



# Alpha-1 Alliance

## Alpha-1 Antitrypsin Deficiency Policy Report **Scotland**

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## 1. Foreword

As a patient suffering from Alpha-1 antitrypsin deficiency (Alpha-1), I welcome the policy recommendations in this report. Alpha-1 is a rare genetic condition that usually affects the lungs and liver, frequently causes disability and can significantly reduce life expectancy.

The report allows patients with Alpha-1 to make their voice heard and share their experiences of living with the condition with parliamentarians and officials. The testimonies by patients, their families and carers from across the country clearly demonstrate that more needs to be done for the Alpha-1 patients in Scotland.

It is important not to lose sight of patients affected by rare diseases and to invest in specialist services and effective therapies that are needed by only a small group of people.

I welcome that the Alpha-1 community has come together to form the Alpha-1 Alliance and look forward to working with NHS Scotland and MSPs on implementing the recommendations included in this report. Together we have the opportunity to ensure that the expertise within NHS Scotland is used to best support the specific needs of Alpha-1 patients.

**Karen North**  
Chair UK, Alpha-1 Alliance

As a practising clinician and scientific researcher who has worked for three decades within the field of Alpha-1 antitrypsin deficiency (Alpha-1), I welcome this report and fully endorse the policy recommendations arising from it. In the course of my own clinical practice, many Alpha-1 patients have described their experiences to me, and these are mirrored closely in the findings of the survey that are summarised in this report. It provides a representative account of how sufferers from this condition and their families are affected in many aspects of their lives and acts as a compass to guide the development of care for Alpha-1 patients in Scotland.

Although scientific understanding of this rare, inherited condition has improved vastly over the course of the 50 years since its initial discovery, awareness of Alpha-1 amongst healthcare professionals, as well as recognition by policymakers, has lagged significantly behind that of other rare diseases of comparable severity and frequency.

In the last 20 years, the clinical management of this complex condition has advanced in other European countries, whereas there has been little progress in improving care for affected patients in Scotland. The necessary clinical expertise and the will to improve the care of Alpha-1 patients already exist within NHS Scotland but there is currently no care pathway that delivers optimal and integrated multidisciplinary care for Alpha-1 patients.

I feel privileged to represent the Alpha-1 community as their chairperson in Scotland, and look forward to working with the Alpha-1 Alliance in order to support patients in obtaining equitable access to integrated healthcare services for their condition in Scotland that rival those of other European countries.

**Professor William MacNee**  
Chair Scotland, Alpha-1 Alliance



### 3. Introduction

Alpha-1 antitrypsin deficiency (Alpha-1) is a rare genetic disorder that leads to significant disability and early mortality. Alpha-1 most commonly results in lung and liver disease, which may be so severe as to require organ transplantation in early life. It can also affect other organs such as the skin. Alpha-1 associated lung disease is often initially diagnosed as asthma or chronic obstructive pulmonary disease (COPD), and the average delay in correctly diagnosing Alpha-1 is greater than seven years.

Patients with lung disease may be affected in a variety of ways. The most common symptom is shortness of breath, which can significantly limit their ability to work and perform normal daily activities, and which may progress with age, leading to potentially fatal lung failure. Patients are particularly prone to chest infections that worsen these symptoms and lead to an increased risk of hospitalisation and death. Chest infections are a particular problem in the winter months, so that patients tend to avoid going 'out and about'. Patients with liver disease are often tired and weak. They can also feel sick and lose their appetite, which interferes with their normal daily activities. When this is severe, individuals experience jaundice, sickness, diarrhoea and potentially fatal liver failure.

Although extensive expertise in Alpha-1 exists in Scotland, there is considerable variation in patient access to specialists, and no access to specific therapies for Alpha-1 associated lung disease. There is also no care model within NHS Scotland that provides integrated clinical management for the unique needs of the different clinical aspects of Alpha-1.

This report, and the recommendations contained within it, is informed by the findings of a survey of Scottish Alpha-1 patients, their families and carers. It contributes to a campaign to establish a nationally commissioned highly specialist service for Alpha-1 that provides equitable access for patients to optimal, integrated clinical care for this complex disease and the unique needs of Alpha-1 patients.

Find out more online  
[www.alpha-1-alliance.org.uk](http://www.alpha-1-alliance.org.uk)

## 4. The Alpha-1 Alliance

In 2012, the Alpha-1 community established the Alpha-1 Alliance, an unprecedented coalition of patients and leading clinicians in the field of Alpha-1 from across the UK. The objectives of the Alliance are to raise the profile of the unmet medical need of Alpha-1 patients and to campaign for better and equitable access to healthcare services for these patients, including the provision of coordinated multidisciplinary specialist care via nationally commissioned expert centres and timely access to any existing and forthcoming licensed and effective medicines for this condition. The Alpha-1 Alliance is chaired by Karen North in the UK and Professor William MacNee in Scotland.

## 5. Purpose of the report

- To identify the areas of unmet medical need for Alpha-1 patients.
- To evaluate differences in access to and the quality of available medical care for Alpha-1 patients across Scotland.
- To establish the impact of the unmet medical need on relatives and carers of Alpha-1 sufferers.
- To improve parliamentarians' and policymakers' understanding of the needs of the Alpha-1 community, including those of families and carers.
- To ensure the Alpha-1 community's views are represented in policy discussions about relevant healthcare services.
- To secure ongoing political support for the improvement of healthcare services available to the Alpha-1 patient community.

