ANNUAL REPORT 2025

ALPHA-1 FOUNDATION IRELAND



MISSION STATEMENT

Alpha-1 Foundation Ireland is a charity dedicated to raising awareness, increasing diagnosis, promoting research, and improving the treatment of Alpha-1 Antitrypsin Deficiency (Alpha-1).

VISION

That everyone with Alpha-1 in Ireland is diagnosed and receives specialist care and treatment in a timely fashion.

CORE ACTIVITIES OF ALPHA-1 FOUNDATION IRELAND



ALPHA-1 FOUNDATION IRELAND TEAM

Anne Marie O'Dowd, CEO
Dr Tomás Carroll, Chief Scientist
Ronan Heeney, Medical Scientist
Rosaleen Carroll, Alpha-1 Nurse Specialist
Dr Laith Bedri, National Alpha-1 Registry Manager

TEL: 01-809 3871 **EMAIL:** alpha1@rcsi.ie **WEB:** www.alpha1.ie

@Alpha1_Ireland Find us on Facebook

Alpha-1 Foundation Ireland Charity Code: CHY22304

ANNUAL REPORT 2025

ALPHA-1 FOUNDATION IRELAND

CONTENTS

1. Executive Summary 2
2. The National Alpha-1 Antitrypsin Deficiency Targeted Detection Programme – An Update 3
3. The National Alpha-1 Antitrypsin Deficiency Registry 6
4. Living with Alpha-1 – Cáol's Story 8
5. Recent Events 11
6. Report on the National Alpha-1 Patient and Family Conference 2025 13
7. Alpha-1 Research Highlights in 2025 14
8. Financial Report 15
9. Acknowledgements 16

1 Executive Summary



2025 was another busy year for Alpha-1 Foundation Ireland. The charity is based at RCSI Beaumont and works closely with the Alpha-1 clinical and research teams. The National Centre of Expertise for Alpha-1 is at RCSI Beaumont Hospital, expertly lead by Professor Gerry McElvaney. 308 new referrals were made to the centre following a diagnosis of Alpha-1 via the National Targeted Detection Progamme. Patients attending this clinic are offered the opportunity to participate in the National Registry for Alpha-1, particularly if they have a ZZ diagnosis – the most severe form of the condition. At the end of 2025, there were 788 people with Alpha-1 taking part in the National Alpha-1 Registry, 299 of whom had the ZZ phenotype. Having a robust and up-to-date registry is important in order to track numbers of people, which in turn informs the level of services required, but it can also provide swift access to clinical trials. In 2025, the numbers of registry participants greatly increased, helped to a huge extent by the recruitment of a full-time Registry Manager in July, Laith Bedri, who was a really important addition to the Alpha-1 team (see Chapter 3).

Notably 2025 saw the first ever Clinical Fellowship in Alpha-1 with Dr. Suzanne Roche appointed as the inaugural Fellow. This fellowship means that a senior doctor trains and specialises in Alpha-1, and becomes a future expert and leader in the field.

We know that Alpha-1 is an under-diagnosed condition so education and outreach are crucial. In 2025 visits to universities, respiratory hubs and hospitals highlighted Alpha-1 and ensured health care staff know how to order an Alpha-1 test and what to do if someone is diagnosed with Alpha-1. Chapter 2 explains who should be tested as well as the process that follows. Alpha-1 Foundation Ireland advocates for patients on a broad level – as members of the European Alliance of Alpha-1 Patient Organisations; the International Respiratory Coalition as well as through national advocacy groups in Ireland.

One of our most important events annually is the National Patient & Family Conference which took place in October in Marino Institute of Education. Patients and their families came together, with doctors and researchers, to hear about the latest clinical trials and emerging therapies. They also heard about the role of the specialist nurse in the clinic as well as ideas on how to live well with the condition. The day concluded with an amazing and inspiring presentation from a patient about her journey with Alpha-1.

Core funding from the HSE ensures the continuation of the National Detection Programme and the National Registry, without which this work would not be possible. Along with the dedicated work of the Alpha-1 teams, it moves us closer to our vision to ensure every patient with Alpha-1 receives a diagnosis and the specialist care that they need. Our work continues!

