

# MISUNDERSTOOD, MISTREATED AND FIGHTING TO BE HEARD

The devastating journey of achalasia  
diagnosis in the UK



**ACHALASIA ACTION**

This report was prepared by Achalasia Action and Common Purpose Collective. We are grateful for the contributions of:

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Special thanks to the rest of the Board of Trustees of Achalasia Action, the steering group of volunteers and all participants who made this research possible.

#### Achalasia Action

In January 2020, Achalasia Action was officially registered as a Charitable Incorporated Organisation with the Charity Commission for England and Wales (Charity no. 1187367).

Achalasia Action's charitable objectives include:

- To advance education about achalasia and associated conditions.
- To preserve and protect the health of people with achalasia, including their friends and families.
- To encourage and support research into achalasia.

To achieve these objectives, our activities include:

- **Support:** providing a friendly and well-informed community, education and guidance to support those with achalasia and their loved ones.
- **Inform:** raising awareness of the disease amongst patients, their families, the medical profession, policymakers and the wider public.
- **Research:** collaborating with the medical community and encouraging achalasia-related research.

For more information about support group meetings, subscribing to our newsletters, or discussing your personal situation, please visit [www.achalasia-action.org](http://www.achalasia-action.org) or call our helpline at 0300 772 7795.

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# 01

## Foreword

Receiving a diagnosis of achalasia is rarely straightforward. Our own personal experiences underscore just how challenging and isolating the process can be. Both of us experienced difficulties in misdiagnosis, with our achalasia symptoms originally mistaken for other issues, such as anxiety – a misdiagnosis that we know, thanks to this research, is particularly common among women with achalasia.

The findings in this report clearly demonstrate that a swift correct diagnosis is not typical. Misdiagnosis is, regrettably, the norm rather than the exception. In addition, multiple misdiagnoses before finally receiving an accurate diagnosis of achalasia are common.

Achalasia is also inherently isolating, as well as damaging to someone's physical and mental health. As we can attest from our own experience of getting a correct diagnosis, persistent symptoms – pain, difficulties in daily life, and the narrowing of one's social world – are challenging enough without the added pressure of continuous self-advocacy. People with achalasia regularly encounter misunderstanding from friends, family, employers and medical professionals alike, perpetuating a cycle of doubt and isolation.

This report represents the culmination of months of work, led by the achalasia community. Achalasia Action would like to offer our heartfelt thanks to the participants who took part in our consultation and interviews and the volunteers who steered this research. Their willingness to share often painful experiences has given this report its depth and credibility, and their collective voice reminds every reader that no one faces achalasia alone.

The result is a clear and unignorable illustration of a stark truth: the burden of achalasia misdiagnosis and late diagnosis is heavy and widespread, and our current healthcare system, despite the commendable efforts of countless dedicated professionals within the NHS, is simply not meeting the needs of people with this rare disease.



Our own response to these findings is one of anger and disappointment, but not surprise. For those with achalasia, all the experiences detailed in this report will be familiar. What this report shows is that the problems of misdiagnosis are systemic, not individual; widespread, not isolated. This is not an individual struggle; it is our collective fight for recognition, understanding, and better care. Simply put, what we have now is not good enough.

It is time for change. Our healthcare system can and must do better. This report must serve as a catalyst for systemic improvement in diagnosing and managing achalasia. That journey starts now.



**Silvia Davey**  
Chair of the Board of Trustees,  
*Achalasia Action*



**Jack Eddy**  
Board of Trustees and  
Head of Policy and Advocacy,  
*Achalasia Action*

# 02

## Executive Summary



This comprehensive report illuminates the severe challenges and systemic shortcomings faced by individuals living with achalasia in the United Kingdom (UK). Based on the experiences of 350 participants - representing around 5% of the total UK achalasia patient population - the research uncovers a troubling landscape characterised by persistent delays, frequent misdiagnoses, and a healthcare system inadequately prepared to manage this rare condition.

Key findings from our research indicate alarming patterns. Over half of the respondents endured multiple incorrect diagnoses before receiving an accurate diagnosis of achalasia. Commonly mistaken conditions included acid reflux, anxiety, and eating disorders. More than a quarter of participants waited longer than three years for an accurate diagnosis, with some extreme cases stretching over a decade. Women, in particular, faced disproportionate delays and misdiagnoses, often due to gender biases resulting in physical symptoms being erroneously attributed to psychological factors.

The diagnostic journey was notably fragmented, involving interactions with numerous healthcare professionals - ranging from general practitioners to specialists - many of whom lacked sufficient awareness or understanding of achalasia. This fragmented pathway not only delayed diagnosis but also significantly impacted patient trust and satisfaction of the healthcare system, exacerbating feelings of isolation and medical dismissal. Patients frequently noted being misunderstood or dismissed by healthcare providers, intensifying their frustration and sense of helplessness.

The physical consequences of delayed diagnosis and misdiagnosis were profound. Participants detailed substantial deterioration in health, including rapid weight loss, malnutrition, chronic pain, daily symptoms such as severe chest spasms and persistent swallowing difficulties. The psychological repercussions were equally severe, with high rates of anxiety, depression, distress, and significantly reduced quality of life. Participants also noted considerable disruption to personal relationships, social isolation, and impaired career progression. Financial strain was another critical issue, as many patients resorted to private healthcare due to excessive NHS waiting times, highlighting inequities in access to timely medical care.

This research identifies profound systemic healthcare failures. Many patients reported inconsistent and ineffective communication between healthcare professionals, inadequate patient education about achalasia post-diagnosis, and limited aftercare support. These gaps undermined patient confidence in the healthcare system, contributing to long-term reluctance in seeking further medical assistance.

This report is a critical call to action, outlining systemic changes in the NHS that require concerted action across the system. Our key recommendations include:

- **Enhancing healthcare professional education and awareness on achalasia**  
Bodies with a significant role in setting national training curricula for primary care and gastroenterology should ensure that training emphasises early recognition of key achalasia symptoms and reduction of gender bias in symptom interpretation and clinical assessments, to improve early recognition and appropriate referral for achalasia.
- **Improving achalasia diagnostic pathways**  
Publish a nationally agreed diagnostic algorithm that sets out the earliest appropriate referral point from primary care for persistent upper GI symptoms, irrespective of age, weight loss or gender. NHS capacity should also be expanded for oesophageal manometry (e.g. additional tertiary centre sessions and cross trust booking). Decision support prompts should also be added in GP software to lower the threshold for urgent referral, rather than expecting GPs to request manometry or barium studies themselves.
- **Improving patient communication and support**  
Develop robust communication strategies and structured aftercare plans to support patients post-diagnosis, including clear informational resources and accessible psychological support services.
- **Renewing the policy agenda for rare diseases**  
Renew the UK Rare Diseases Framework and the reinstatement of the Rare Disease Advisory Group (RDAG), with a focus on improving diagnostic delay for conditions that do not have a clear genetic link.

We call upon healthcare providers and purchasers, policymakers, professional membership bodies, and government stakeholders to urgently prioritise and implement these recommendations. Addressing these critical gaps will ensure that future achalasia patients receive timely, accurate diagnoses and comprehensive, compassionate care, significantly reducing unnecessary suffering and improving overall patient outcomes.

As the only charity solely focused on supporting people with achalasia, Achalasia Action will focus on advocating with key stakeholders to achieve the above recommendations. As a patient support charity, we will consider the following recommendations that people with achalasia steering this project have asked for, to inform future Achalasia Action strategies and activities:

- Gather and share knowledge about achalasia symptoms via case studies and a diagnostic check-list for primary care professionals.
- Further develop educational and support resources for people with achalasia, including an achalasia business card, a workplace adjustments card, guidance on how to navigate social situations and resources specifically supporting women.
- Establish supportive achalasia communities for people with a new achalasia diagnosis, including peer-led support services and buddy systems for newly diagnosed patients, creating positive and non-intimidating spaces for people when they are first diagnosed, and facilitating regional achalasia support groups.
- Provide self-advocacy support for people with suspected achalasia via services such as peer-to-peer support programmes, webinars, or buddy systems and resources including templates for symptom diaries and questions to ask during appointments.
- Expand equity, diversity and inclusion in achalasia support service, by developing and co-producing targeted outreach strategies and culturally-sensitive achalasia resources in multiple languages that address the specific needs and concerns of different communities.

# 03

## Introduction



## Introduction

Every day in the UK, someone with achalasia is struggling to swallow their food and experiencing extreme pain at mealtimes, whilst facing a long and uncertain journey to understanding what is causing their symptoms. For the estimated 6,000 people living with achalasia in Britain, the journey to a correct diagnosis is often marked by years of confusion, misdiagnosis, and mounting health complications that could have been prevented by earlier diagnosis and intervention.

The human impact of delayed diagnosis and misdiagnosis reaches far beyond the physical symptoms. We see careers disrupted, social lives diminished, and families struggling to support loved ones through years of uncertainty. The psychological toll of living with unexplained symptoms, combined with the frustration of multiple misdiagnoses, creates a burden that no one should have to bear.

Between December 2024 and March 2025, Achalasia Action undertook service user-led research to understand these challenges. Our community opened their lives to us, sharing deeply personal stories of their diagnostic journeys. Their experiences revealed not just individual struggles, but systemic failures in how achalasia is recognised and diagnosed within our healthcare system. We are profoundly grateful to all those who contributed their time and energy to this research, often revisiting difficult memories to help create change for others.

This report represents more than just research findings: it is a call to action. By documenting people's experiences and identifying clear opportunities for improvement, we aim to catalyse meaningful change in how achalasia is recognised, diagnosed, and how people experiencing life-changing symptoms are treated. The recommendations we present are practical, achievable, and have the potential to transform lives.

## The impact of achalasia on people's lives

Achalasia is a rare swallowing condition that affects the muscles of the oesophagus, the food pipe that runs from the throat, down through the chest and into the stomach. In healthy individuals, coordinated muscle contractions in the oesophagus move food downward, while a ring of muscle at the bottom of the oesophagus (the lower oesophageal sphincter) relaxes and opens to let food enter the stomach. In people with achalasia, this essential mechanism fails.

The condition occurs when oesophageal muscles are weak, don't contract properly, or don't function at all, creating two critical problems: the oesophagus loses its ability to propel food effectively, and the lower

oesophageal sphincter fails to open. Food and drink are blocked in the oesophagus and can remain there for a long time, sometimes for hours or days. This food build-up can create pressure, similar to a blocked hosepipe, until the food either painfully goes down into the stomach or is regurgitated back up through the mouth.

Because of this, people with achalasia experience a range of symptoms such as, but not limited to, difficult and painful swallowing, food regurgitation, malnourishment, rapid weight loss, lack of energy, regular acid reflux and extremely painful chest spasms, which often evolve and worsen over time. The condition can have a severe and multifaceted impact on a person, affecting not only their physical health but also their social life, mental well-being, and the ability to work or carry out everyday activities.

## A hidden crisis

It is estimated that around 6,000 people are living with achalasia in the UK. However, these numbers may underrepresent the true scale of the problem. The condition's rarity means that healthcare professionals have often never encountered achalasia, making recognition, diagnosis and treatment exceptionally challenging.

