Alpha-1 Alliance

Alpha-1 Antitrypsin Deficiency Policy Report

www.alpha-1-alliance.org.uk

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1. Foreword

As an MP with a young constituent who suffers 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 lung 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 England.

It is important not to lose sight of patients affected by rare diseases and to invest in specialised services and effective therapies that are needed by only a small group of citizens. I welcome that the Alpha-1 community has now come together and look forward to working with the Alpha-1 Alliance, NHS England and MPs on the recommendations included in this report. Together we have the opportunity to ensure that the expertise within the NHS is used to best support the specific needs of Alpha-1 patients.

Mark Pawsey MP

Chairman's Statement

As a practising clinician and scientific researcher who has worked for two 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 are affected in many aspects of their lives and acts as a compass to guide the development of care for Alpha-1 patients in England.

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 workers, and recognition by policy-makers, 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 England. The necessary clinical expertise and the will to improve the care of Alpha-1 patients already exist within the NHS but there is currently no care pathway that delivers optimal and integrated care for Alpha-1 patients.

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

Dr Ravi Mahadeva

2. Executive Summary

Alpha-1 antitrypsin deficiency (Alpha-1) is a rare genetic condition that affects both children and adults and can cause severe lung and liver disease. There is considerable variation across the country in patient access to specialists and appropriate therapies, and no care model currently exists within the NHS that provides integrated clinical management for the unique needs of the different clinical aspects of Alpha-1.

The Alpha-1 Alliance, an unprecedented coalition of the Alpha-1 patient community and expert clinicians, commissioned a national survey of Alpha-1 patients, their families and carers to establish the level of unmet medical need that Alpha-1 patients currently experience in England. The report demonstrates that considerable gaps exist in the delivery of integrated healthcare services in the NHS for this complex disease. Based on these findings, our recommendations are for a multi-disciplinary service that will:

1. Raise awareness and knowledge of the condition amongst clinicians and other healthcare professionals.

2. Improve patient information about the disease, its implications and available treatment options.

3. Ensure early and correct diagnosis.

4. Detect high-risk patients.

5. Integrate and coordinate all aspects of clinical care and genetic counselling.

6. Link genetic, respiratory, hepatology, transplantation and paediatric services.

7. Ensure equitable access to all clinical services required for optimal care.

8. Improve access to augmentation therapy and future effective therapies for those patients who will benefit.

9. Provide national benchmarking for optimal standards of integrated care.

10. Deliver all of the above through national Alpha-1 centres of excellence in tertiary hospitals with interdisciplinary medical expertise.

A national highly specialised service that implements these recommendations would facilitate the consistent delivery of integrated multi-disciplinary care. Such a holistic approach for care is widely believed within the clinical community to have considerable long-term benefits for Alpha-1 patients.

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 commonest symptom is shortness of breath, which can significantly limit their ability to work and perform normal daily activities, and 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 England, there is considerable variation across the country in patient access to specialists, and no access to augmentation therapy for Alpha-1-associated lung disease. There is also no care model within the NHS 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 patients, their families and carers. It contributes to a campaign to establish a nationally commissioned highly specialised 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

4. The Alpha-1 Alliance

The Alpha-1 community has established the Alpha-1 Alliance, which is an unprecedented coalition of patient groups and leading clinicians in the field. The objectives of the Alliance are to raise the profile of the unmet medical need of Alpha-1 patients and to campaign for better access to specialist services for these patients, which include integrated care and augmentation therapy, via a nationally commissioned specialised service. The Alpha-1 Alliance includes the Alpha-1 UK Support Group, Alpha-1 Awareness UK and Alpha-1 Advocacy & Action and is chaired by Dr Ravi Mahadeva, who represents other leading specialists.

5. Purpose of the report

- To identify the areas of unmet medical need for Alpha-1 patients.
- To evaluate differences in access and quality of available medical care for Alpha-1 patients across England.
- To establish the impact of the unmet medical need on relatives and carers of Alpha-1 sufferers.
- To improve parliamentarians' and policy-makers' understanding of the needs of the Alpha-1 community, including those of their 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 that are available to the Alpha-1 community.