## 6. Methodology

The survey was conducted online amongst Scottish Alpha-1 patients, their families and carers from November 2012 to August 2013, and was accessible via the main Alpha-1 patient support charities' websites. Results were collected anonymously. The majority of questions were asked only of patients; those questions that were asked of family members and carers are specified in the report. The questions were developed jointly by the Alpha-1 Alliance Secretariat in consultation with the Alliance Executive and clinical experts in the field.

Several survey findings were complemented by data from a systematic audit of Alpha-1 patients identified in Scotland since 1999, which was conducted in 2014 by Professor William MacNee from the University of Edinburgh and his team. The audit is based on patient-level data provided by the Information Services Division (ISD), a division of National Services Scotland, part of NHS Scotland.

## 7. Demographics

36 respondents from all parts of Scotland returned completed surveys; 31 responses were submitted by patients and 5 by family members or carers. The results presented in this report include all responses. The vast majority of patient respondents reported suffering from the most severe form of Alpha-1, genotype PiZZ. Most respondents reported experiencing predominantly lung-related symptoms, and one fifth of respondents also reported liver problems.

The systematic audit of ISD data identified 191 PiZZ patients in Scotland (94 female, 96 male, 1 unknown).

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[www.alpha-1-alliance.org.uk](http://www.alpha-1-alliance.org.uk)

## 8. Survey results

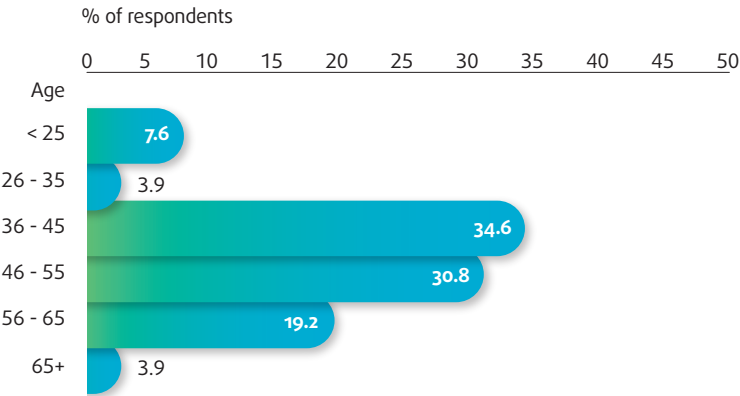
### 8.1 Diagnosis

#### Age at diagnosis

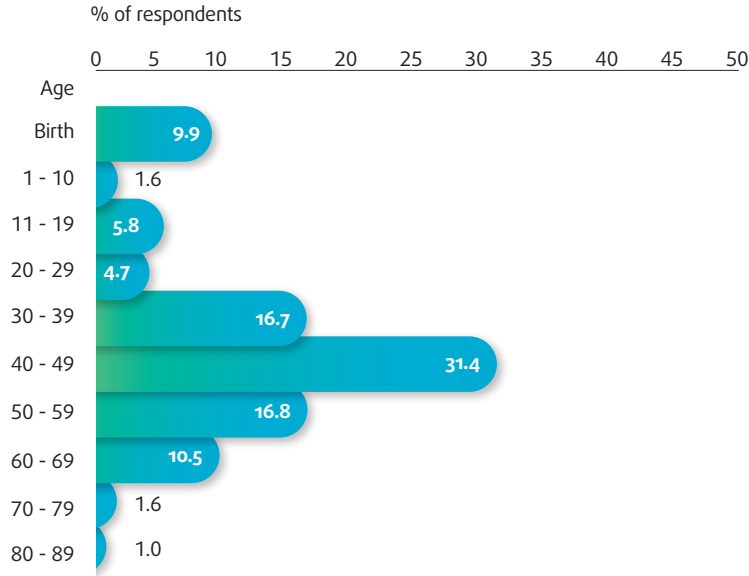
The majority of respondents were diagnosed with Alpha-1 between the ages of 36 and 65, with over one third having received the diagnosis between 36 and 45 years of age.

Data from the Scottish patient audit confirmed the survey results, with two thirds of patients found to have received the diagnosis of Alpha-1 between the ages of 30 and 59 years.

How old were you when you were first diagnosed with Alpha-1?



Age at diagnosis of Alpha-1?





## Misdiagnosis

Many respondents reported that they were initially misdiagnosed and received treatment for other respiratory conditions, most commonly asthma, before being correctly diagnosed with Alpha-1.

The long journey through the healthcare system that respondents encountered before receiving the correct diagnosis was reported to have impacted substantially on their quality of life. Patients felt helpless about not knowing the cause of their progressively worsening symptoms and about the lack of response to the medications that they had been prescribed.