Although achalasia has no cure, there are a limited number of treatments that can help improve achalasia symptoms, most of which are surgical. It is critical that people with achalasia receive a correct and fast diagnosis, as it is the first necessary step towards access to treatments that can help ameliorate their lives.

# 04

## Methodology

### **A service user-led approach**

As a charity led by people with achalasia, it was important that Achalasia Action took a research approach that centred around people living with the condition. This research has been conducted as a partnership between Achalasia Action, a research consultant from Common Purpose Collective (a co-production research consultancy) and those who use Achalasia Action's services.

We recruited a steering group of seven volunteers living with achalasia from across England and Wales. The steering group members brought varied diagnostic experiences, represented different age groups and came from a range of geographical locations. The steering group co-produced the research methodology, co-facilitated the focus groups, collaborated to validate the data analysis, and helped develop the recommendations you will find in this report. This research could not have happened without their hard work, expertise and willingness to share their lived experience so openly.





**Annie-Rose Tate**  
North Wales  
Female  
9 years living with achalasia

**Amanada Ladell**  
London  
Female  
30 years living with achalasia

**Linda Arundale**  
West Yorkshire  
Female  
10+ years living with achalasia



**Winette Deflorimonte**  
Croydon  
Female  
15 years living with achalasia

**Pamela Browne**  
Ingatestone, Essex  
Female  
30+ years living with achalasia

**Fred Caygill**  
Exmouth, Devon  
Male  
Diagnosed in 2023

**Emma Wabnij**  
Durham  
Female  
Diagnosed in 2024

## Quantitative research

Through a facilitated session, our steering group designed a national survey that was distributed via Achalasia Action’s social media channels and newsletter. The survey gathered responses from 350 individuals living with achalasia from the UK, equating to just under 6% of the estimated population affected by achalasia in the UK. All participants provided informed consent before completing the survey, and all data was anonymised to protect participant privacy. The questions in our survey focused on collecting factual data, such as demographic information, misdiagnosis timelines and quantifiable measures of the impact of a late diagnosis or misdiagnosis. This has allowed us to identify clear patterns in people’s diagnostic journeys.

It should be noted that some percentages reported in our findings do not add to 100% as respondents often provided multiple answers for one question.

## Interviews and focus groups

We conducted 12 one-to-one interviews and 6 focus groups with 20 people to explore in more depth the impacts of prolonged diagnosis and misdiagnosis of achalasia. Overall, we spoke to 32 people living with achalasia from across the UK, all of whom revealed different experiences of being diagnosed with achalasia. The focus groups were co-delivered by the research consultant and a member of the steering group.

Participants provided informed consent prior to participating in interviews and focus groups, and all personal identifying information was anonymised in the analysis and reporting process. We have changed the names of all participants in this report to ensure anonymity. Participants were asked a series of questions about their diagnosis journey, final diagnosis experiences and the impact of getting a diagnosis. We also asked participants to detail any key recommendations they would want to see in the future to improve the diagnosis pathway.

All interviews and focus groups were recorded and transcribed using an online transcribing tool. The research consultant then undertook a process of thematic analysis by grouping common trends and themes. Our steering group then reviewed the preliminary themes during two collaborative workshops to validate the trends. This approach ensured themes accurately represented participant experiences while identifying any gaps.

## Recall bias

While gathering retrospective accounts of diagnosis experiences, we acknowledge the potential for recall bias in both the quantitative and qualitative data, whereby participants’ memories of past events may be influenced by subsequent experiences, or time elapsed since diagnosis. It is important to recognise that the findings in this report are based on personal testimony, and we did not have access to personal medical records or healthcare datasets to validate our findings.

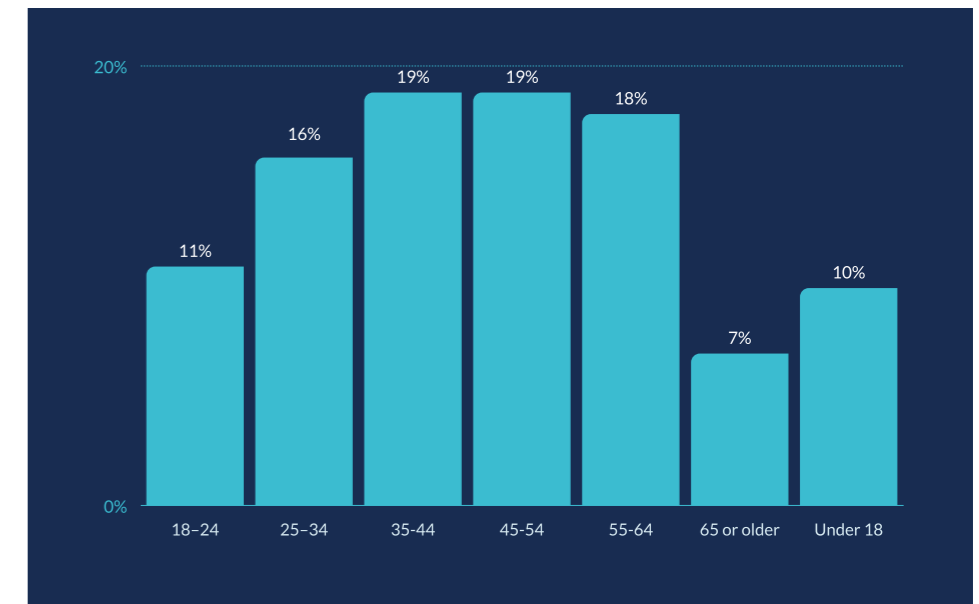
# 05

## Who took part in our research?

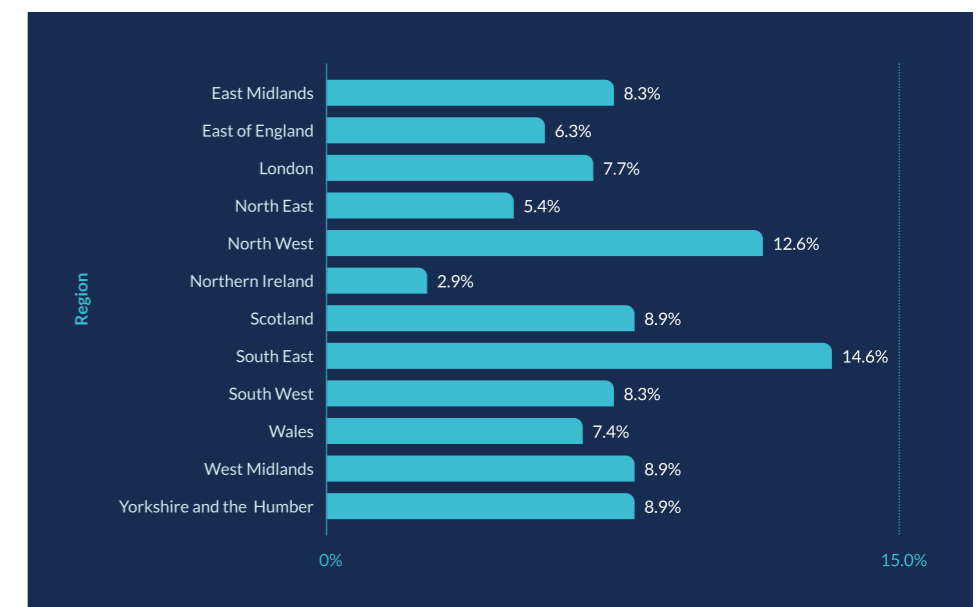
Our findings present data from a national survey of 350 participants, equating to just under 6% of the estimated population affected by achalasia in the UK.

Our survey included participants from all major regions across the UK, ensuring our insights reflect nationwide experiences. There was also representation from people diagnosed across all age groups. This indicates that the patterns and themes explored throughout this report likely cut across regional boundaries and age demographics, suggesting systemic rather than isolated issues:

At what age were you diagnosed with achalasia?



Percentage of participants from each region



As we utilised a convenience sample for this research, it is important to acknowledge limitations in the demographic representation of our sample, particularly regarding gender and ethnicity.

Our participant sample shows a notable gender imbalance with 72% of respondents being women and 28% of respondents being men. This disparity is significant as current medical research does not indicate that achalasia disproportionately affects women. The overrepresentation of women could be attributed to several factors, for example:

- Women may be more willing to participate in health research and share their experiences.
- Women may be more likely to access support through Achalasia Action's online forums where the research opportunity was advertised. For example, we know that on one of our social media platforms (Instagram), 82.5% of followers are women, though we don't have similar demographic data for our newsletter and Facebook audiences, where the research opportunity was also advertised.
- Men with achalasia may be less willing to share their experiences.

This gender imbalance means our findings may disproportionately reflect women's experiences of achalasia diagnosis. A woman's experience could differ from men's in important ways, such as how symptoms are presented to, or interpreted and addressed by, healthcare professionals.

Similarly, our survey sample shows limitations in ethnic diversity, with 95% of the cohort being White British. This figure significantly underrepresents global majority communities compared to the UK population, where 18% identify as non-white<sup>1</sup>.

While these demographic limitations do not invalidate our findings, we acknowledge that our research may not depict the various ways different demographic groups experience getting an achalasia diagnosis. Our findings also do not fully capture intersectional experiences - where structural inequalities based on gender, ethnicity, socioeconomic status, and other factors may create additional obstacles to timely and accurate diagnosis for people with multiple marginalised identities<sup>2</sup>.

Despite these demographic limitations, our mixed-methods approach provides clear and unique insights into the achalasia diagnostic experience across the UK. The consistent patterns emerging across our quantitative and qualitative data suggest systemic issues that are deep rooted in the UK's healthcare system.

In addition to our 350 UK respondents, we also received 112 responses from people living with achalasia outside the UK. While we deeply appreciate these international contributions and acknowledge the global nature of achalasia challenges, as a UK-based charity focusing on national healthcare improvements, this report primarily analyses and presents findings from our UK respondents. The international perspectives, however, continue to inform our broader understanding of achalasia experiences worldwide and reinforce the universal need for improved diagnosis and care pathways.

1. GOV.UK. (n.d.). Ethnicity facts and figures: Government data about the UK's different ethnic groups.

2. Jefferies, D., Williams, E., Buck, D., Babalola, G., & Maguire, D. (2025, September 3). What are health inequalities? The King's Fund.