Anne Marie O'Dowd
CEO, Alpha-1 Foundation Ireland

2 The National Alpha-1 Antitrypsin Deficiency Targeted Detection Programme – An Update



Ronan Heeney, medical scientist on the national detection programme for Alpha-1 which tests over 1,200 people each year.

Alpha-1 antitrypsin deficiency (AATD or simply Alpha-1) can be diagnosed by a simple blood test but unfortunately remains hugely under-diagnosed. A diagnosis of Alpha-1 is a unique opportunity for early intervention which can prevent or postpone lung disease in both the affected individual and their relatives. In May 2004, a national targeted detection programme for AATD was launched by Alpha-1 Foundation Ireland thanks to funding from the HSE. By the end of 2025, the programme has tested more than 26,000 people.

Who Should Be Tested for Alpha-1?

World Health Organisation (WHO), American Thoracic Society (ATS), and European Respiratory Society (ERS) guidelines advocate targeted detection programmes for AATD. These guidelines recommend targeted testing of certain patient groups, with a special focus on chronic obstructive pulmonary disease (COPD)(Table 2.1).



***Figure 2.1.** Simple testing process for Alpha-1 involving a visit to a GP, blood sample, laboratory test and a final report.*

***Table 2.1.** ATS/ERS recommendations for diagnostic testing for AATD (type A recommendations).*

Who Should Be Tested?
Adults with symptomatic emphysema or COPD (regardless of age or smoking history)
Adults with asthma with airflow obstruction that is incompletely reversible after aggressive treatment with bronchodilators
Asymptomatic individuals with persistent obstruction on pulmonary function tests with identifiable risk factors (e.g. cigarette smoking, occupational exposure)
Adults with panniculitis
Siblings of individuals with Alpha-1
Individuals with unexplained liver disease, including neonates, children, and adults, particularly the elderly

How Do We Test for Alpha-1?

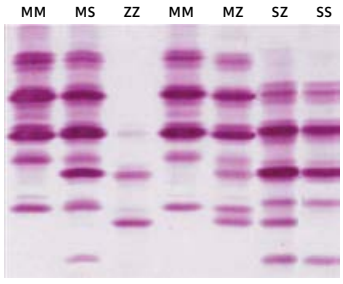


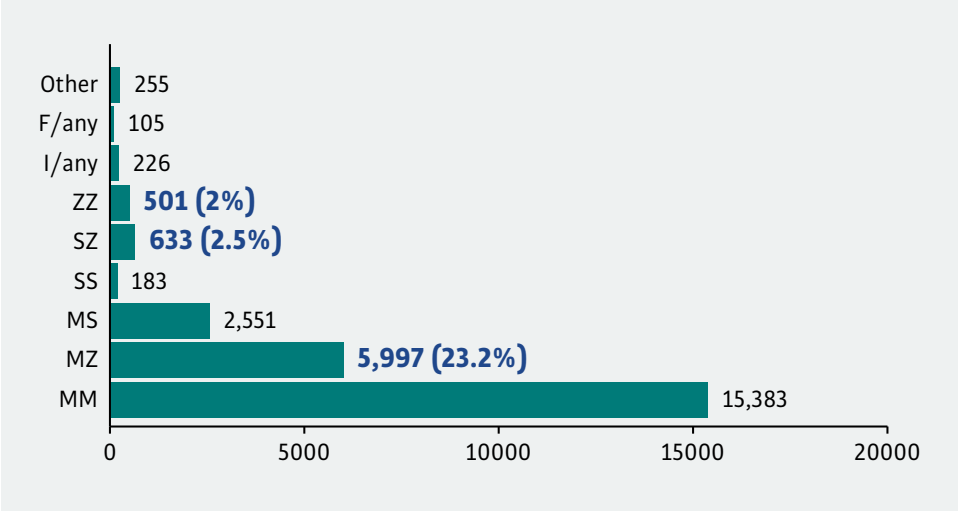
Figure 2.2. *Typical isoelectric focusing gel for AAT phenotyping with the most common phenotypes indicated.*

There are two tests needed to correctly diagnose Alpha-1. The first test measures how much alpha-1 antitrypsin (or AAT) is in the blood. The second test looks at what type of AAT protein is present by a method called isoelectric focusing with immunofixation. This method identifies types of AAT protein circulating in human blood and is more commonly known as "*phenotyping*" (Figure 2.2). It is our primary method of diagnosing Alpha-1 and can recognise most common, rare and new AAT variants.