6. Methodology

The survey was conducted online amongst English 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.

7. Demographics

162 respondents from all parts of England returned completed surveys; 93 responses were submitted by patients and 69 by family members or carers. The analysis that informs this report includes all responses. The vast majority of patient-respondents reported to suffer from the most severe form of Alpha-1, genotype PiZZ. Most respondents reported to experience predominantly lung-related symptoms, although 18% of respondents reported liver problems.

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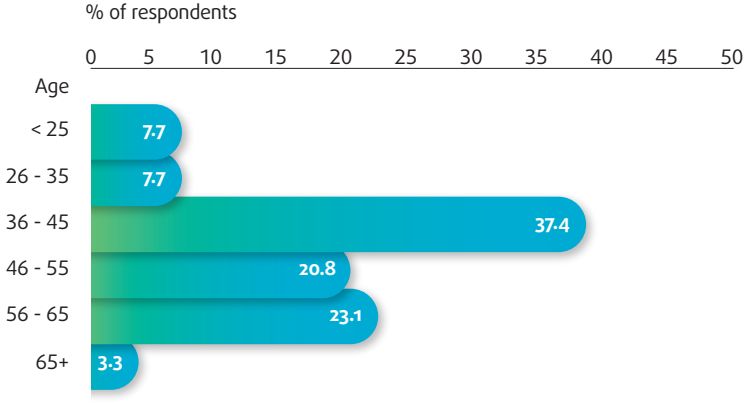
8. Survey results

8.1 Diagnosis

Age at diagnosis

The majority of respondents were diagnosed with Alpha-1 between the ages of 36 and 65, with 37% having received the diagnosis between 36 and 45 years of age.

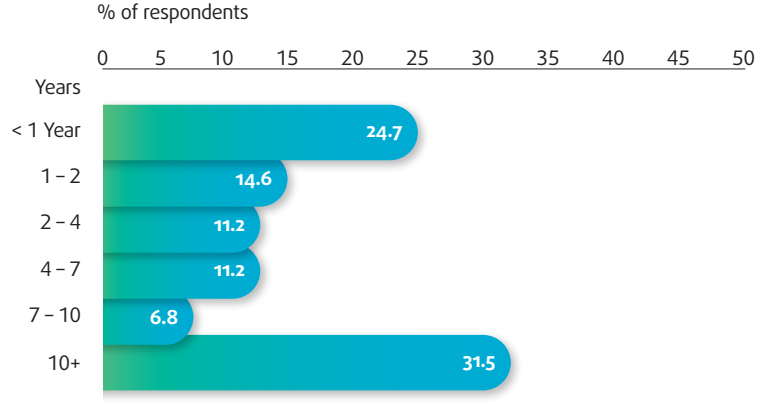
How old were you when you were first diagnosed with Alpha-1?



Delayed diagnosis

Nearly a third of patients experienced a delay of more than 10 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?



> "My symptoms appeared shortly after birth, and I was diagnosed at age 64.

Misdiagnosis

Many respondents reported that they were initially misdiagnosed and received treatment for other respiratory conditions, the commonest being asthma, before being correctly diagnosed.

"First diagnosed with asthma aged 40 but after no signs of improvement after several years of treatment a new diagnosis of COPD was given. At age 65 I was finally sent to see a respiratory specialist who did a blood test which indicated I was an Alpha."

"Years of unexplained severe long lasting chest infections treated as asthma."

"I guess it was over a ten year period that I was treated for bronchitis. I was reviewed on a yearly basis and, although I was getting shorter of breath, no more tests were done."

Many respondents reported that the long journey that they encountered through the healthcare system without receiving the correct diagnosis had 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.

"It was like going through a tunnel with no light at the end."

"I felt completely lost and deserted by the NHS."