# 06

## Prolonged diagnosis and misdiagnosis

### The experience of delayed diagnosis

3. Genetic Alliance UK (2020). RARE EXPERIENCE 2020. Rare-Experience-2020-Report-updated-May-2021-.pdf

### Diagnosis by time

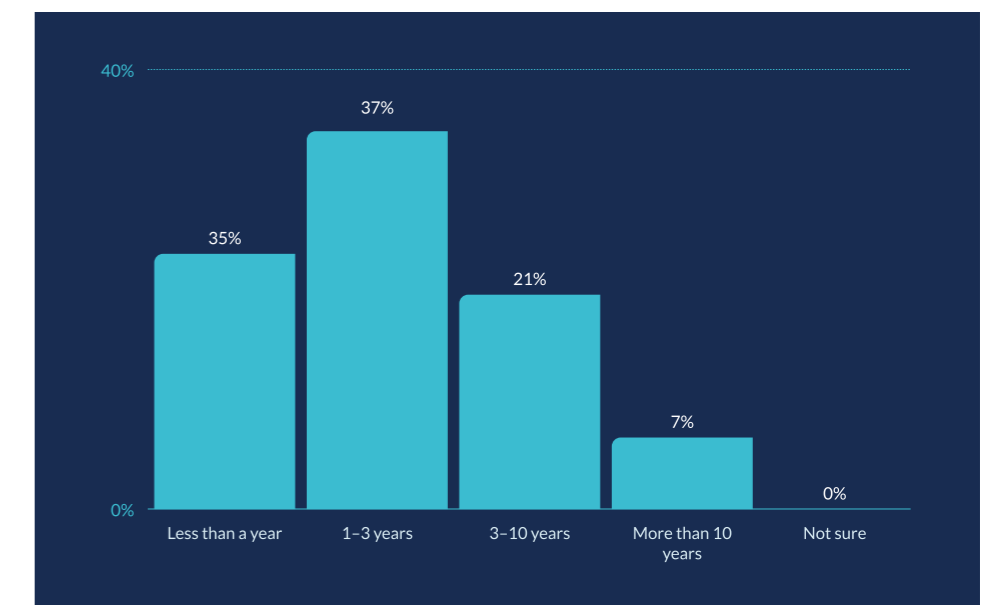
Our research reveals the challenging journey people face to get an achalasia diagnosis. For many, it causes years of uncertainty, endless healthcare appointments and a significant physical and emotional toll.

In this chapter, we explore the extent of the crisis people waiting for an achalasia diagnosis are experiencing. From excruciatingly long waits to multiple misdiagnoses, we hear from people whose lives have been severely impacted in a variety of ways.

Our survey revealed interesting insights into diagnostic timelines, exposing the significant delays that many people face.

For example, 28% of survey respondents endured waits exceeding three years before receiving a correct diagnosis.

More than one-third of respondents (35%) received an accurate diagnosis within one year of seeking medical support, demonstrating that timely diagnosis is possible when the right diagnostic pathways are followed. Among other respondents, more than a third (37%) waited between one and three years for a diagnosis. While this indicates significant room for improvement, it also suggests that the majority of people with achalasia (72%) are diagnosed within three years. This is comparatively better than some other rare disease diagnostic journeys, as the Genetic Alliance UK's Rare Experience 2020 report<sup>3</sup> indicates that 35% of people with rare diseases face waits of 5+ years for a diagnosis.



During our interviews, we heard some cases that were on the extreme end of this spectrum. One person we spoke to waited an extraordinary 17 years for a diagnosis; Anne first experienced achalasia symptoms at age 13 but wasn't diagnosed until she was 30.

**Anne:** *“As I got older and into my 20s it was very, very stressful. These ‘attacks’ were quite frequent because I didn’t realise what I was dealing with and I didn’t know how to deal with it.”*

The prolonged wait for diagnosis severely affected her personal relationships and mental wellbeing.

**Anne:** *“It impacted on relationships as well because not a lot of people know or understand what it is. Even in my 20s, trying to have a relationship... you could try and explain to somebody what’s going on when you’re having a spasm. They don’t really understand.”*

Anne’s story represents the extreme end of what our research shows is a widespread problem of wait times for an achalasia diagnosis. Her almost two decades of unexplained symptoms, medical dismissal, and deteriorating health illustrate the severe consequences of diagnostic delays that many face.

## The cycle of misdiagnosis

Our research uncovered a concerning pattern of multiple misdiagnoses, forcing people into a healthcare labyrinth before receiving the correct diagnosis. The journey to correct diagnosis proves exceptionally challenging, with only 19% of respondents receiving an accurate diagnosis the first time they are diagnosed. The majority (52%) received between one and three misdiagnoses, while 30% suffered through more than four incorrect diagnoses before receiving a conclusion.

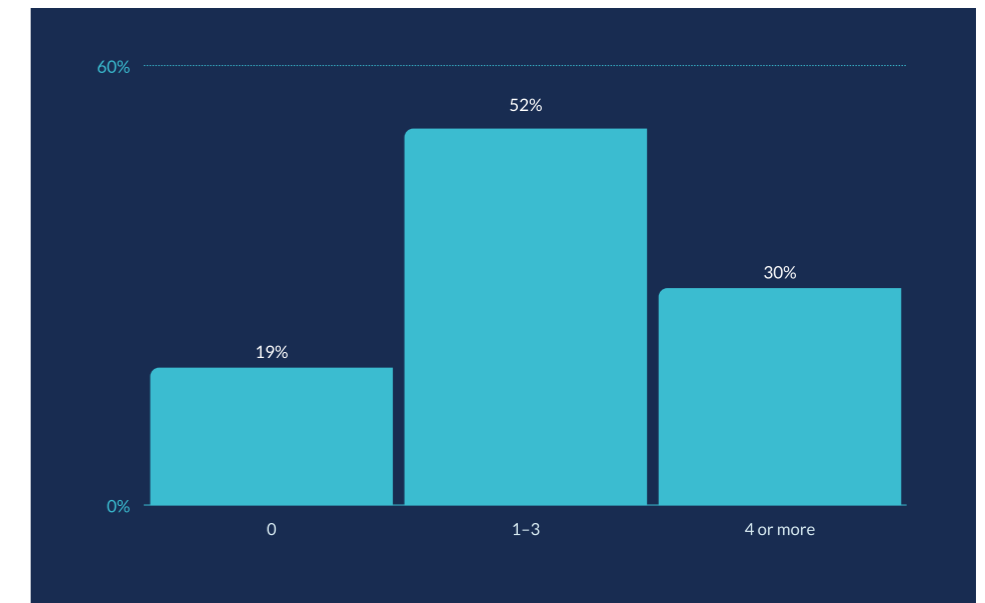
These findings become even more concerning when compared with broader rare disease data. Findings in Genetic Alliance UK’s Rare Experience 2020 report<sup>4</sup> showed that 44% of people with a rare disease received no misdiagnosis at all, and only 18% received two or more misdiagnoses. This stark difference suggests several factors at play: (i) symptom overlap between achalasia and far more common conditions, such as reflux and eosinophilic oesophagitis, (ii) limited front-line access to, and interpretation of, specialist tests for achalasia, and (iii) a general knowledge gap about achalasia within primary and emergency care. We examine these contributing factors later in the report.

4. Genetic Alliance UK (2020). RARE EXPERIENCE 2020. Rare-Experience-2020-Report-updated-May-2021-.pdf

The most frequently reported misdiagnoses reveal how achalasia symptoms are routinely misinterpreted. Affecting over 70% of all participants – many of whom were given unnecessary medication to treat the condition – acid reflux emerged as the predominant incorrect diagnosis. Anxiety followed as the second most common misdiagnosis, experienced by 38% of survey respondents, highlighting a troubling tendency to attribute physical symptoms to psychological causes. Eating disorders were also frequently incorrectly diagnosed in approximately 20% of cases, often leading to inappropriate treatment and a lack of both mental and physical support. It is worth noting that these misdiagnoses can occur concurrently, with patients being diagnosed with both reflux and anxiety, for example.

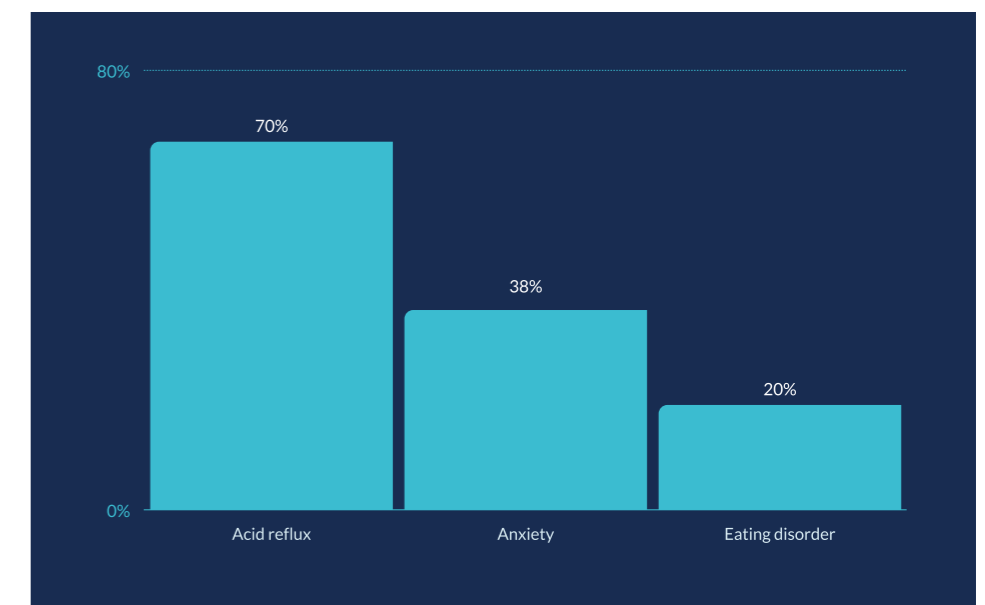
### Total number of misdiagnoses

*\*Due to rounding of individual values, percentages may not sum to exactly 100%.*



### Types of misdiagnoses

*\* Percentages do not add to 100 as some respondents received multiple misdiagnoses*



For those misdiagnosed with an eating disorder, the impact was particularly devastating:

Rebecca:

*“I remember there being a big assumption about my age, that I’m young... there was definitely one or two doctors that felt that I had an eating disorder, which was an interesting one. And I think that was highlighted by the fact that I was a young woman, which is actually probably the most damaging assumption for me. And it still sits with me today in that it’s all in my head.”*

Each misdiagnosis represents not simply an error in judgement, but months and years of unnecessary suffering, inappropriate treatments and progressive physical deterioration that may have been prevented through earlier recognition of the condition.

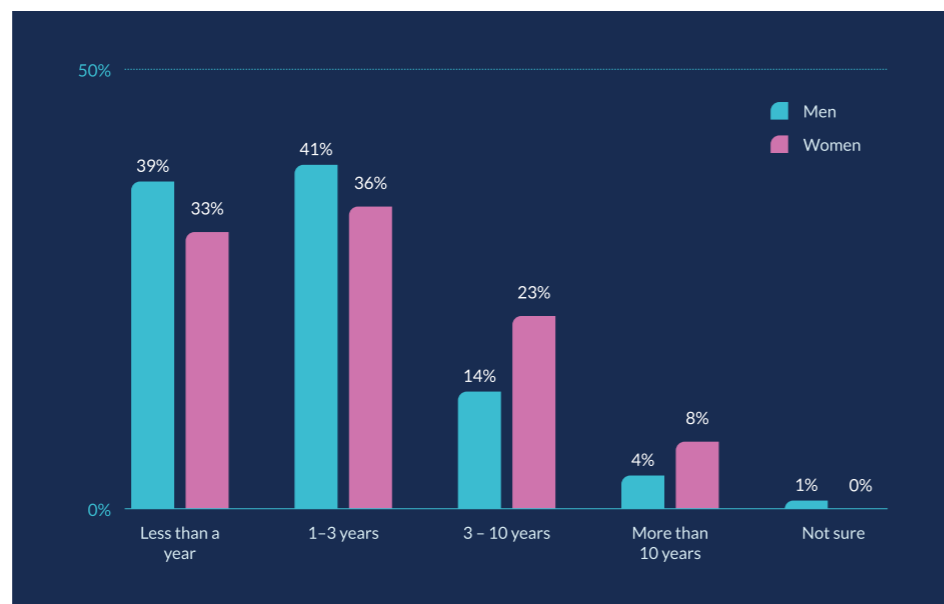
When diving deeper into the data, our research indicates striking gender disparities. Women regularly face longer and more challenging journeys to an accurate diagnosis than their male counterparts. While 39% of men were diagnosed with achalasia in less than a year of seeking support, only 33% of women experienced the same.