*"I was devastated, with fear of an unknown future."*

*"The doctors always diagnosed my condition as asthma, and the medication prescribed was inhalers and tablets. I found that these treatments were not sufficient to help my problems."*

*"I have been totally exhausted for years; I was looking for a diagnosis, but not one for which there is no medication available to help my condition."*

*"I was labelled as neurotic at the age 40, after having been investigated many times from the age of 25 without getting a diagnosis. In my case multiple organs are involved - chest, skin, liver."*

*"After having been treated for asthma for years, at the age of 40, my respiratory nurse felt my lungs were in such a bad condition that further investigation was necessary. She did an Alpha-1 test just to rule it out!"*

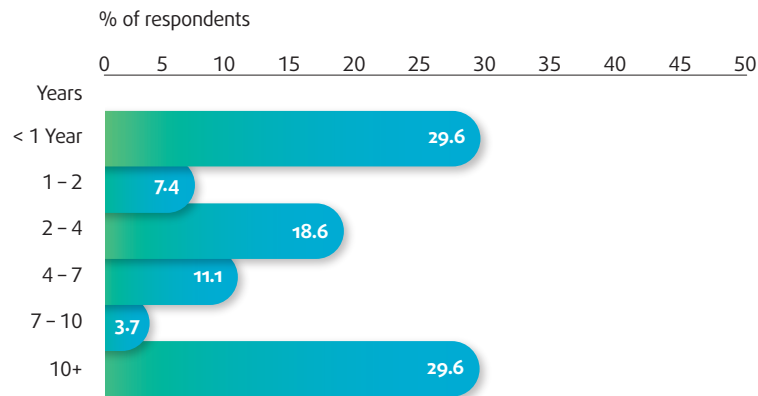
*"I was relieved to finally know what was wrong."*

*"I had a very awful experience. For a number of years I was treated for bronchitis. I saw different GPs, then a respiratory consultant. He told me, after looking at my long history of chest infections, that nothing serious was wrong with me."*

## Delayed diagnosis

A third of patients experienced a delay of more than seven years after the initial onset of their symptoms before receiving a correct diagnosis of Alpha-1. Many of the respondents reported that they had to consult many different doctors before finally receiving the correct diagnosis, which left them in a position of uncertainty as to the cause of their symptoms, sometimes for many years.

After first experiencing symptoms, how long did it take for you to receive a diagnosis of Alpha-1?



“I had been to my doctors with chest infections many times and they could not tell me what was wrong. I saw at least five doctors.

## Need for early diagnosis

Respondents commonly felt that an early correct and accurate diagnosis, combined with advice on lifestyle changes, would have been greatly beneficial in decelerating the progression of their condition.

*"Knowing the diagnosis has helped me to do all I can to reduce the condition's present and potential future impact on my well-being."*

*"I was extremely concerned and worried about the damage it may already have inflicted before I was diagnosed."*

*"I have changed my lifestyle completely - given up smoking was the single biggest health improvement for me."*

*"I was a bit shocked at being told at the age of 52 that I had Alpha-1 because, obviously, I was born with this deficiency. I believe the test for Alpha-1 should have been done sooner, especially since I was diagnosed with COPD when I was relatively young."*

## 8.2 Burden of disease

The survey indicated that Alpha-1 affects patients' lives at many different levels and represents a major burden for patients, and for their families and carers.

*"Since diagnosis, my health has noticeably deteriorated and my quality of life is very poor now."*

### General activity and mobility

The inability to be active and mobile as a result of the shortness of breath and associated immobility that most patients experience was highlighted by many respondents.

*"Alpha-1 totally dominates my life, I am now pretty well housebound."*

*"I become breathless if I have to walk upstairs, and I can't lift or carry anything."*

*"If I am out shopping, I have to try and park as near as I can - I have a blue badge that I could not manage without."*

*"I live at the bottom of a hill and when we moved in, I was able to walk up the hill. I have been unable to do this for some years now."*

### Everyday activities

Respondents reported that, as the condition progresses, it has a significant impact on their ability to live a normal and fulfilled life. Many patients struggle to perform normal everyday activities.

*"Showering, dressing, tying shoes, shopping, doing the garden, holidaying - anything physical is a struggle."*

*"I am always tired and unable to do even simple tasks without feeling breathless."*

*"Everyday tasks are done in stages ... huffing and puffing my way through, with regular breaks to regain my breath."*

*"Swimming was something I used to do daily and I miss it a lot."*

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online visit  
[www.alpha-1-alliance.org.uk](http://www.alpha-1-alliance.org.uk)

## Work and social life

Respondents were asked to detail how their health problems limit their ability to work, or to take part in recreational and social activities. The responses clearly demonstrate how severely Alpha-1 affects patients' ability to engage in employed work and lead a fulfilling personal life, with a profoundly deleterious effect on their quality of life.

"I had to give up the job I loved - as a social worker I had to do home visits and carry children who were in care which I am no longer able to do."

"It affects all the things I loved to do in life: working STOPPED, playing in the park with the kids STOPPED, sex life STOPPED, friends STOPPED visiting, social life STOPPED."

"I cannot do a full-time job as I just don't have the energy."

"Alpha-1 has prevented me from taking part in family gatherings, social nights out, or performing any form of physical exercise."

"Socially I used to love going out and really enjoyed dancing - I can't even get up for one dance now."

## Independence

The survey showed that many patients feel distressed about losing their independence and becoming a burden for their families, often at a young age and at a time when they are trying to bring up a family.

"Now my wife not only looks after my two kids who are 5 and 6 years old, she needs to look after me too."

"I am very lucky to have my husband - without his support and love I would be in hospital a lot more often."

"I cannot survive without the aid of my wife and I have to rely on her doing many things needed to keep me alive."