"I had a 'mystery illness' in 1994 for which I was hospitalized for 2 - 3 weeks and although every test and scan was carried out, there was no diagnosis made."

"I saw 10 or 15 doctors before receiving a diagnosis. I had to ask to have the test done."

"Having been diagnosed with asthma 27 years ago I've lost count of how many doctors it has taken before I was referred to a lung specialist."

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.

"Because my brother was diagnosed with Alpha-1, I was tested which I think was a major step in saving my health and changing my lifestyle before it was too late."

"My health has been reasonably stable mainly as a result of early diagnosis."

"If I had been diagnosed earlier, I would have avoided some of the activities which have exacerbated my condition."

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.

"It's a steady decrease of quality of life."

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.

"My liver and lungs are affected, and my physical stamina has gone. Things I enjoyed doing are now history for me."

"When you can't breathe properly, life changes."

"I get severely breathless on exertion, walking up hills and carrying bags, and I feel tired most of the time."

"I am almost housebound relying on my mobility scooter to get me out & about."

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.

"I am 39 and barely able to dress myself! It has taken only five years to get to this stage."

"I can do very few 'everyday' tasks if they require any moderate exertion."

"Walking, climbing stairs, doing housework became virtually impossible."

"I have to pace myself in getting dressed or bathing."

Work and social life

Respondents were asked to detail how their health problems reduce 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 in a fulfilling personal life, with a profoundly deleterious effect on their quality of life.

"I had to give up work because of chronic lung problems and chest infections."

"I had to retire early as I was unable to fulfil my work commitments due to my breathing."

"My social life suffers, at work I struggle in many ways, sometimes I feel isolated from friends."

"I cannot make any arrangements to visit family and friends as I am always suffering from chest infections."

"My circle of friends is now very small and I have lost all my old friends due to me being mostly housebound."

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 the time they are trying to bring up a family.

"I struggle to stay at home and look after the kids whilst my wife singlehandedly supports the family."

"It's heartbreaking having your family worry about you, becoming a nuisance to them, seeing the fear in their eyes when you are poorly."

"It is very difficult for my husband who is trapped in my same world."

"I am pretty much houseband these days and need oxygen 24/7. I have been struggling to breathe now for over 20 years and some days it all seems a bit too much. I can no longer drive my car and I have to rely on my son to take me anywhere."

"I have found it hard to get others to do so many of things for me that I routinely did just a year or two ago."

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.

"I have severe bouts of depression."

"I find it hard to deal with psychologically at times."

"Mentally it's a challenge as I have two young children that I wish to see grow up."

"When I realized there is no effective treatment for Alpha-1 in this country, I became more and more depressed."

"I don't know what the future holds for me I'm too scared to look."

8.3 Impact on patients' family 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.

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"My husband's condition has changed my lifestyle – loss of independence, loss of income, holidays are difficult as he can't cope with heat, cold or hills."

"I have to care for her full-time and am not able to return to work."

"There aren't many things we can still do together."

"I can't keep a job as I had to keep taking time off to look after my daughter, I have to be her nurse as well as her mum."

"It means having to take a lot of time off work for hospital appointments, sickness etc."

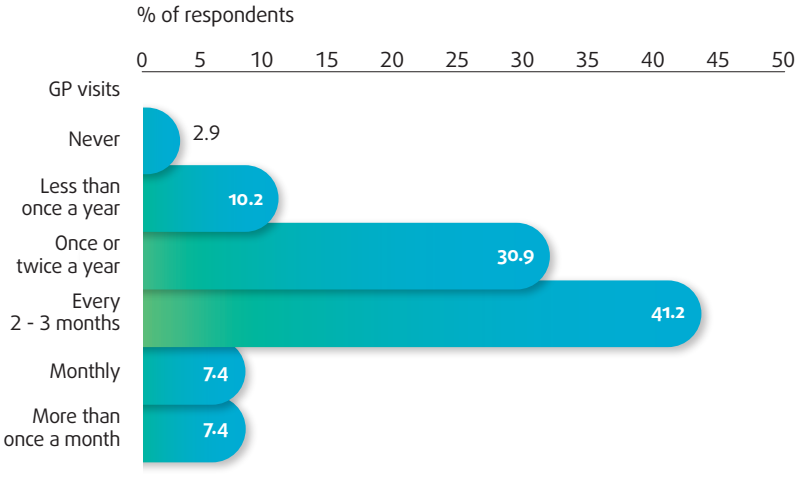
"The constant worry about my children's health and welfare is stressful and not good for my health either!"