This gender disparity widens for people facing more lengthy diagnostic delays. Women were 13 percentage points more likely to experience severe delays, with 31% of women waiting more than three years for a diagnosis, compared to 18% of men. This pattern shows that women with achalasia bear a disproportionate burden in navigating the health system.

## Gender disparities

### Diagnosis time by gender

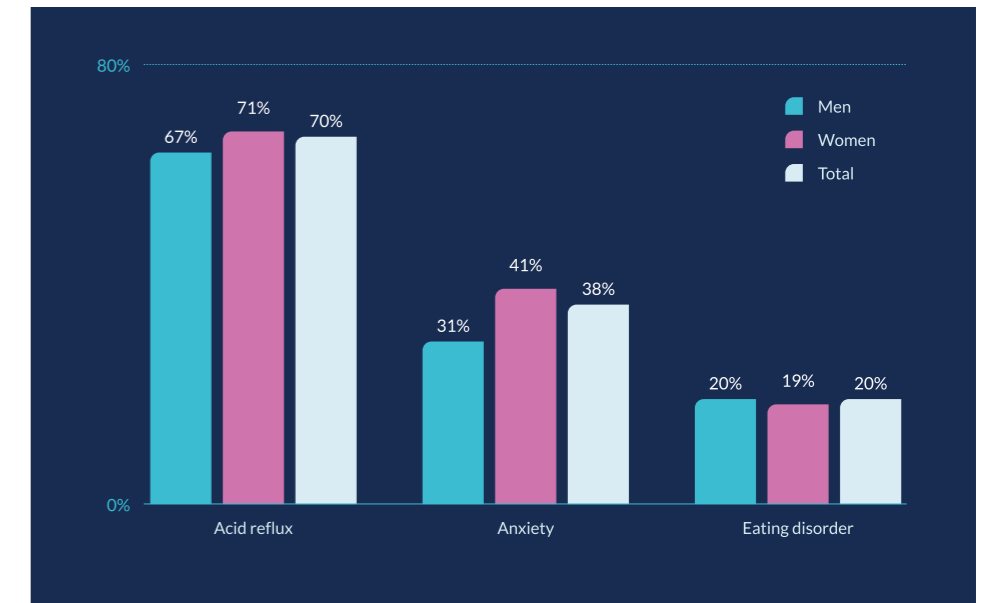
*\*Due to rounding of individual values, percentages may not sum to exactly 100%*



### Most common misdiagnosis

*\*Percentages do not add to 100 as some respondents received multiple misdiagnoses*

These gender disparities also extend beyond the timeline for getting a diagnosis of achalasia. Women reported higher rates of multiple misdiagnoses, with 33% receiving more than four incorrect diagnoses compared to 27% of men. Even more revealing is the types of misdiagnoses - women were 10 percentage points more likely to have their physical symptoms incorrectly attributed to anxiety (41% for women; 31% for men), indicating gender bias in symptom interpretation.

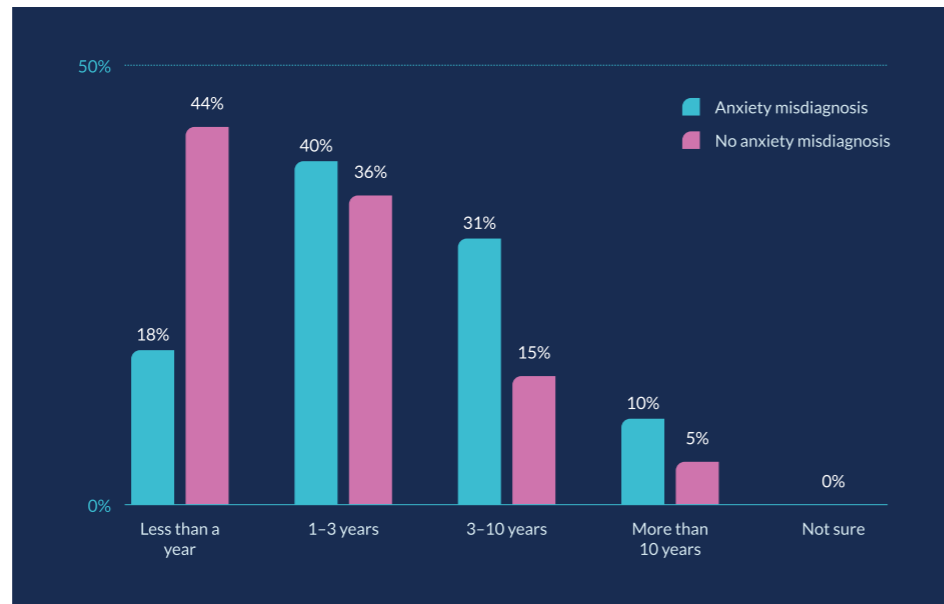


The consequences of being misdiagnosed with anxiety are especially striking. People misdiagnosed with anxiety were 21 percentage points more likely to wait more than three years for a correct diagnosis, and 5 percentage points more likely to wait over a decade.

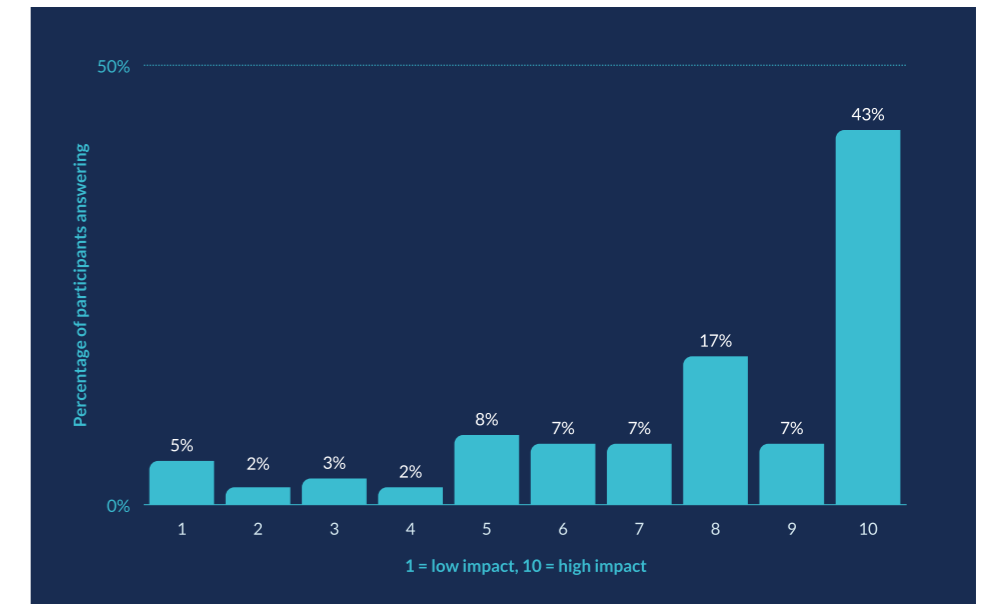
Women face a compounded challenge: their physical achalasia symptoms are more frequently misattributed to anxiety, which not only delays proper treatment but also extends their diagnostic journey compared to men. This creates a cycle where psychological misdiagnosis leads to longer suffering from untreated physical symptoms. The impact of having physical symptoms misinterpreted as psychological ones will be explored later in this report.

## Anxiety misdiagnosis vs No anxiety misdiagnosis

\*Due to rounding of individual values, percentages may not sum to exactly 100%



## How much did prolonged diagnosis/misdiagnosis affect your physical health



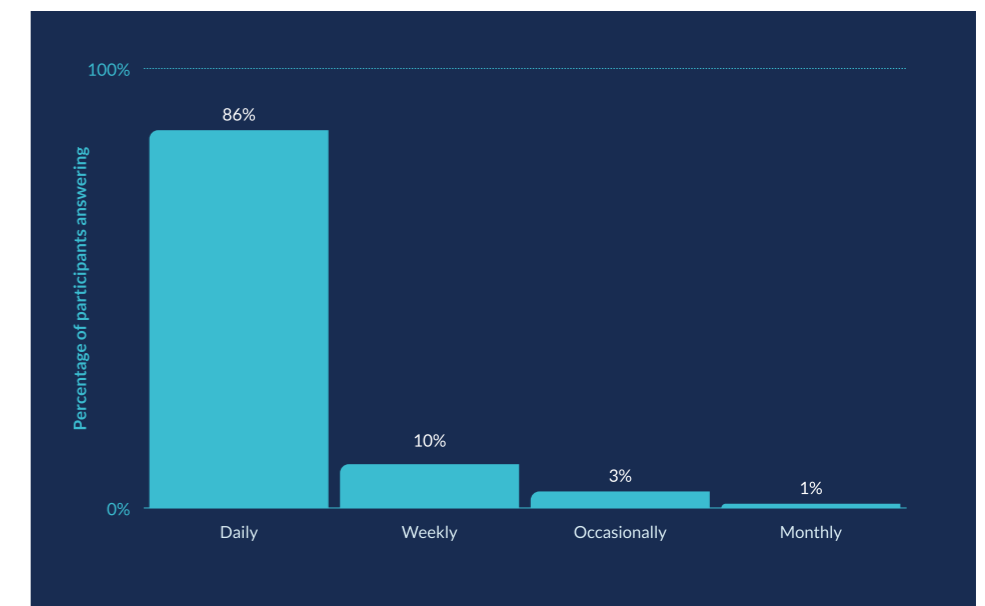
Dean:

*"I might be wrong, but I have a real strong feeling that men and women are treated differently."*

Janet:

*"Being a woman of 63, you know, I also appreciate that there is unfortunately a very bad habit amongst a small but significant minority of the medical community to assume that things going wrong in women's lives are actually connected with hysteria, anxiety, you know, and so I was immediately kind of a bit horrified at that"*

## How often did you experience symptoms before diagnosis?



## The physical toll of prolonged diagnosis and misdiagnosis

Our survey revealed the extreme impact on physical health that people face when waiting for a diagnosis. When asked to rate the impact of prolonged diagnosis or misdiagnosis on a scale of one (low impact) to ten (high impact), 43% of respondents rated it 10/10, while 74% rated it within the 7-10 range. These strikingly high ratings demonstrate the debilitating nature of undiagnosed achalasia and highlight how quality of life is compromised during the pre-diagnosis period.