"I retired early and am now confined to home and permanently attached to an oxygen concentrator."

## Mental health

Several respondents reported that the disease also affects their social well-being and mental health. They also feel anxious about their future and that of their families.

*"Sometimes I feel so alone and lonely, and with people who I thought were close friends no longer visiting or phoning I begin to feel like a leper."*

*"My moods are low and some days I don't want to get out of bed."*

*"I'm on medication for depression - Alpha-1 made me housebound and I now live a quiet and sedate life style."*

*"I struggled for a long time with poor mental health as I had lost all my confidence after being so ill."*

*"I find it very difficult to enjoy anything and get depressed about knowing that I cannot do the things with my family that I used to do."*

## 8.3 Impact on families and carers

The survey suggests that most carers of Alpha-1 patients are family members who, as a direct consequence of the condition of their loved ones, experience a significant impact on their own ability to have a productive and fulfilling life. In our survey, family members and carers reported that the flexibility in their own work and social lives was significantly reduced by having to care for an Alpha-1 patient. They also reported experiencing anxiety about the effects of the disease on the patient, and on their families.

*"I lost my spouse to Alpha-1. My life was torn apart as a result because of the loss I have suffered."*

*"I haven't had children of my own partly because I fear passing Alpha-1 on to them."*

*"This disease is horrific. My two brothers suffered terribly and passed away at young ages of 40 and 35 years."*

*"I have been a constant source of support for my brother, but it is difficult."*

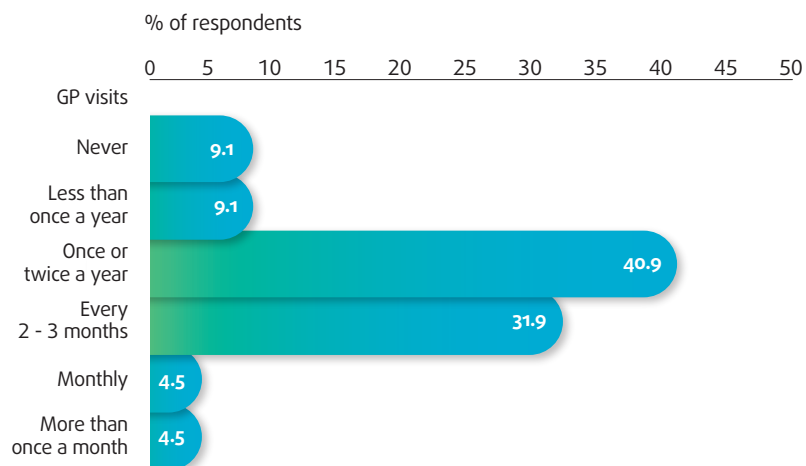
*"The family has become closer as we are frightened we will lose him."*

## 8.4 Access to medical services and treatment

### Primary care

Over 40% of respondents reported having to visit their GP more than once every three months to receive help for their condition, indicating a high level of unmet medical need. Some respondents commented on encountering barriers to easy access to doctors, leading to potentially harmful delays in obtaining appropriate treatment.

On average, how frequently do you visit your GP because of your condition?

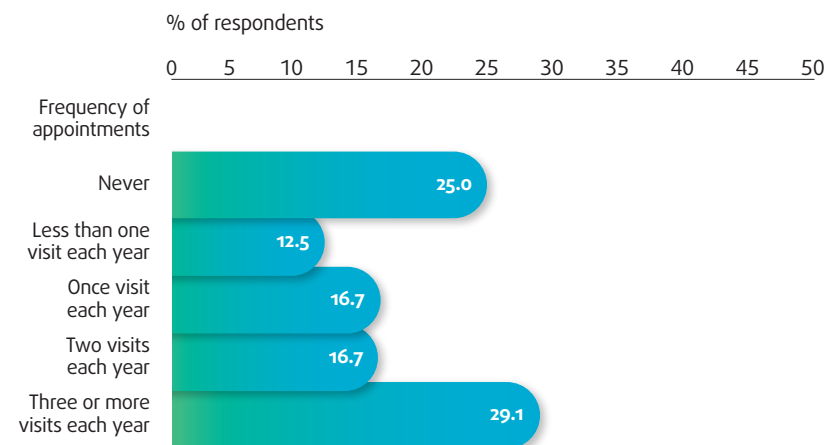


“ I would like to have better access to my doctor when I have a chest infection.

### Secondary care

Utilisation of medical services in secondary care was also high amongst respondents, with almost half of the patients needing to attend clinical specialists in relation with their Alpha-1 at least twice a year. Only 38% of respondents said that they had ever seen a specialist respiratory nurse for their condition. Of those, the majority found the consultations beneficial and desire more frequent access to these nurse specialists.

On average, how frequently do you attend appointments with a specialist clinician for your condition?



“ When I do get to see my lung specialist, there is only very little consultation time with him.

### Accident & Emergency attendances and hospital admissions

42% of respondents reported having to attend Accident & Emergency in the preceding five years, indicating that Alpha-1 patients experience episodes of acute medical need that are not preventable by current routine care arrangements.

In addition, 46% of respondents were admitted to hospital for their condition in the preceding five years, with many patients reporting multiple admissions in this period.