What Have We Found in Ireland?

Since 2004, more than 26,000 individuals with COPD, asthma, and liver disease, as well as first-degree relatives of people with AATD have been tested in the HSE-funded National Targeted Detection Programme.

Figure 2.3. *Results from the National AATD Targeted Detection Programme showing the major AAT phenotypes identified among more than 26,000 individuals tested (figures to end of 2025).*



A total of **501 ZZ individuals** with severe Alpha-1 have been identified, in addition to a larger number with moderate Alpha-1, who are at risk of developing lung problems if smoking or vaping. Moderate Alpha-1 includes those with MZ (**5,997**) or SZ (**633**) forms of Alpha-1 (Figure 2.3). Other clinically significant forms of Alpha-1 have been found including **35 IZ** and **20 FZ** individuals. A number of very rare deficiency-causing AAT variants have also been identified. These include $M_{heerlen}$, M_{malton}, $M_{wurzburg}$, $N_{whitstable}$, S_{munich}, and $Z_{bristol}$ and six different Null (Q0) mutations ($Q0_{bolton}$, $Q0_{cork}$, $Q0_{dublin}$, $Q0_{lisbon}$, $Q0_{porto}$ and $Q0_{amersfoort}$). **$Q0_{cork}$ and $Q0_{dublin}$** were discovered for the first time here in Ireland.

Table 2.2. *Simple explanation of the most common AAT phenotypes.*

AAT Phenotype/ AAT Genotype*	AAT Deficiency?	What does it mean?
MM	No	Does not have the disorder – has two normal copies of the AAT gene.
MS	Mild	No evidence of increased risk of lung or liver disease but does carry one altered AAT gene.
MZ	Moderate	Significantly increased risk of lung disease **in smokers**. Increased risk of liver disease.
SS	Moderate	Presumed increased risk of lung disease **in smokers**. No evidence for increased risk of liver disease.
SZ	Moderate	Significantly increased risk of lung disease **in smokers**. Increased risk of liver disease.
ZZ	Severe	Significantly increased risk of lung disease in **smokers and ever smokers**. Increased risk of liver disease.