The relentless nature of the symptoms people experience also compounds their severity. When asked, 86% of people said that they experienced symptoms daily while waiting for a diagnosis. The constant presence of symptoms - difficulty swallowing, regurgitation, chest pain and weight loss - means that affected individuals rarely experience relief during the diagnostic journey.

Tamsin's story, shared during a one-to-one interview, vividly illustrates the devastating physical toll that people waiting for an achalasia diagnosis endure.

What started as a minor inconvenience quickly evolved into a serious medical challenge and the progression of her condition became hard to ignore. Tasmin recalled losing “half a stone in a week,” which was a critical turning point for her. Her weight plummeted dramatically, she went from a size 14 to a size 10-8, and eventually to a size four. “I went [to] less than seven stone,” she explained, highlighting the extreme physical toll of her undiagnosed condition.

**Tasmin:** *“I was on my lunch break, and I was just having a sandwich, and I just noticed that I couldn’t actually swallow it very well. It was just getting a little bit stuck, and I didn’t really think anything about it.”*

The consequences for Tamsin went far beyond weight loss. She recalled:

**Tasmin:** *“I just basically went to bed all day because I just couldn’t do anything. I had no energy because it impacted me, I couldn’t work, I couldn’t do anything,”*

As her body continued to get weaker, her menstrual cycle ceased, and even walking became nearly impossible. The situation spiraled toward what Tamsin and her family recognised as potentially fatal:

**Tasmin:** *“We all just thought if something’s not done, I’m gonna go into organ failure or something.”*

Tamsin’s story, while shocking, reflects the lived experience of so many people who are waiting for an achalasia diagnosis.

## The psychological and social toll of prolonged diagnosis and misdiagnosis

The significant mental toll of waiting for a diagnosis was a common and repeated theme shared by those we spoke to throughout our research. This ranged from experiencing anger that affected relationships with loved ones, shame that affected their self-confidence, to a sense of grief surrounding the unexplained symptoms. This social impact should not be downplayed, with respondents highlighting that relationships, friendships and families were significantly impacted by their experience.

**Jemma:** *“I lost confidence with the doctors. I was just getting so angry with everybody. Like even in the house, I was just so angry with the family, I didn’t know what else I could do”*

**Amy:** *“I feel like I’m grieving, you know. I just want to know if this procedure’s going to work and if I’m going to eat again”*

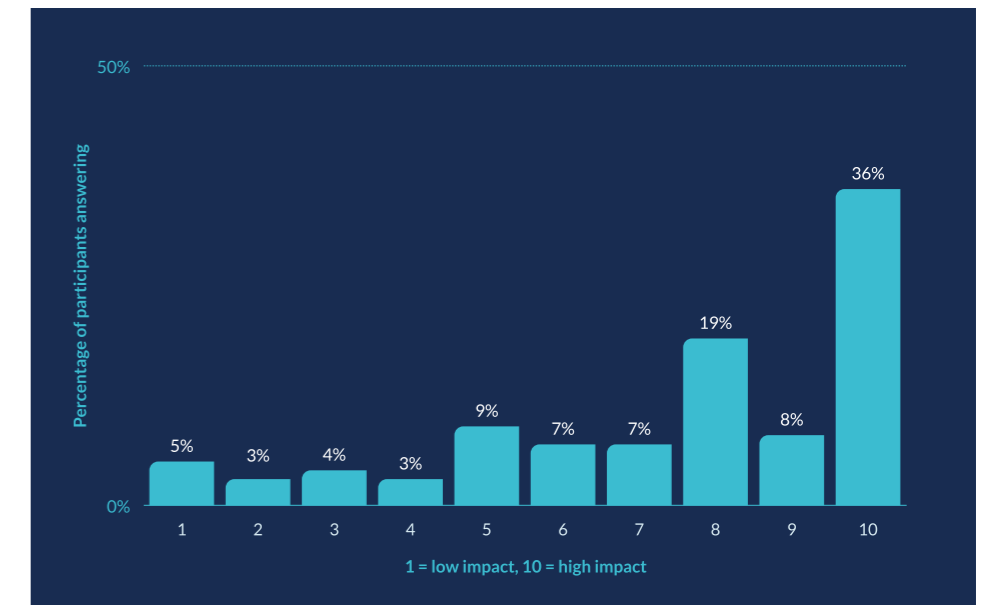
**Dana:** *“A lot of like self blame, even though I know that’s not rational, but I still feel like, yeah, it still feels like my fault in some way. “*

**Rebecca:** *“My mood was just completely gone. I used to walk into the kitchen and if I saw my housemate cooking, I’d be like, you’re so selfish that you cook in front of me, which is mad. Like, I hate that I did that. It’s just, it changed this, the whole thing, that whole period of time changed who I was as a person.”*

The survey data confirms the testimony that we collected through our interviews. 36% of respondents reported maximum psychological distress, rating the impact at 10/10. Approximately 70% rated the psychological impact in the high range (7-10) and only 12% rated it as a score of three or below. This underscores how few escape the mental health consequences of prolonged diagnostic journeys.

### How much did the prolonged/ misdiagnosis affect your mental health?

*\*Due to rounding of individual values, percentages may not sum to exactly 100%*



The data and personal testimonies collected through our research paints a clear picture of the significant challenges faced by those awaiting an achalasia diagnosis. The prolonged waiting periods, multiple misdiagnoses, and profound physical and mental health impacts highlight critical gaps in current diagnostic pathways for achalasia and their effects on people with the condition. The gender disparities are clear, demonstrating the systemic bias that is embedded in our healthcare system, especially when dealing with symptoms of a rare disease. These findings underline the urgent need for improved diagnostic pathways, which will be explored later in this report.

# 07

## Systemic health service failures

This chapter explores the structural failures within healthcare systems that contribute significantly to delayed achalasia diagnosis. Our research reveals a fragmented healthcare journey characterised by inconsistent diagnostic pathways, limited awareness among healthcare professionals, and critical gaps in patient information and support.

### The complex journey to diagnosis

A key finding from our research is the striking absence of a standardised diagnostic pathway for achalasia. When asked about their diagnostic journey, participants reported encountering a disjointed series of different healthcare professionals and tests. When asked what diagnostic tests participants received before being diagnosed, 13 different tests were mentioned, reflecting both appropriate diagnostic procedures, but also tests that suggest patients were placed on complex diagnostic pathways.

Among these tests, six are recognised diagnostic procedures typically used to correctly identify achalasia:

Name of test	Description
Manometry	Measures muscle tone and contraction and relaxation in the oesophagus and sphincter.
Barium Swallow	X-ray using contrast to view swallowing function and the shape and anatomy of the oesophagus and stomach.
Endoscopy	Camera test to examine the oesophagus and stomach lining.
Esophageal pH Test	Assesses acid levels in the oesophagus over time.
Biopsy	Tissue samples taken to check for inflammation or disease.
EndoFLIP	Measures oesophageal pressure and diameter in real time.

The remaining seven investigations that were mentioned can be clinically helpful to rule out aspiration sequelae or concurrent pathology, but they are not central to confirming achalasia itself.

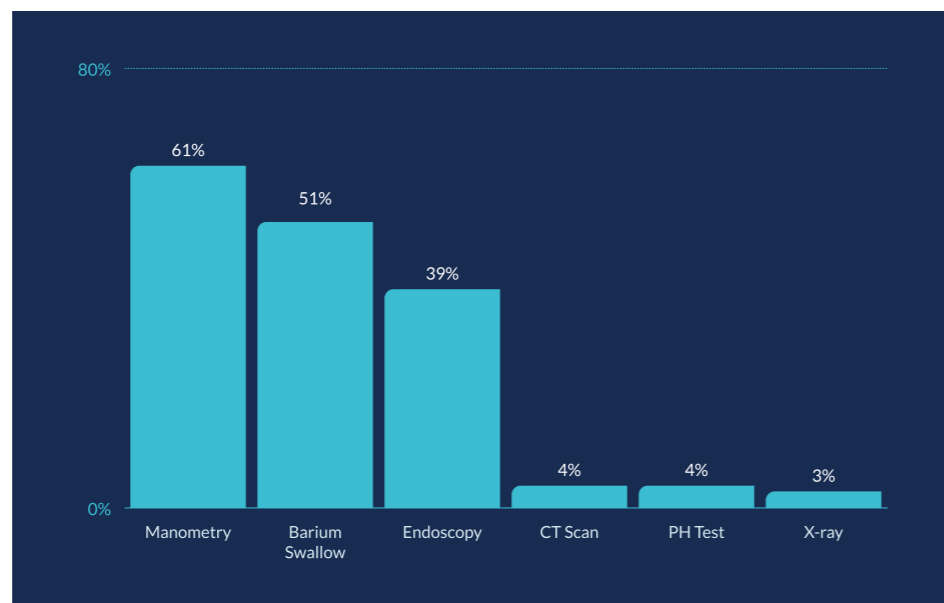


Name of test	Description
CT Scan	Detailed body imaging using X-rays.
X-ray	Basic imaging to check for structural abnormalities.
Blood Tests	Checks for signs of inflammation.
Ear Nose and Throat Tests	Examines ENT structures for contributing issues.
Haptoglobin Test	Blood test for signs of haemolysis or inflammation
MRI	High-resolution imaging using magnetic fields.
Lung Testing	Evaluates breathing and lung function.

As a result, many patients spend months in parallel pathways – respiratory, ENT, gastro-oesophageal reflux – before the right oesophageal motility tests are considered, prolonging the overall journey to treatment.

### Percentage of participants who had the test

Note: Percentage do not add to 100% as respondents often get multiple diagnostic tests



The data presents a clear pattern: manometry, barium swallow and endoscopy are the most common diagnostic tests for achalasia<sup>5</sup>. However, manometry, which is widely considered the gold standard test for diagnosing achalasia, was administered to only 61% of participants. This means nearly 4 in 10 patients received their diagnosis without this crucial test. When conducting interviews and focus groups, the long wait for a manometry test and the lack of availability of manometry testing on the NHS was raised repeatedly.

5. Pohl, D., & Tutuian, R. (2007). Achalasia: an overview of diagnosis and treatment. *Journal of Gastrointestinal and Liver Diseases*, 16(3), 297-303

Abbie:

*“It seemed a bit crazy that they were giving me these endoscopies. I had I don’t know how many endoscopies before I had the manometry, which looking back was a hell of a waste of time and money”*

This patchwork approach extends to the healthcare professionals that patients encounter during their diagnostic journey. Our survey revealed that patients typically consult numerous healthcare professionals before receiving their diagnosis. When asked which healthcare professionals they saw before receiving a diagnosis, 17 different types of healthcare specialism were mentioned.