### Prescribed medicines

The majority of the respondents receive only standard prescription medications for relief of their symptoms and additional treatments on those occasions when they suffer acute episodes, such as respiratory infections. Most patients reported having easy access to routine prescriptions via their GP or specialist nurse, but difficulty in obtaining emergency medication was identified as a problem.

*"I was told that I am on all the available medication suitable for my condition."*

*"I have been given easy access to medication by my doctors."*

*"I receive standard COPD treatments; nothing targeted at Alpha-1 specifically."*

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Many respondents reported to be dependent on long-term supplementary oxygen therapy, although access to regular monitoring varied.

*"I have purchased my own portable oxygen concentrator - the one available on the NHS is not suitable for my lifestyle."*

*"I am now reliant on supplementary oxygen 24 hours a day."*

*"I'm on home oxygen and was told they will visit me six-monthly, but I have had problems. I think the visits should be a bit more frequent than once every six months anyway."*

## Genetic counselling

The survey showed that genetic counselling was rarely proactively offered to patients by NHS Scotland doctors following a diagnosis of Alpha-1. Many respondents reported that genetic counselling was only provided after it had been explicitly requested by them. Only 17% of respondents had received any genetic counselling. Of those, however, 80% rated their overall experience of genetic counselling as 'very good or good'.

In general, respondents found genetic counselling important and very helpful in understanding the disease and its implications for other members of their family.

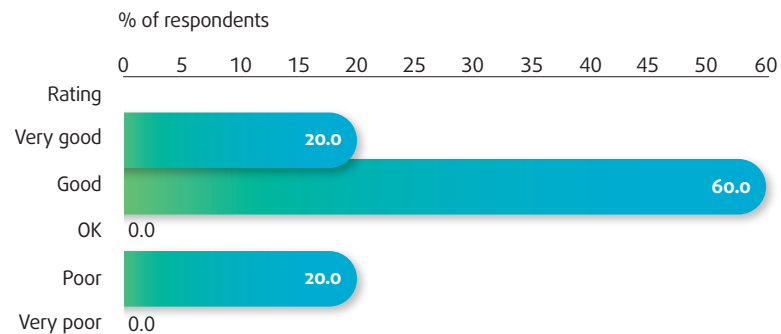
*"It was very important and helped alleviate many fears and concerns."*

*"The counselling was good."*

*"Genetic counselling provided the opportunity for other family members to be tested."*

*"They explained the nature of the disease, and I then informed my siblings who were also tested."*

### How would you rate your overall experience of receiving genetic counselling?



*"I would have liked the opportunity to speak to a genetic specialist about my condition."*

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### Pulmonary rehabilitation

Only half of the respondents had managed to participate in pulmonary rehabilitation programmes. Although the quality and benefit of pulmonary rehabilitation was assessed very positively by patients, the limited access to this service appears to be a major problem and was repeatedly criticised.

*"I found it very beneficial. It was about two and half years ago and I would like very much to have another session."*

*"I had a very good respiratory nurse. It's a shame that these nurses cannot visit at home when you reach the later stages of your illness."*

*"I did gain benefits from pulmonary rehabilitation as it improved my fitness levels, and they also gave advice on diet."*

*"It was extremely beneficial; unfortunately it took me two years to get a place."*

*"Pulmonary rehab proved invaluable in boosting my confidence. The sad thing is that, at the time I did it, there were no follow-up classes available."*

### Organ transplantation

Some patients reported either having undergone lung transplantation or being on the waiting list for transplantation, which is considered the only effective means for prolonging life for Alpha-1 patients currently available in Scotland.

*"Drastically, in 2010 I received a bilateral lung transplant."*

*"I was assessed for lung transplantation, but I was told I'm too ill for it now."*

### Regional inequalities in access to appropriate healthcare

Results from the survey suggest that wide variation exists across Scotland in access both to healthcare professionals with specific expertise in Alpha-1 and to tailored treatments across Scotland.

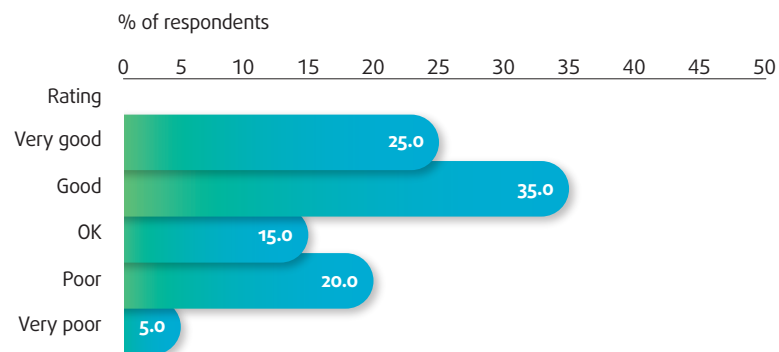
*"I have to travel to separate clinics for the different problems with my condition."*

*"The pulmonary rehab was not available locally and I had to go to Edinburgh. I feel that these courses should be run regularly by every local health authority."*

## 8.5 Quality of care

Reports of the quality of treatment and support patients receive for their condition varied significantly and were rated as 'very good or good' by 60% and as 'poor or very poor' by a quarter of respondents. This indicates that there are no uniform care standards for the treatment of Alpha-1 in Scotland and that regional variations exist in the quality of care and the availability of support for this condition. The lack of awareness of the disease by healthcare professionals and the available options for treatment were criticised by many of the respondents.