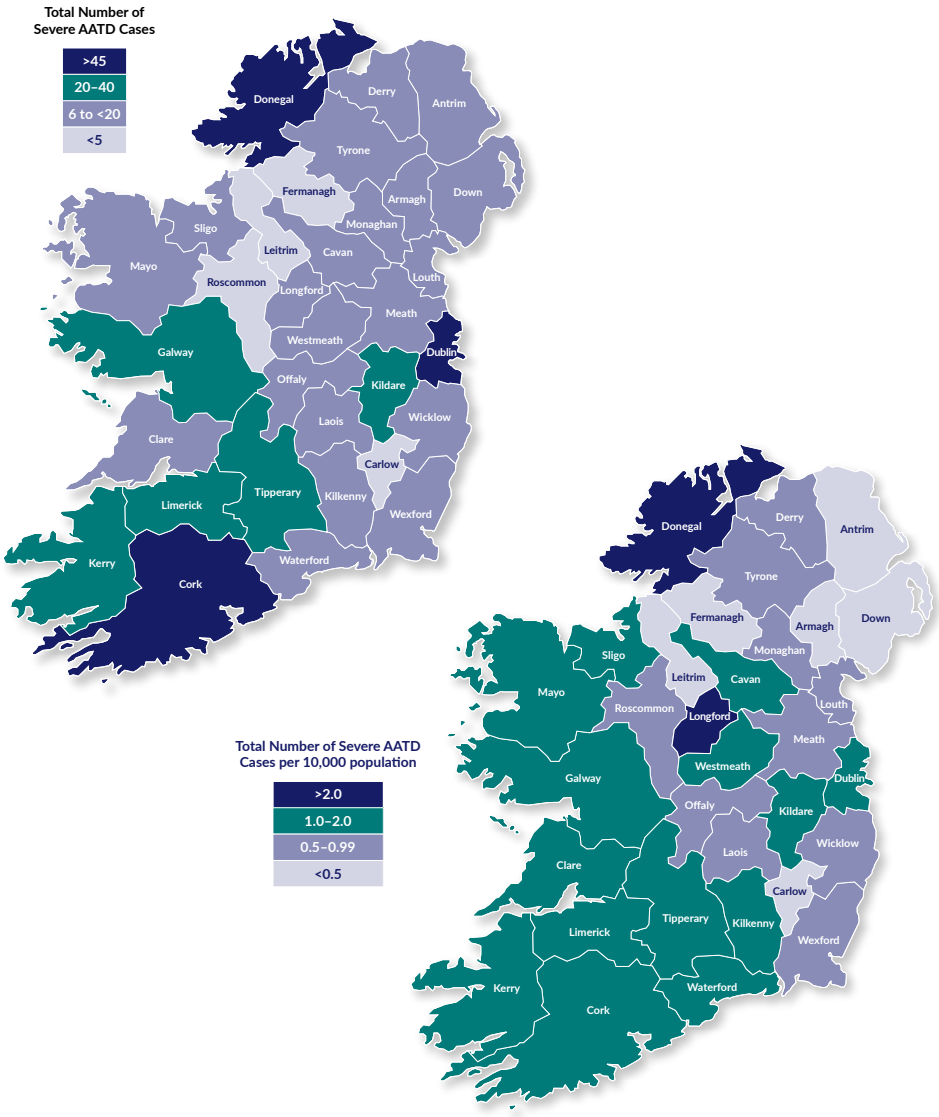
The goal of the national detection programme is to ensure people with Alpha-1 are correctly diagnosed and are given the opportunity to receive expert medical care, advice, and support. People with Alpha-1 can be referred to the **National Centre of Expertise for AATD in RCSI Beaumont Hospital** under the care of **Professor Gerry McElvaney**. A strong focus on family screening can help diagnose other family members with Alpha-1,

which can help prevent or postpone the development of serious health problems in the wider family. Importantly, a correct diagnosis means people can benefit from lifestyle changes such as:

- stopping smoking and vaping
- reducing risks in the workplace (such as fumes, gases, or dusts)
- receiving specialist medical care from Alpha-1 experts
- getting opportunities to enrol in clinical trials that test new treatments

Figure 2.4.
LEFT: *Distribution of severe AATD cases detected to date (600 cases across 32 counties – this includes people diagnosed outside of the TDP and outside of Ireland).*

RIGHT: *Distribution of severe AATD cases adjusted per 10,000 population.*



Alpha-1 Education and Outreach in 2025

Education and raising awareness of AATD is one of our core activities. We held seminars on Alpha-1 to a mixture of respiratory and laboratory teams in Mullingar, Portiuncula (Galway), and Beaumont Hospitals. We also presented to medical students in UCD, nursing students in DCU, and M.Sc. students in RCSI. We presented research on Alpha-1 at the 16th Jenner Glycobiology and Medicine Symposium, the Irish Thoracic Society conference, and the European Respiratory Society conference. The aim of these presentations is to increase awareness of Alpha-1 among healthcare professionals and to encourage testing. While respiratory (and liver) medical teams care for patients most at risk due to AATD, hospital laboratories measure alpha-1 antitrypsin levels during normal blood investigations and thus help reach a diagnosis of AATD.

3 The National Alpha-1 Antitrypsin Deficiency Registry

What is the Alpha-1 registry?

The registry is a confidential database that stores relevant medical information of individuals diagnosed with alpha-1 antitrypsin deficiency (AATD). Results of tests that monitor lung and liver function such as blood tests, breathing tests, CT scans of the lungs and ultrasound or transient elastography (also called a Fibroscan) of the liver are recorded in the registry and updated at regular intervals. The registry also collects other information relevant to AATD. For example, smoking status and occupation can help to quickly identify risks to lung health (e.g. a factory worker exposed to fumes or dusts).