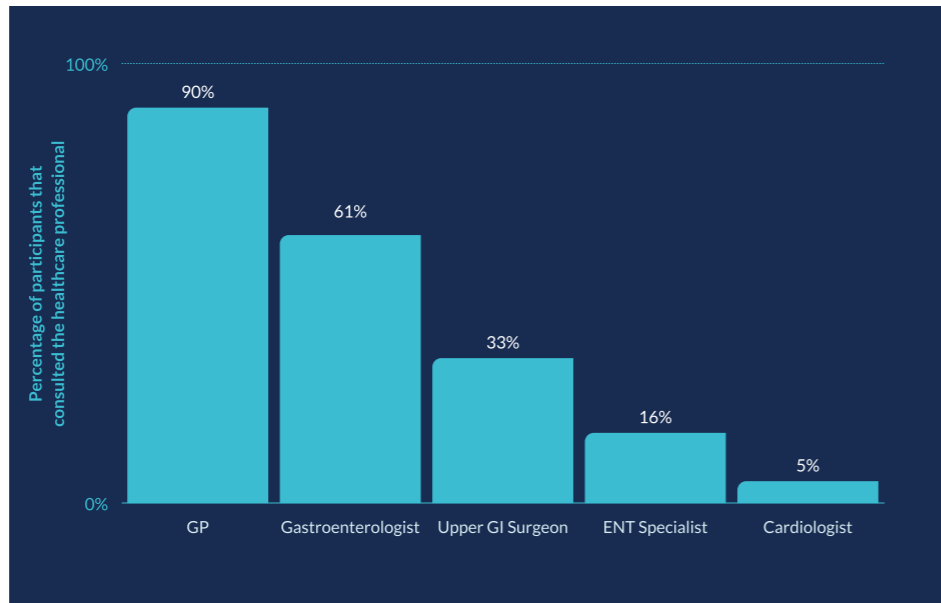
Professional or Service	Description
General Practitioner (GP)	First point of contact for medical concerns and referrals.
Gastroenterologist	Specialist in digestive track disorders.
Upper Gastroenterologist	Focuses on oesophagus, stomach, and upper digestive tract.
ENT Specialist	Treat ear, nose and throat conditions.
Cardiologist	Specialist in heart and circulatory system
A&E/Emergency Department	Urgent care for acute or severe symptoms.
SaLT/Speech Therapist	Assesses and treats swallowing and speech issues.
Physician Assistant	Supports doctors in diagnosis and treatment.
Paediatrician	Medical care for infants, children and adolescents.
Respiratory/Chest Specialist	Focuses on lung and breathing disorders
Mental Health Team	Provides support for psychological and emotional wellbeing.
Asthma Team	Manages and supports asthma care.
Autoimmune Specialist	Diagnoses and treats immune system disorders.
Endoscopist	Performs internal camera-based examinations like endoscopies.
Thoracic Specialist	Treats diseases of the chest, including lungs and oesophagus.
Dietician	Offers nutritional guidance and dietary support.
Psychiatrist	Medical doctor specialising in mental health treatment.

The number of different professionals involved further highlights how fragmented the diagnostic journey can be. Without a clear and standardised pathway, patients navigate a complex system, being passed from team to team often without a resolution.

It is clear from the data that the pathway to getting a diagnosis lacks consistency. Nonetheless, we can also see clear patterns within the data that show where the greatest impact could be felt if changes were implemented.

### Types of healthcare professionals consulted by participants

Note: Percentage do not add to 100% as respondents often consult multiple healthcare professionals



Our survey shows that 90% of participants sought support from their GP during their journey to diagnosis. This overwhelming majority highlights primary care as the critical point of contact for people seeking support for symptoms of achalasia. GPs serve as essential first advocates and navigators for patients, guiding them toward specialist referrals and further testing. This makes them the single most influential touchpoint for potential achalasia patients. Consequently, targeted interventions at the primary care level could have the most substantial impact on reducing diagnostic delays.

### Knowledge, communication, and information from health professionals

A significant finding from our research was the overwhelming consensus regarding healthcare professionals' lack of awareness about achalasia. When asked what could have helped secure an earlier diagnosis, 295 out of 350 survey participants specifically mentioned "better awareness of achalasia among healthcare professionals."

The personal testimonies from our research participants highlight the impact of this lack of awareness:

Dana:

*"I feel like my journey to get diagnosed was a really long and stressful one... I just don't want other people to have to go through that as well. And I agree with [name of other focus group participant] that there's a lack of knowledge in doctors and clinicians and definitely a lack of empathy as well."*

Jane:

*"I think one of the biggest gaps that we've got is the clinicians, the doctors, everybody involved in the healthcare system just having a lack of knowledge. And I think what then can come with that is lack of empathy as well."*

Our research also uncovered concerning patterns in communication between healthcare providers and with patients. Many interview and focus group participants described fragmented communication between different specialists and departments, creating frustrating delays and repetitions in their care:

Jane:

*"No one really talks to each other. And it's just really, really annoying when you think you're getting somewhere. And then you realise that they haven't told that person."*

Equally concerning was the significant information void that many people experienced after receiving their diagnosis. Participants frequently reported being given minimal or no information about their condition, treatment options, or support resources:

James:

*"But for me, the worst was effectively being given my diagnosis over the phone by a secretary, and no elaboration. If I hadn't read about achalasia myself, if I didn't already know what that condition was, a secretary going, "you've got achalasia, you're on the waiting list for surgery."*

Rose:

*"There was no, 'here's a leaflet about it. Here's someone to speak to about it. Go and speak to this person about it'. Nothing about that at all. It was just, 'we suspect you've got achalasia'. And then you go home."*

These systemic failures create an environment where patients often feel abandoned and unsupported, left to navigate a complex condition with minimal guidance. The consequences of these failures extend beyond delays in diagnosis - they erode trust in healthcare systems, create unnecessary suffering, and potentially lead to poorer long-term outcomes for people living with achalasia.

The experiences documented in our research point to clear opportunities for improvement in healthcare professional education, communication protocols, and patient information provision, all of which will be explored in our recommendations section.

# 08

## Medical dismissal

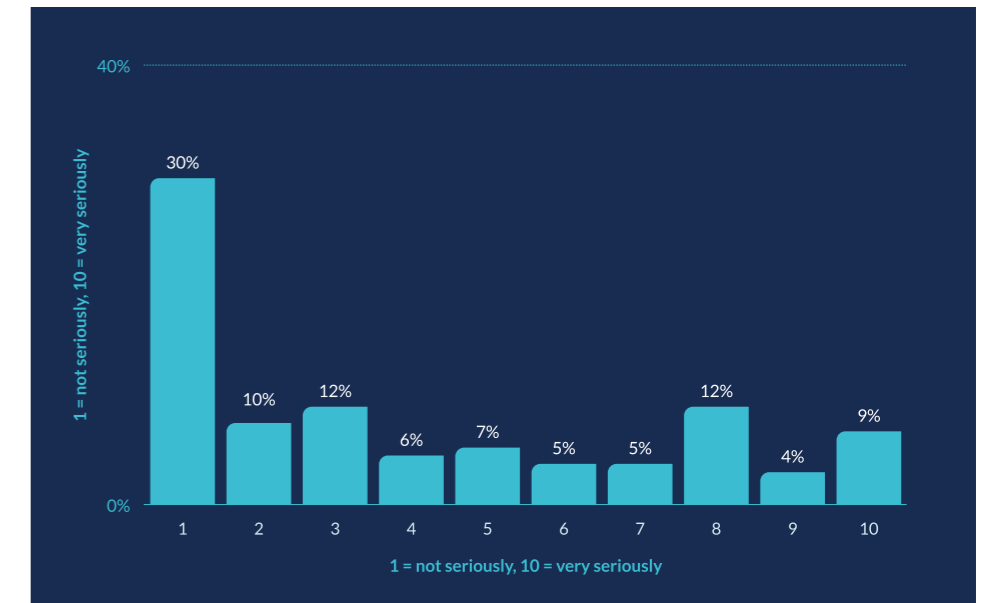


### The extent of medical dismissal

Were you taken seriously by your healthcare provider before your diagnosis?

Throughout our research, we heard repeated stories of people feeling unheard, invalidated and dismissed by their healthcare professionals. This chapter explores these patterns of dismissal and disbelief, shining a light on the impact on people with achalasia.

In our survey, we asked participants to rate how seriously they were taken by healthcare providers before diagnosis on a scale of 1-10 (with 1 being “not seriously at all” and 10 being “very seriously”).



52% of participants gave a rating of 3 or lower, meaning that they felt they were not taken seriously during the pre-diagnosis experience. Of these, 30% gave a rating of 1, indicating that they felt completely dismissed by healthcare providers. Only 9% of participants gave a rating of 10.

This widespread experience of medical dismissal appears to align with our findings on misdiagnosis rates and diagnostic delays. Based on the patterns observed, we can infer that when patients aren't taken seriously, their symptoms are often incorrectly attributed to more common conditions, particularly acid reflux (reported by 70% of participants), anxiety (39%), or eating disorders (20%). These misdiagnoses then lead to inappropriate treatments and further delays in receiving the correct diagnosis and care.

While pre-diagnosis experiences were challenging for many, our research revealed a different picture once patients reached the healthcare professional who ultimately diagnosed their achalasia. When we asked participants to rate how seriously they were taken by these diagnosing professionals on a scale of 1-10, the results were overwhelmingly positive.

A substantial 77% of participants gave ratings of 8 or higher, with over half (53%) giving the maximum rating of 10. This indicates that many specialists do recognise and validate achalasia symptoms once patients reach the right healthcare provider. Only 4% gave the lowest possible rating of 1, suggesting that negative experiences with diagnosing professionals are relatively rare.

These findings highlight that significant expertise and empathy exists within the healthcare system, particularly among specialists familiar with achalasia. As one participant, Erica, expressed:

**Erica:** *“My medical treatment and my journey has been like, entirely improved since I’ve had the diagnosis.”*

The contrast between pre-diagnosis and post-diagnosis experiences suggests that improving referral pathways to connect patients with these knowledgeable professionals earlier could significantly improve the overall patient journey. When patients finally receive their diagnosis, they typically encounter healthcare professionals who take their symptoms seriously and provide appropriate validation and care.

### **The psychological burden of symptoms being misattributed to other causes**

The psychological burden that patients face when their symptoms are dismissed or misattributed to psychological causes by healthcare professionals emerged as a central theme in our research.

Two participants’ experiences illustrate how devastating this can be for patients navigating the long path to diagnosis.

At 63 years old, Sarah experienced classic achalasia symptoms of regurgitation and difficulty swallowing. Her journey through the healthcare system demonstrates the profound impact that not being believed had on her psychological wellbeing.

When she consulted her GP about regurgitating food, Shirley recalled: “The GP said that’ll be acid reflux.” Even when she explained there was “no discernible acid,” the doctor remained unconvinced, simply prescribing omeprazole without considering alternative diagnostic tests.

As her symptoms progressed to include what she called “explosive regurgitations,” Sarah sought additional medical help. During one particularly distressing consultation, a physician openly laughed when she described her severe pain:

**Sarah:** *“he literally laughed out loud when I told him that, as though he thought I was making it up or exaggerating.”* ”

This experience was deeply humiliating for Sarah, who at the time was navigating her diagnosis journey alone.

Perhaps most distressingly for Sarah, one referral letter suggested a “functional cause” for her symptoms. As a former barrister with experience in personal injury cases, Sarah interpreted this as medical code implying she was fabricating her condition. Crucially, no healthcare professional explained what “functional cause” meant in this context, leaving her to draw on her legal background to interpret the terminology.