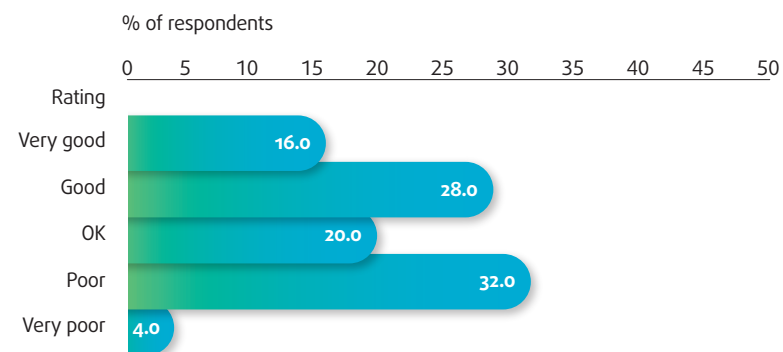
How would you rate the quality of the treatment and support that you currently receive?



## Awareness and knowledge amongst healthcare professionals

Many respondents highlighted the lack of medical knowledge about the disease amongst GPs. 44% rated their GP's level of knowledge about the condition as 'good or very good', but more than a third of the respondents felt that their GP had 'poor or very poor' knowledge of Alpha-1. This result is reflected in the long delay patients experience before receiving the correct diagnosis, which is likely to be a consequence of a lack of awareness of the condition.

How would you rate your GP's level of knowledge of Alpha-1 and its effects on your health?

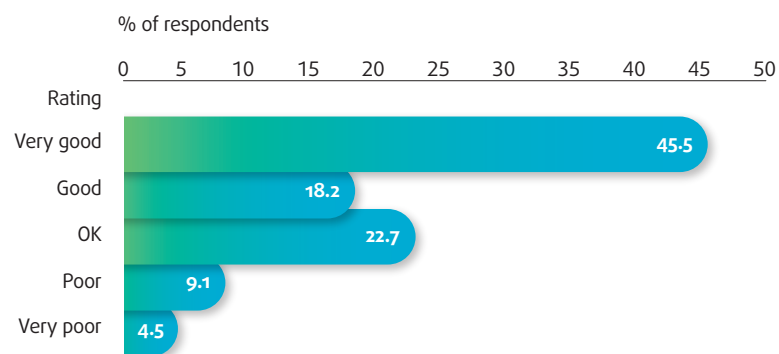


“The doctor at my surgery had to look up Alpha-1 online as they had never heard it.

My doctor also has Alpha-1, so he has quite a lot of knowledge.

The survey suggested that the level of knowledge about the disorder amongst specialist clinicians also differs across Scotland, although it was rated as 'very good or good' by nearly two thirds of respondents.

#### How would you rate your specialist clinician's level of knowledge of Alpha-1 and its effects on your health?



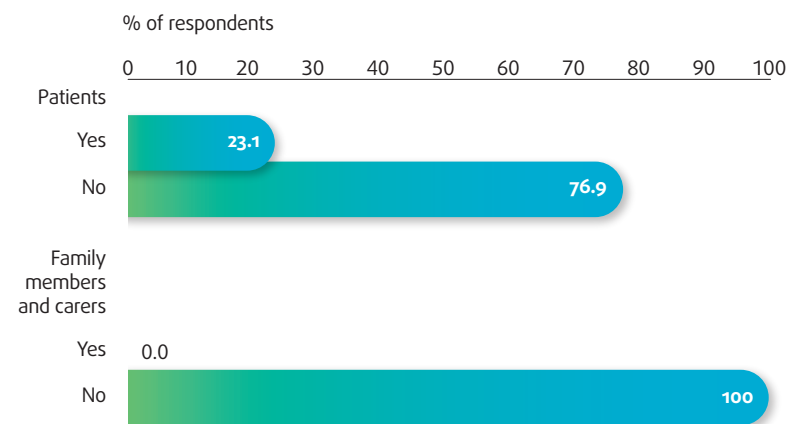
“My consultant is ok and seems to know the symptoms etc. However, he is not an Alpha-1 specialist.

*My liver consultant was most helpful.*

#### Information provided by the NHS

Respondents were generally not satisfied with the level of information they received for their condition, with all family members and carers and 77% of patients feeling that NHS Scotland does not provide sufficient information about the disease, available healthcare services and treatments for Alpha-1.

#### Do you feel that you have enough information about NHS treatments and services available to patients with Alpha-1?



“I was told that, as I am so good with computers, I should conduct my own research.

*I do find it upsetting that information leaflets are not commonly provided by local hospitals and GP practices.*

### Medical treatment, care and support

Respondents expressed their concern about the lack of adequate treatment and care options that exist in Scotland for their disease.