8.4 Access to medical services and treatment

Primary care

44% of respondents reported having to visit their GP more than once every three months to receive help for their condition, indicating a high unmet medical need.

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

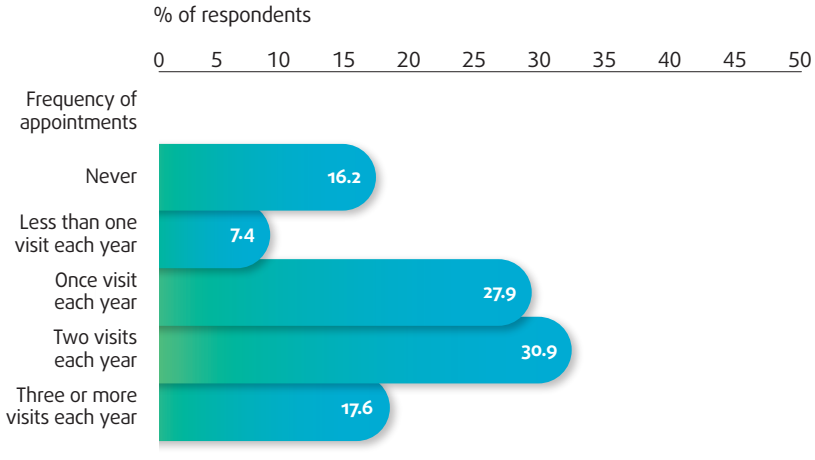


> "Not always easy to get an emergency GP appointment."

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 43% of respondents said that they had seen a specialist respiratory nurse for their condition. Some respondents commented on encountering barriers to easy access to doctors leading to potentially harmful delays in obtaining treatment.

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



Accident & Emergency attendances

39% 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. A third of respondents were admitted to hospital for their condition in the preceding five years.

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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. Many respondents reported being dependent on long-term supplementary oxygen therapy, access to which varied.

"The only treatment I receive are my prescribed inhalers."

"I am on oxygen 16 hours a day."

"I have problems with the oxygen provider."

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. Several patients highlighted the cost of prescription charges when multiple medications were needed.

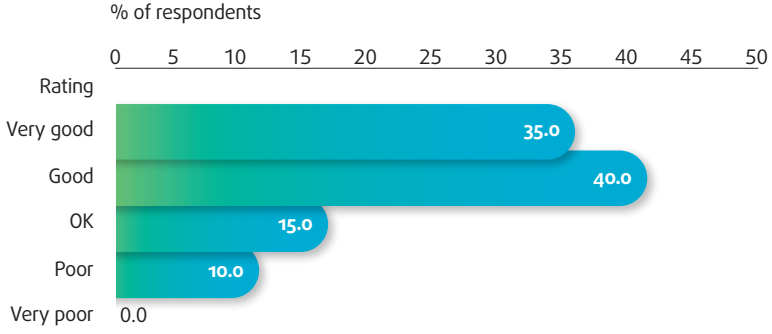
"Sometimes it is difficult to get a supply of steroids at short notice. Doctor expects me to turn up at the surgery when I least can go out."

"Access to this treatment is not easy as I now have to get 4 prescriptions a month which I cannot afford."

Genetic counselling

The survey showed that genetic counselling was rarely proactively offered by NHS doctors to patients following a diagnosis of Alpha-1. Many respondents reported that genetic counselling was only provided after it had been explicitly requested by them. Only 26% of respondents had received any genetic counselling. Of those, three-quarters rated their overall experience of genetic counselling as 'very good or good'.