**Sarah:** *“When I read this, I almost felt as though somebody had actually punched me in the stomach, I felt so humiliated and horrified to think that this man to whom I’d been revealing what was going on in my life would be taking an objective look at me and imagining that I was a fantasist or an exaggerator.”*

The psychological impact was profound:

**Sarah:** *“It made me feel physically sick. It was psychologically damaging.”*

This experience further isolated Sarah when navigating her symptoms, as she explained:

**Sarah:** *“I couldn’t tell my sister what had been said, because I felt ashamed of the suggestion that I was imagining it or exaggerating it, or just some inadequate person who was experiencing something that wasn’t really happening.”*

Another interview participant, Erica, spoke about the misattribution of physical symptoms to psychological causes. For nearly two years, healthcare providers repeatedly suggested her swallowing difficulties stemmed from anxiety or other psychological issues rather than investigating physical causes.

**Sarah:** *“It was all quite a lot of assumptions that it was a psychological issue. And that it wasn’t anything to do with a physical cause. And I felt quite strongly that that wasn’t the case,”*

Erica explained. Despite her medical knowledge as a mental health nurse, her concerns were dismissed:

**Sarah:** *“I would say, you know, I work in mental health, I would say if I thought it was anxiety.”*

Rather than investigating physical causes, Erica was directed toward psychological treatments:

**Sarah:** *“I had treatment for anxiety... two different treatments, you know, evidence-based treatment for anxiety... But it didn’t work for me, because obviously, that wasn’t my issue.”*

The psychological misdiagnosis escalated to questioning whether she had an eating disorder:

**Sarah:** *“Then there was a bit of a discussion about whether I had an eating disorder, whether I had bulimia. And I started to think, oh my God, I’ve got bulimia. And I’m so in denial that I don’t know I’ve got bulimia.”*

The misdiagnosis had far-reaching consequences beyond her health. Erica reduced her working hours and experienced career setbacks:

**Sarah:** *“I reduced my hours at work. And I went down some gradings at work... I think if I’d got a diagnosis earlier, I could have taken proper sick leave.”*

Perhaps most distressing was the effect on her young family:

**Sarah:** *“There was two whole years of not knowing what to say. So they were scared. I was scared. We’re all in the scared kind of thing dealing with these symptoms all together. And that is something that I’m really upset about because I think it’s had an impact, not just on me and I’m an adult, but on my young children.”*

## Long-term impact on trust in healthcare services

These experiences of medical dismissal have lasting consequences that extend far beyond the initial diagnosis period. Our research reveals how these negative encounters can fundamentally alter patients’ relationship with the entire healthcare system, creating barriers to future care and trauma that may persist for decades.

Many participants described developing significant anxiety about engaging with healthcare services after their experiences:

**Anna:** *“I feel like if I were to contact the NHS now, I’m scared that they’re going to upset something... that trauma that I’ve had... has definitely impacted my stay in hospital this time. Massively (...) It’s definitely impacted my ability to seek help from NHS services.”*

Perhaps most concerning is how persistent this trauma can be. One participant reported that these feelings of medical distrust endured for decades after their diagnosis:

**Anna:** *“It really gave me a lot of trauma about not being believed. And it does, I mean, it’s 20 odd years, it’s 25 years ago now, and it still does. It gives me trauma to this day.”*

Beyond the emotional toll, frustration was also highlighted at the inefficiency and waste within a system that failed to address their concerns appropriately:

**Ross:** *“...it doesn’t instill any confidence at all, really... what a waste of time of the NHS, her time, my time, I got nothing out of it.”*

This erosion of trust creates a concerning cycle with significant implications. Research shows that medical mistrust leads to “reluctance to accept diagnoses, limited engagement with healthcare providers, lower adherence to treatment protocols, reduced uptake of preventive measures, and poorer overall physical and mental health outcomes”<sup>6</sup>. Due to their poor experience of receiving an achalasia diagnosis, patients may delay seeking further help for their achalasia symptoms, or even other healthcare conditions. It also leads to patients approaching interactions with skepticism or avoiding healthcare services altogether.

This damaged trust represents a failure that undermines the effectiveness of healthcare delivery for a substantial population of people living with achalasia. Rebuilding trust is essential for improving health outcomes.

6. Souvatzi, E., Katsikidou, M., Arvaniti, A., Plakias, S., Tsiakiri, A., & Samakouri, M. (2024). Trust in healthcare, medical mistrust, and health outcomes in times of health crisis: A narrative review. *Societies*, 14(12), Article 269.

# The burden of self-advocacy

7. Halley, M. C., Halverson, C. M. E., Tabor, H. K., & Goldenberg, A. J. (2023). Rare disease, advocacy and justice: Intersecting disparities in research and clinical care. *American Journal of Bioethics*, 23(7), 17–26.

8. Tumiene, B., Peters, H., Melegh, B., Peterlin, B., Utkus, A., Fatkulina, N., Pfliegler, G., Graessner, H., Hermanns, S., Scarpa, M., Blay, J.-Y., Ashton, S., McKay, L., & Baynam, G. (2022). Rare disease education in Europe and beyond: Time to act. *Orphanet Journal of Rare Diseases*, 17(1), 441.

Navigating healthcare systems for anyone with a chronic health condition is a challenge, but for those with a rare disease like achalasia, this challenge becomes even more burdensome. Research has consistently shown that people with rare diseases must develop sophisticated self-advocacy skills to receive the care they need<sup>7</sup>. The need for self-advocacy is particularly critical for individuals with rare diseases for several reasons. For example, healthcare professionals receive minimal training about rare diseases during their training and most clinicians only encounter a handful of rare conditions throughout their career<sup>8</sup>. Additionally, the siloed nature of specialist care means that patients tend to become the primary advocates of their own needs by translating and communicating information between healthcare professionals.

The burden of self-advocacy should not be understated, and its impact was consistently highlighted throughout our research. This chapter explores the extraordinary measures some patients take to secure a correct diagnosis of achalasia. Our research reveals that diagnosis and treatment often depend not on the healthcare system's ability to identify and address symptoms, but on patients' capacity to persistently advocate for themselves. For many, the path to diagnosis becomes a test of determination and, for those who lack the ability and resources to self-advocate, the condition may remain undiagnosed for too long.

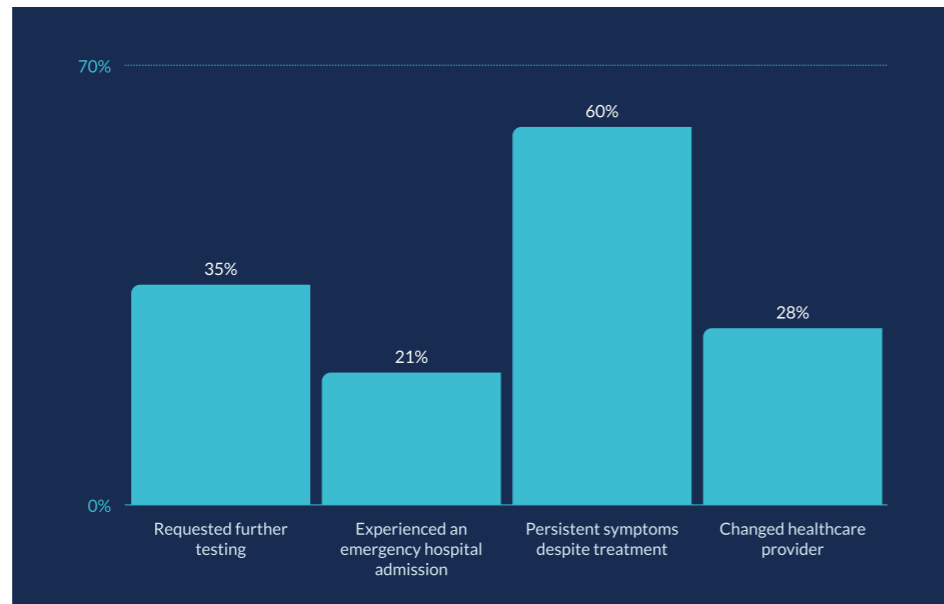
## The necessity of patient persistence

Through our survey, we asked participants what led to them receiving an achalasia diagnosis. The findings show that diagnosis often depends on patient actions rather than standard diagnostic pathways.



## What actions lead to a diagnosis

Note: Percentage do not add to 100% as respondents often consult multiple healthcare professionals



Over one-third (35%) of patients had to actively request further testing to reach a diagnosis, showing how patients must push for the correct care. 60% of patients only received their diagnosis after experiencing persistent symptoms despite treatment, indicating that many healthcare providers continued with ineffective approaches until the patient's condition became undeniable.

Particularly concerning is that 21% of participants experienced their symptoms getting so bad they had to seek support from emergency services to receive care that led to a diagnosis. This represents more than one in five people with achalasia whose condition deteriorated to the point of needing emergency intervention - a situation that could have potentially been avoided with an earlier diagnosis. Not only does the significant presence of people with achalasia in emergency services represent the severe nature of achalasia symptoms, it also shows a breakdown in the primary care pathway. This highlights both the urgent need for better recognition in primary care settings to prevent individuals deteriorating to this extent and better education in emergency medicine to ensure people are quickly directed to the correct diagnostic and treatment pathway.

These statistics paint a picture of a healthcare system that fails to proactively identify achalasia, placing the burden of diagnosis squarely on patients themselves. Whether it is by having the confidence to request the correct diagnostic testing, having the persistence to continuously reach out for support despite being incorrectly treated, or having to proactively find a different healthcare provider, being a self-advocate is often key to getting an achalasia diagnosis.

## The extreme measures required

Throughout our interviews and focus groups, participants described taking increasingly desperate measures to be heard by healthcare professionals. Many detailed how conventional approaches to seeking medical care proved ineffective, forcing them to adopt extraordinary tactics:

*"So it literally got to the point he was calling the secretary every day and leaving a message saying, look, I'm really worried, please, please help."*

Some participants described refusing to leave medical appointments until they received appropriate care or tests:

*"Don't get up, refuse... I remember just like holding on to a chair saying, 'I'm not leaving until you do something.'"*

*"In my mind, I had decided until we know exactly what this is and I have a confirmed diagnosis, I am not leaving this. I don't care. I don't care if I'm here for months on end."*

This level of persistence shows how people are often forced into assertive or even desperate measures to be taken seriously, far beyond what should be necessary in routine care. These accounts show a system where standard communication methods are not successful, forcing people into uncomfortable positions where self-advocacy is the only way to legitimise their symptoms.

We also heard from many who took on the role of medical researchers in the absence of adequate support. This shows how people with achalasia often reach the point where they had to research their own potential diagnosis in order to identify the diagnostic tests that they were not receiving:

*Carly:*

*"I had Googled my own symptoms and realised it must be achalasia and went in and said, this is the test I want, which was the oesophageal manometry. (...) Everything came from me. I had to do all the research."*

## Paying to be heard

When NHS pathways failed to provide adequate and timely diagnosis and treatment, many of our research participants resorted to using private healthcare services. Our survey revealed that 16% of participants were diagnosed by a private healthcare provider. While we cannot tell how many of these participants are regular users of private healthcare services, we did hear throughout our interviews and focus groups that this was a last resort due to the long waits within the NHS.