*"There are no specific treatments or services for patients with Alpha-1 - the only support is via support groups run by patients for patients."*

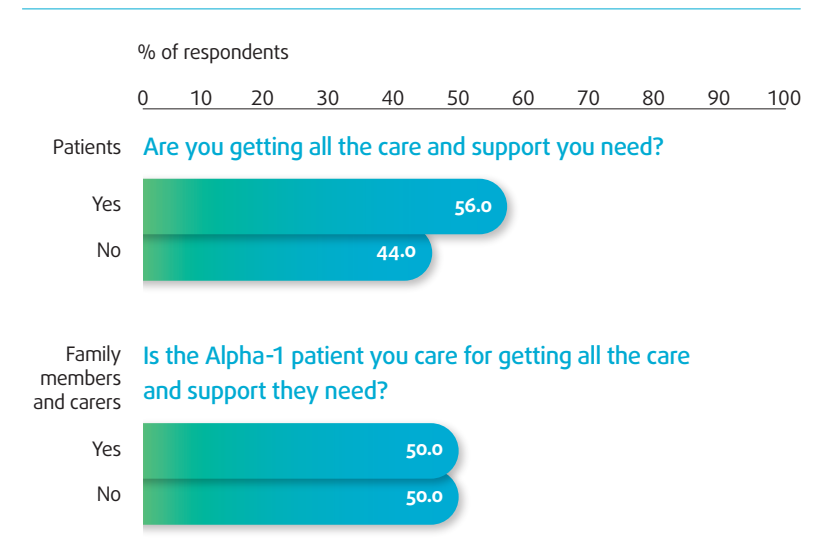
*"I receive standard COPD treatments, but nothing targeted at Alpha-1 specifically."*

*"There is a lack of information, advice, treatment options and recognition of the disease."*

*"I don't feel supported by the NHS, in contrast to one of my sisters who is on augmentation therapy in Belfast."*

*"I don't believe I have ever had any real support relating to my Alpha-1."*

Overall, 44% of patients felt that they are not receiving the care and support they need for their condition on various levels. This opinion was mirrored by family members and carers. In particular, respondents expressed their dissatisfaction with the lack of knowledge about Alpha-1 amongst healthcare professionals, and with the inadequate support structure in the NHS for their unique and complex needs.



*“ I feel let down by the NHS.”*

## 8.6 How should NHS services improve to meet Alpha-1 patients' needs?

### Information, care and support

Respondents were asked what additional information, care and support they feel they need to cope better with their condition. Patients as well as their family members and carers felt the need for NHS Scotland to improve in all of these areas. The majority of respondents stated that awareness and knowledge about the condition needed to improve amongst healthcare professionals, particularly GPs and specialist nurses. The level of information provided by NHS Scotland on the disease, its implications for patients and their families, the location and availability of specialist clinicians with expertise in Alpha-1 and available treatment options were also highlighted as areas that require urgent improvement. Several respondents felt that, due to the low profile of Alpha-1 and the current lack of treatment options for the condition that are available in Scotland, they did not receive the same level of attention and care as other rare respiratory conditions, such as cystic fibrosis.

*"Being diagnosed with Alpha-1 should no longer be the beginning of a journey into the unknown."*

*"We need more information about recent research and available treatments and better access to Alpha-1 specialists."*

*"It would be important to have systems in place to immediately help newly diagnosed sufferers."*

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*"Everyone knows about diabetes, why shouldn't everyone know about Alpha-1?"*

*"Access to consultants who know about and specialise in Alpha-1 would help to improve Alpha-1 care and diagnosis of related conditions."*

*"Education of clinicians and nurses about Alpha-1 should be improved, and a much wider recognition of the disease amongst healthcare personnel would help."*

*"Doctors need to take the illness as seriously as cystic fibrosis, as Alpha-1 can also lead to death."*

*"There should be more continuous support for our condition and a support structure for our families."*

Medical services and treatment options

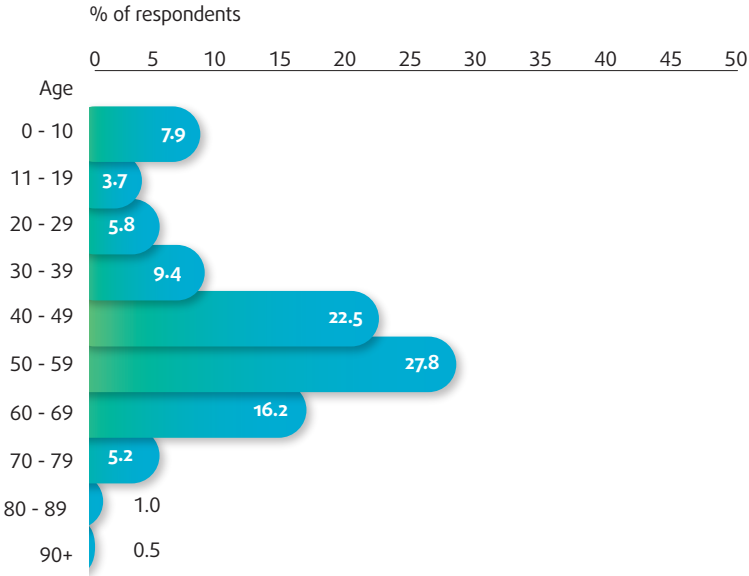
Respondents were asked what services and treatments for Alpha-1 sufferers they feel NHS Scotland should provide or improve access to. Respondents most frequently called for more equal access to services, a nationally uniform approach and best practice standards, and access to treatments that are available in many other countries. The need for a multidisciplinary approach was also highlighted.