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



"I had to ask on many occasions before I was referred for genetic counselling. The service was very good once I got there.

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

"It improved my understanding of the condition."

"It made my wife realise how important it was to get herself tested for the sake of our children."

"We built a family tree and discussed the implications on having children."

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Pulmonary rehabilitation

43% of respondents said that they had seen a specialist respiratory nurse for their condition at least once, but only 54% of patients 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 the general info and education provided in the course via lectures and videos to be excellent."

"It was beneficial but I was only given one course five years ago and not been offered any since."

"There is a 9 month waiting list to get pulmonary rehab in my area."

"It took 3 years to get a place."

"I cannot get pulmonary rehab because I live too far from nearest clinic."

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 England.

"I was lucky enough to have a life-changing double lung transplant just over a year ago. Life is so, so much better now."

Regional inequalities in access to appropriate healthcare

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

"There is no treatment for people like me locally."

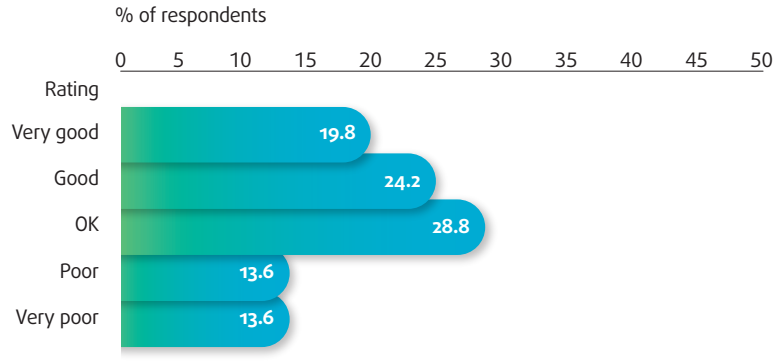
"Good support from local respiratory team if required, but only Monday to Friday."

"It seems to be a postcode lottery - many Alphas live near Birmingham (ADAPT) or Cambridge - for me there is nothing."

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 44% and as 'poor or very poor' by 27% of respondents. This indicates that there are no uniform care standards for the treatment of Alpha-1 in England.
The lack of awareness of the disease and the available options for treatment were criticised by many of the respondents.

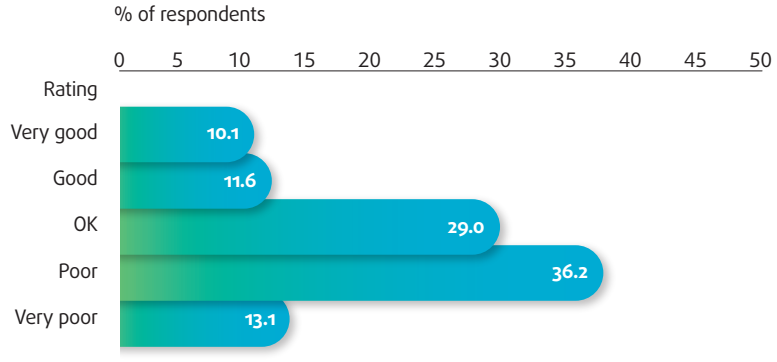
How would you rate the quality of the treatment and support that you currently receive?



Awareness and knowledge amongst healthcare professionals

The majority of respondents highlighted the lack of medical knowledge about the disease amongst GPs. Only 22% rated their GP's level of knowledge about the condition as 'good or very good', whereas almost half 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.

