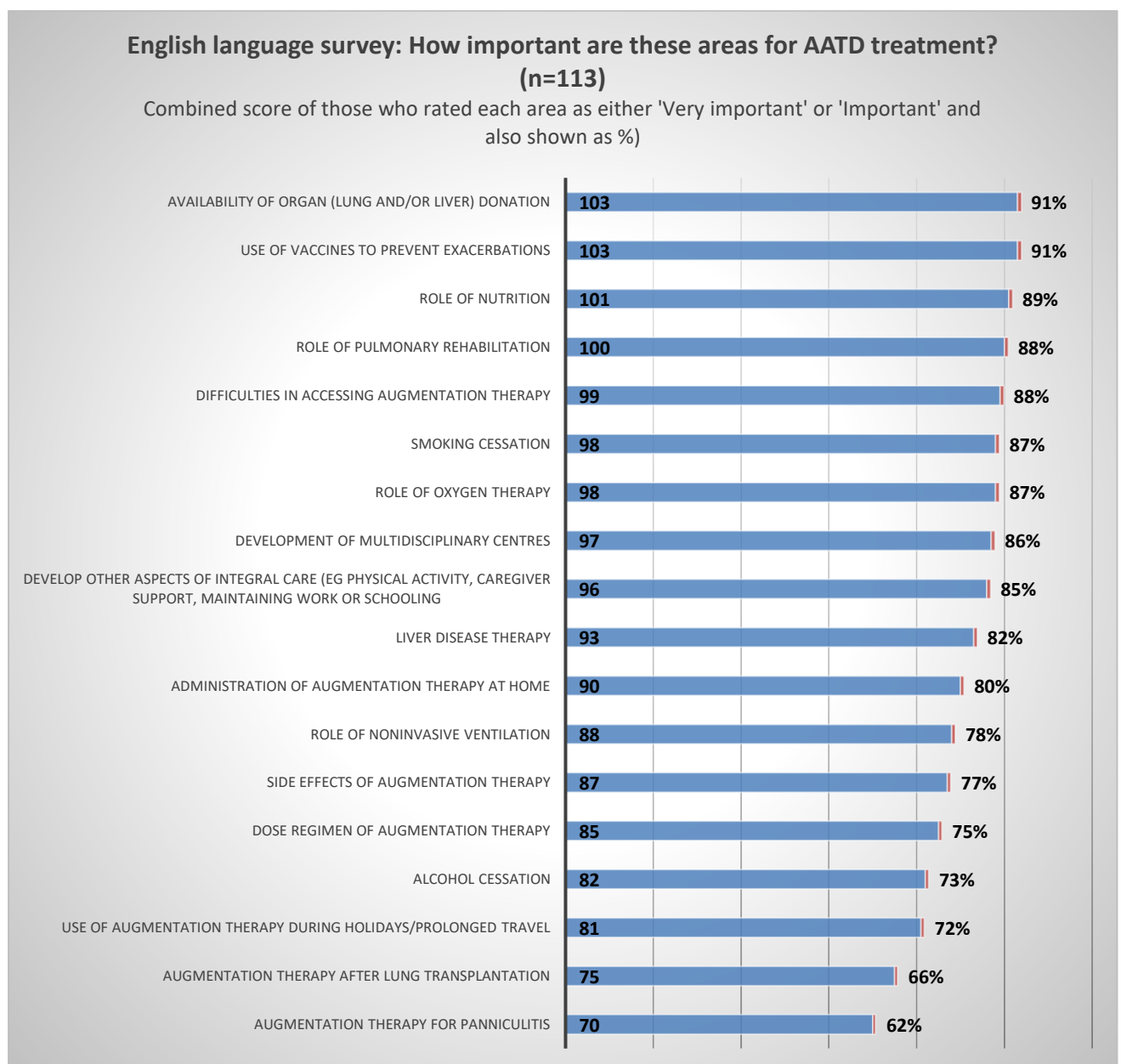
*“It is very important that general practitioners are educated because most of them know nothing about it”*

*“My answers reflect the incredibly difficult first 2-3 months post diagnosis, trying to get the NHS and GP to understand the diagnosis and support me. My own intensive research initially led me to have more knowledge than the GPs I spoke to and only through my persistence, did I finally get to a GP who was happy to learn and refer me to one of the specialist hospitals that I'd discovered existed in the UK, after finding my way to a Facebook support group. After then, I was 'in the system' and this was a massive relief in those early days, post diagnosis.”*

### Improving treatment

Respondents were asked to rate a list of areas as to how important each is to improve treatment of AATD. The results in the chart below show the combined scores of those who rated an area as ‘Very important’ and ‘Important’:

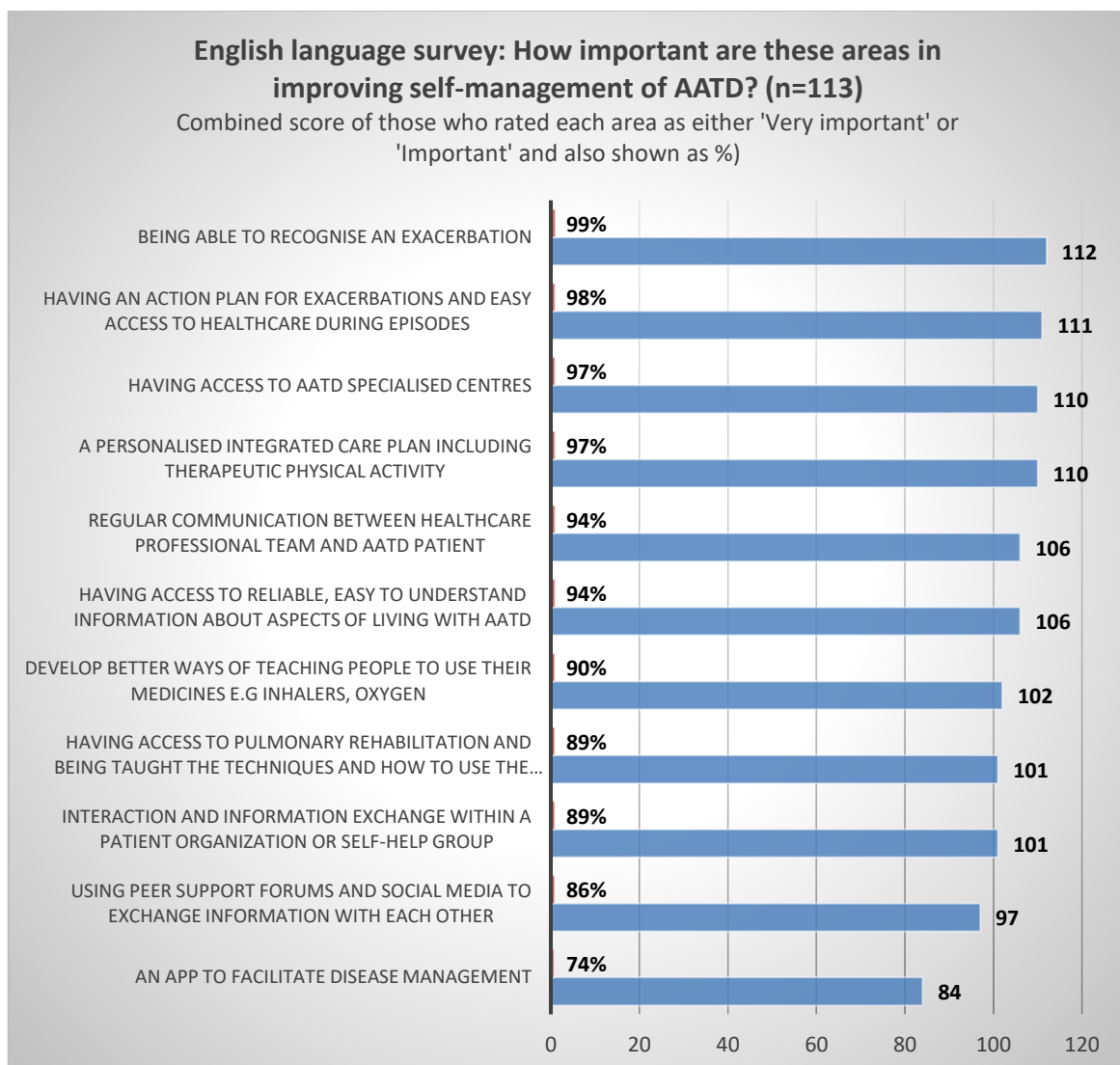
1. Availability of organ (lung and/or liver) donation (91%)
2. Use of vaccines to prevent exacerbations (91%)
3. Role of nutrition (89%)



## Self-management and education

Respondents were asked to rate a list of areas as to how important each is to improve self-management of AATD. The results in the chart below show the combined scores of those who rated an area as 'Very important' and 'Important'. The top 3 most important being:

1. Being able to recognise an exacerbation (99%)
2. Having an action plan for exacerbations and easy access to healthcare during episodes (98%)
3. Access to AATD specialised centres (97%) and A personalised integrated care plan including therapeutic physical activity (97%)



*"I have used an app called MyCoptd which has many features you mention already".*

*"I have experienced very long wait times for pulmonary rehabilitation. Approximately 6 months for a 6 weeks course with no follow up. I found this to be typical through contact from social media."*

*"Experts should teach other professionals and share their experience over years so that there is no gap when they retire... New experts have been appointed here but they have zero experience."*

*"Help in getting medical Insurance when you have AATD that covers all the meds." "Information on getting on the liver transplant list."*