- "All patients should have the same care, attention and treatments available to them."*
- "Specialist treatment centres that would have the expertise to diagnose, monitor, follow-up and treat when the condition gets worse."*
- "Availability and frequency of pulmonary rehabilitation sessions should be improved."*
- "Access to augmentation therapy and other effective Alpha-1 specific treatments, in line with the standard in other countries."*
- "Home visits from respiratory nurses and general assessments should be more frequent."*

High burden in the working population

The Scottish patient audit found that over three quarters of patients with the most severe form of Alpha-1 are below 60 years of age, and 50% are less than 50 years old. This group of patients represents a working population, and the disease will therefore impact significantly on these individuals' ability to sustain their livelihood. This data confirms that Alpha-1 is a disease that first affects people at a young age and, because of its chronic progressive nature, will worsen with age and reduce life expectancy. Therefore, early effective management is essential to prevent or delay disease progression, and this is best achieved through dedicated specialist centres with appropriate expertise in Alpha-1.

Current age of Alpha-1 patients in Scotland (PiZZ genotype)



### Support for a nationally commissioned highly specialist service for Alpha-1

All respondents to our survey were unanimous in their strong support for the establishment of a national highly specialist service for Alpha-1 in Scotland. Many patients expressed the notion that such an expert-led service could fill the gap they currently experience in their clinical care, improve access to optimal treatment, provide multidisciplinary management of their condition and facilitate more continuity of care.

*"A national service would help to establish standard procedures for identifying and treating patients, raising general awareness and training interested clinicians."*

*"Doctors currently treat us as COPD patients, not as Alpha-1 patients. We have associated problems which are completely unrelated to COPD (skin disease, liver disease and many more) which an Alpha-1 specialist would pick up as related issues. Recognising this would improve response and treatment times resulting in the saving or prolonging of life."*

*"Specialist centres would provide points of contact for experts who understand the condition."*

*"It would hopefully give access to augmentation therapy which has been available for over 20 years in other countries where it has shown to be a safe and beneficial treatment for affected patients."*

## 9. Summary

The results of a national survey of Alpha-1 patients, their family members and carers highlighted the acute unmet medical need that Alpha-1 patients currently experience in Scotland. A number of particular issues with the current level of care were emphasised by patients including:

- Lack of awareness and knowledge of the condition amongst healthcare professionals, resulting in long delays before receiving the correct diagnosis.
- Lack of patient information about the disease, its implications and available treatment options.
- Inequitable and restricted access to services and adequate therapies (e.g. genetic counselling, respiratory rehabilitation, supplementary oxygen).
- The need for coordinated services for Alpha-1 patients – in particular, agreed standards of care and identifiable specialist services with expertise of the condition.
- Lack of multidisciplinary care pathway that integrates lung, liver and transplant services.
- Lack of access to specific licensed treatments, that are available in many other countries, such as augmentation therapy.

## 10. An example of a care model – ADAPT and the UK national Alpha-1 patient registry

The Antitrypsin Deficiency Assessment and Programme for Treatment (ADAPT) and the associated national Alpha-1 registry was established at Queen Elizabeth Hospital Birmingham in 1996 and has been funded predominantly by industry. The demographic and clinical data collected by ADAPT are amalgamated in the national disease registry. Clinical research at ADAPT has contributed significantly to the worldwide knowledge-base and the current understanding of Alpha-1 and associated conditions.

ADAPT is run by a team of expert clinicians, specialist nurses and a research team and offers a comprehensive programme of care to Alpha-1 patients which includes:

- Information on the disease, including the genetic and healthcare implications for patients and their families.
- Comprehensive, state-of-the-art clinical assessment.
- Advice on appropriate lifestyle and self-management to minimise progression of the disease.
- Recommendations on clinical management for implementation by local treating clinicians.
- Annual follow-up and clinical review at the ADAPT centre.

Patients are usually referred to ADAPT by their specialist clinician following a diagnosis of Alpha-1, or they may get in touch with ADAPT via a patient support group. ADAPT provides financial support towards patients' travel costs to the centre.

Many respondents to our survey reported their positive experiences of ADAPT. The comprehensiveness and quality of the service provided at ADAPT were praised, and patients feel that ADAPT offers significantly more support and care to them and their specific needs than the NHS Scotland currently provides.

*"I gained much of my knowledge from ADAPT in Birmingham."*

*"ADAPT is amazing and without them I would feel quite worthless. They keep me informed and I feel looked after."*

*"I have access to experts in Alpha-1 at ADAPT."*

*"My consultants do not have the specialist equipment that ADAPT have in Birmingham and which tests more sensitively and accurately."*

*"ADAPT are exceptional. But it's a long journey every time - 400 plus miles."*

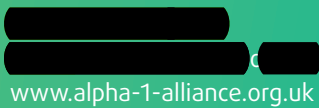
*"All the advice and recommendations for treatment I receive is from the Alpha-1 specialists at ADAPT. My GP and consultant only prescribe what they are told."*







## Alpha-1 Alliance



[www.alpha-1-alliance.org.uk](http://www.alpha-1-alliance.org.uk)

The Alpha-1 Alliance is sponsored by its member patient groups, and formerly, by unrestricted educational grants provided by CSL Behring and Grifols.

The Secretariat to the Alpha-1 Alliance is provided by Advocate.

Industry sponsors have no influence on the decisions and activities of the Alpha-1 Alliance.