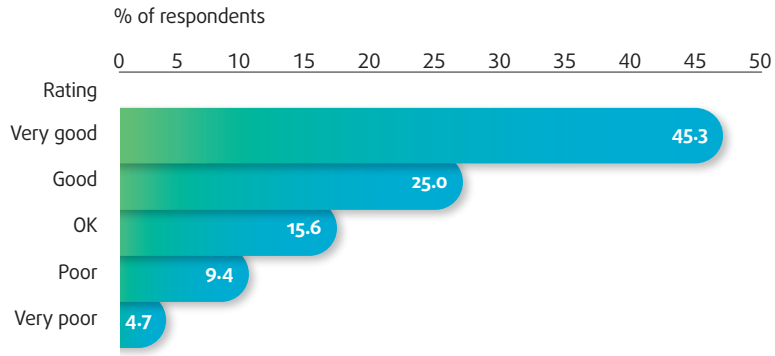
How would you rate your GP's level of knowledge of Alpha-1 and its effects on your health?



> "My GP was not familiar with Alpha-1.
>
> I was only diagnosed by accident by a locum GP who was concerned at the number of chest infections I was getting.

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

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



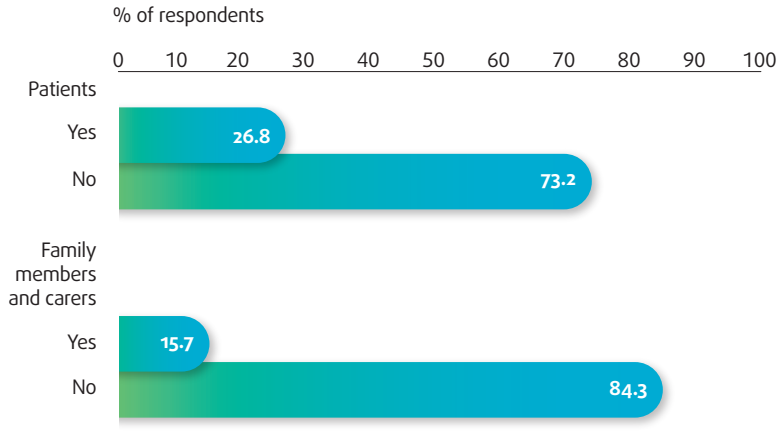
"My consultant has in depth knowledge.

The consultant is very knowledgeable about Alpha-1, but her registrars are not.

Information provided by the NHS

Respondents were generally not satisfied with the level of information they receive for their condition, with 73% of patients and 84% of family members and carers feeling that the NHS does not provide sufficient information about available 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 to look on the internet.

I had no information given to me by any medical professional. All my knowledge of the disorder is from online research and leaflets.

Medical treatment, care and support

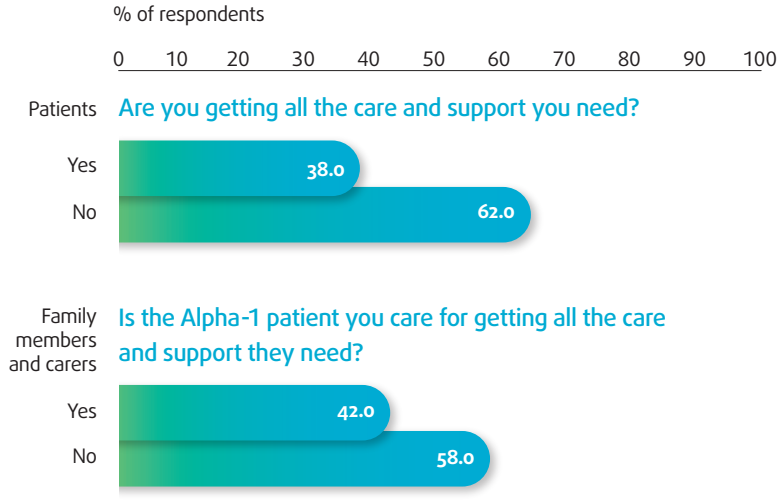
Respondents expressed their concern about the lack of adequate treatment and care options for their disease that exist in England.

"There is no treatment in England, so feeling ill is the norm."

"I don't feel that doctors consider new treatments, just rely on the standard ones available."

"The treatment I get for my Alpha-1 is non-existent."