*Julie:* “I wasn’t getting anywhere. So I actually got a consultant privately, because I was just really fed up with waiting. So I paid for a private consultation.”

*Andrea:* “I was seeking a private route to have the operation. Because the NHS was just a very long wait.”

For some, this financial burden created additional emotional distress, including guilt about using family resources for essential healthcare:

*Dana:* “My mum had been made redundant from work, a job she’d been at for about like 20 odd years and had a redundancy payout and used most of that to pay for my consultations, my surgery... I have a lot of guilt around that.”

Others felt they had no choice but to find the money for private care, despite the financial strain:

*Mary:* “And I was like, I can’t, I can’t live another two years with this. I can’t. So I decided to save up and go see somebody privately, locally.”

The decision to pay for private healthcare is rarely made lightly by patients. Through our qualitative research we heard that it often comes after multiple failed attempts to receive adequate care through the NHS. This represents not a preference but a desperate measure taken when all other options have been exhausted. This creates a system where diagnosis speed may be determined by financial capacity rather than medical need.

## The inequity of self-advocacy

The reliance on both persistent self-advocacy and financial resources to access timely diagnosis reveals deep inequities in care access. As participants in our research interviews and focus groups noted, the necessity for self-advocacy means that those without the personal resources, confidence, or capability to push for care may struggle to, or may perhaps never receive appropriate diagnosis and treatment:

*Mary:* “But how do you make a doctor listen to you? I mean, the way I did it was I just kept going back until they had no other option. “I’m going to keep coming back to your door...” But I think if you’re not a persistent person, you will fall to the wayside, you will just get left. But I don’t know how you would make it... I don’t know how you would make them listen.”

This highlights the existence of a two-tier healthcare system: one for those with the resources, determination, and capacity to advocate relentlessly or pay privately; and another for those without these advantages. This may systematically disadvantage:

- Those who lack confidence or assertiveness skills
- People with language barriers or communication difficulties
- People with other cultural barriers or a natural deference to healthcare professionals
- Individuals with limited health literacy or internet access
- Those without financial resources for private healthcare options
- Groups who may already experience medical dismissal more frequently based on marginalised identities.

When effective diagnosis and treatment become dependent on a patient’s ability to advocate persistently and potentially pay privately, the system fails its fundamental purpose of providing universal health care based on need rather than means or personal capability. These findings show that improving achalasia diagnosis cannot focus solely on medical knowledge or testing availability – it must also address the structural issues that currently force individuals to persistently self-advocate to receive appropriate care.

# 10

## Conclusion and Recommendations: building a better future for achalasia diagnosis in the UK



Our research reveals important opportunities to transform achalasia diagnosis in the UK. We heard about the experiences of people who faced significant delays, multiple misdiagnosis and significant physical and psychological suffering. The experiences of the people who decided to share their stories with us serve as a wakeup call and a guide for meaningful change. To address the challenges effectively, this will require collaboration from healthcare providers, policy makers and patient support groups.

As we look to the future, it is important to take inspiration from some of the great care that exists. Though many people are let down through their journey to diagnosis, one thing is clear from our research: there are still examples of great practice that can be looked to as a beacon of hope. From the 35% of patients who received a diagnosis within a year, to the 77% whose experience drastically changed when interacting with the clinicians who diagnosed them correctly, the transformative impact of knowledgeable, empathetic care is profound.

With this in mind, increased medical awareness of achalasia is absolutely critical to ensure that those who are facing symptoms are able to reach the right medical care at pace. An improved awareness of achalasia among GPs, where 90% of people first go to find support, would make the most impact in transforming the diagnosis journey. However, to address the issues we have heard throughout our research, it will take much more than raising awareness. We need standardised diagnostic pathways, better communication protocols between healthcare providers, additional support resources for those navigating the diagnostic journey and a removal of bias that leads to people being dismissed when seeking help.

The recommendations that follow offer practical and actionable steps towards this goal. Some of these recommendations are things that Achalasia Action are already working towards, some are things we as a charity can strive towards in the future. However, many of these recommendations sit outside of the remit of our charity and will require significant policy and service delivery change in the NHS.

# Recommendations

## For the healthcare system

The findings of this report highlight that there is a need for systemic changes in the NHS that require concerted action from healthcare providers and purchasers, policymakers, professional membership bodies, and government.

### 1. Enhancing healthcare professional education on achalasia

- Medical training and continuous professional development (CPD) for healthcare professionals should include mandatory rare disease modules. At a minimum, the Royal College of General Practitioners (RCGP) and the British Society of Gastroenterology, who have a significant role in setting national training curricula for GP and gastroenterology specialisations, should ensure that training emphasises early recognition of key achalasia symptoms.
- Medical training and continuous professional development (CPD) for healthcare professionals should address gender bias in symptom interpretation, with a particular focus on challenging the tendency to attribute unclear and complex physical symptoms to psychological causes in women.

### 2. Improving achalasia diagnostic pathways

- Publish a nationally agreed diagnostic algorithm that sets out the earliest appropriate referral point from primary care for persistent upper-GI symptoms, irrespective of age, weight-loss or gender.
- Integrated Care Systems should ensure increased availability of oesophageal manometry across NHS trusts. Improving access to this test is particularly important, given that NHS England identified manometries as clinically unsuitable for delivery in community diagnostic centres, putting at risk equitable patient access to manometries. Increasing the availability of oesophageal manometry procedures will reduce regional inequalities in access to this essential achalasia diagnostic test.
- The National Institute for Clinical Excellence (NICE) should develop achalasia diagnosis, treatment and management guidelines. Standardised diagnostic pathways, with clear timeframes for progression through diagnostic steps, will support reducing the burden of self-advocacy on patients. In the absence of NICE guidelines, Achalasia Action should lead on a consensus-driven UK guideline development process, and make the guidelines widely available across primary care, gastroenterology and emergency services.

### 3. Implementing supportive communication standards and aftercare plans for people with achalasia

- Integrated Care Systems, NHS providers and primary care providers should improve the standards of communicating an achalasia diagnosis, by ensuring face-to-face explanation of the condition upon diagnosis, setting expectations about appropriate levels of support that patients should receive when diagnosed, and providing resources for healthcare professionals to use for signposting.
- NHS providers and primary care providers should ensure consistent aftercare plans are in place for people newly diagnosed with achalasia, including named healthcare contacts and follow-up appointments.
- Because anxiety and depression are common during the diagnostic journey, every new achalasia after-care plan should offer access to mental-health or wellbeing support (eg referral to talking therapies or specialist counselling) and signpost peer networks.

### 4. Renewing the policy agenda for rare diseases

- The Government should ensure the timely renewal of the UK Rare Diseases Framework and the reinstatement of the Rare Disease Advisory Group (RDAG). The statuses of the Framework and the RDAG are unclear due to the restructuring of NHS England and the Department of Health and Social Care. Assuming the Framework is renewed, any post-2026 strategy for rare diseases must include specific focus on improving diagnostic delay for conditions that do not have a clear genetic link, like achalasia.

## For Achalasia Action

The findings of the report also highlight actions that Achalasia Action can directly undertake to support people with achalasia. The recommendations outlined below were co-developed with steering group members and are set out as aspirations to inform future Achalasia Action strategies and activities.

### 1. Gather and share knowledge about achalasia symptoms

- Compile case studies of diagnostic journeys to share with bodies who set national training curricula for GP and gastroenterology specialisations. Case studies should outline both successful pathways to diagnosis and learning opportunities from delayed diagnoses.
- Develop a GP symptom checklist/tool for achalasia in collaboration with achalasia specialists, and primary care professionals. The symptom checklist could also help inform the introduction of achalasia flags in primary care Clinical Decision Support Systems.

### 2. Further develop educational and support resources for people with achalasia

Achalasia Action provides a variety of educational and support resources for people with achalasia on the Achalasia Action website, and has also published two information booklets: 'Achalasia Explained' and 'Living with Achalasia'. Achalasia Action will focus on further developing these resources by:

- Creating an achalasia business card with a QR code linking to comprehensive resources on the Achalasia Actions website. This portable tool would help newly diagnosed people access reliable information and explain their condition to others. The QR code would link to short, digestible explainers for friends and family, a guide for restaurants, bars and pubs to support with dietary requirements and the workplace adjustment guide (see below).
- Designing, in collaboration with people with achalasia, a workplace adjustments guide that provides information for employers on reasonable adjustments including flexible mealtimes, access to appropriate food preparation facilities and considerations for meetings.
- Further developing the existing 'Living with Achalasia' booklet to include information on how to navigate social situations that involve food and eating, and advice on addressing the anxiety and social isolation experienced by many people with achalasia.
- Producing targeted materials and resources empowering and supporting women with achalasia to effectively communicate symptoms and navigate healthcare systems where their concerns and symptoms may be disproportionately dismissed.

### 3. Establish supportive achalasia communities for people with a new achalasia diagnosis

Achalasia Action provides extensive support services, including a monitored Facebook group and Instagram page, online and in person peer-support meetings, and phone and email peer-support on a one-to-one basis. Given the findings of this report, Achalasia Action will focus on further developing its support services by:

- Developing peer-led support services and buddy systems for newly diagnosed patients, which will provide emotional support and practical guidance and information during a challenging transition period. Matching people who are on a similar journey will enable advice giving and remove isolation.
- Creating more positive and less intimidating spaces for people when they are first diagnosed, focusing on support and practical action. This will ensure that people can navigate their achalasia journey without feeling overwhelmed or scared by other peoples' negative diagnosis and treatment experiences.
- Facilitate regional support groups where people with achalasia can share experiences and practical advice for managing the condition at a local level. This will enable people who live in the same region to share recommendations about healthcare professionals and teams, support services they accessed, and meet in person to connect and support each other.

### 4. Provide self-advocacy support for people with suspected achalasia

Achalasia Action provides extensive, accurate and accessible information on achalasia symptoms and diagnosis. However, we heard that people often don't feel confident in communicating with healthcare providers about their symptoms and experiences. We will therefore focus on:

- Developing advocacy support services and resources for people who may lack the requisite confidence, language, or health literacy to navigate complex healthcare systems effectively. The support could be provided via peer-to-peer support programmes, webinars, or buddy systems.
- Creating resources for people with achalasia symptoms to support them in effectively communicating with healthcare providers about their symptoms and experiences, including templates for symptom diaries and questions to ask during appointments.

## 5. Expand equity, diversity and inclusion in achalasia support services

95% of survey respondents were white British, which could indicate a limited reach of Achalasia Action's support services with marginalised communities. We will therefore focus on:

- Developing and co-producing targeted outreach strategies to ensure that support services are accessible and inclusive for marginalised communities.
- Creating culturally-sensitive resources that address the specific needs and concerns of different communities.
- Partnering with community groups serving diverse populations to raise awareness about achalasia and provide pathways to support.
- Ensuring representation of diverse experiences and voices in all communications. This could start with representation in case studies, stories and information materials on the Achalasia Action website.