Overall, 62% 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 inadequate support structure in the NHS for their unique and complex needs.



"I feel let down by the NHS. The GP offers very little support.

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 the NHS 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 the NHS on the disease, its implications for patients and their families, on the location and availability of specialist clinicians with expertise in Alpha-1 and on 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 made available in England, they did not receive the same level of attention and care as other rare respiratory conditions, such as cystic fibrosis.

"I would like to know which specialists and hospitals have experience and skill in dealing with Alpha-1. I just wish they all knew a bit more about it and its consequences."

"There needs to be more awareness and better education and training amongst health and care professionals."

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

"I would like GP's to be much better informed and more proactive."

"It would be great if there were more locally accessible specialist centres who knew about Alpha-1."

"Information on any transplants and how to get on the transplant lists."

"More support at the time of diagnosis and information that's easy to understand."

Medical services and treatment options

Respondents were asked what services and treatments for Alpha-1 sufferers they feel the NHS 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 such as augmentation therapy, that are available in many other countries. The need for a multi-disciplinary approach and better services for affected children was also highlighted.

"We need a uniform approach and not a postcode lottery."

"We need access to augmentation therapy and other new treatment options."

"A minimum yearly check on the severely affected, to include lung and liver checks."

"It's not all about the lungs."

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"National awareness programme and more services locally for children diagnosed with Alpha-1."

"Anything that might come under the heading 'Best Practice'."

"Access to specialist counselling not only for sufferers but for family members also."

"Treatment should be more in line with other countries."

"Screening for carriers and their partners who are planning a family."

Support for a national highly specialised service

All respondents to our survey were unanimous in their strong support for the establishment of a national highly specialised service for Alpha-1. 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 multi-disciplinary management of their condition and grant more continuity.

"The benefit of a national service would be better access to both treatment and related services."

"We need treatment and expert treatment centres."

"It would be helpful to monitor changes in patients due to Alpha-1."

"I am amazed that the NHS and its professionals have not set up a national service already."

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 England. 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 that result 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 (e.g. GP and consultant appointments, genetic counselling, respiratory rehabilitation)
- the need for coordinated services for Alpha-1 patients, in particular, agreed standards of care and identifiable specialist services with expertise in the condition
- inequitable and restricted access to adequate therapies such as supplementary oxygen
- lack of multi-disciplinary care pathway that integrates lung, liver and transplant services
- lack of access to treatments, such as augmentation therapy, 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) programme and the associated national Alpha-1 registry were 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 of 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 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 specialists do not have the specialist equipment like ADAPT in Birmingham does 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 Specialist, my GP and consultant only prescribe what they are told."

11. Recommendations

Alpha-1 is a severe and complex disease and affected patients have a variety of unique medical needs. The existing gaps in the care for Alpha-1 patients in England can be addressed most effectively by a national highly specialised service within the NHS that ensures optimal integrated care for patients. Based on the results of the national patient survey, our recommendations for the remit of such a multi-disciplinary service should include:

1. Raising awareness and knowledge of the condition amongst clinicians and other healthcare professionals.

2. Improving patient information about the disease, its implications and available treatment options.

3. Ensuring early and correct diagnosis.

4. Detecting high-risk patients.

5. Integrating and coordinating all aspects of clinical care and genetic counselling.

6. Linking genetic, respiratory, hepatology, transplantation and paediatric services.

7. Ensuring equitable access to all clinical services required for optimal care.

8. Improving access to augmentation therapy and future effective therapies for those patients who will benefit.

9. Providing national benchmarking for optimal standards of integrated care.

10. Delivery of all of the above through national Alpha-1 centres of excellence in tertiary hospitals with interdisciplinary medical expertise.

A national highly specialised service that implements these recommendations would facilitate the consistent delivery of integrated multi-disciplinary care. Such a holistic approach for care is widely believed within the clinical community to have considerable long-term benefits for Alpha-1 patients.

Alpha-1 Alliance

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www.alpha-1-alliance.org.uk

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