Is my data safe?

Yes. The registry is located on a secure server, hosted by Health Atlas Ireland, a key part of the HSE digital landscape. All data is encrypted and only staff on Professor McElvaney's team at RCSI Beaumont Hospital have access to the registry.



What is the purpose of the registry?

The registry helps to deepen our knowledge and understanding of AATD, improve the clinical care given to people with AATD, and increase recruitment for clinical trials investigating new treatments for AATD. The small number of people affected by a rare condition like AATD means that getting access to the right care, treatment and support can be difficult. Registries play an important role in filling gaps that exist in the care of people with rare disease, particularly in Ireland which has no electronic healthcare record.



Dr Laith Bedri, National Alpha-1 Registry Manager

What is the role of the National AATD Registry Manager?

As registry manager, my role was to oversee registry activity that is aimed at improving diagnosis, treatment and care of patients with AATD. On a day-to-day basis, I collaborate closely with clinicians and researchers to strengthen Ireland's role in global Alpha-1 research. After consenting patients to be a part of the registry, I analyse a range of clinical data. This includes special breathing tests, radiologic findings and patient-reported outcome measures. I collect and enter clinical data into the registry. This helps the Alpha-1 community as the registry drives research that can improve our understanding of how Alpha-1 affects people – so we can learn how to better prevent the health issues people face. The most rewarding part of my experience was to be able to interact with AATD patients in both the hospital setting and at the annual Alpha-1 Patient and Family Conference. Hearing patients share their experiences, concerns, and hopes reminded me of the importance of approaching research not simply as an exercise in analysing data, but as a means of improving the lives of the people behind that data.

What are the key objectives of the Registry?

1. Increase our understanding of AATD (knowledge)
2. Inform and improve clinical care (care)
3. Provide early access to new treatments via clinical trials (treatment)

How can I enrol?

Individuals diagnosed with various forms of AATD are eligible to enrol in the registry. Enrolment is completely voluntary, and an individual must provide their written informed consent prior to enrolment. A member of Alpha-1 Foundation Ireland will provide individuals with an information leaflet and answer any questions at the time of enrolment.

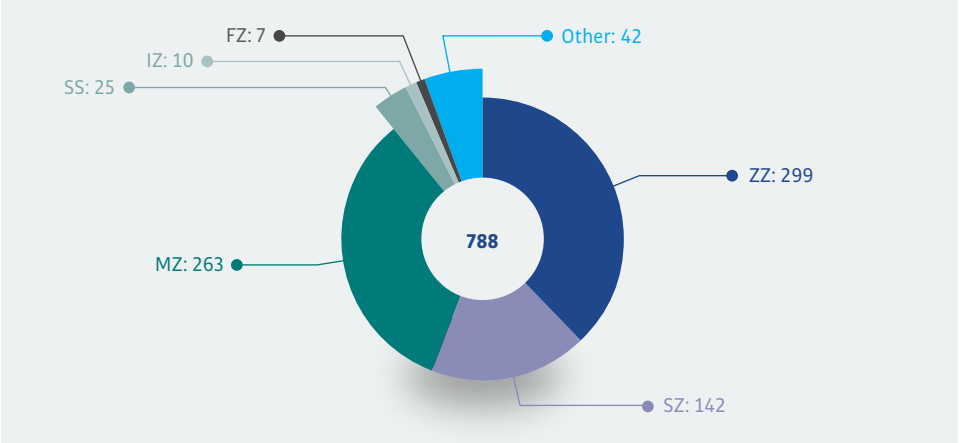
If I have questions?

If you or your family are interested in enrolling or have any questions about the registry, please contact us on 01-8093871. To learn more about our registry visit our website at *https://www.alpha1.ie/irish-alpha-1-registry/*.

Registry Update and Future Plans

At the end of 2025 a total of **788 people with Alpha-1 were taking part in the National AATD Registry** (Figure 3.2) and this number is growing all the time. Joining the registry allows us to build knowledge, improve care and help develop new treatments through involvement in clinical trials.

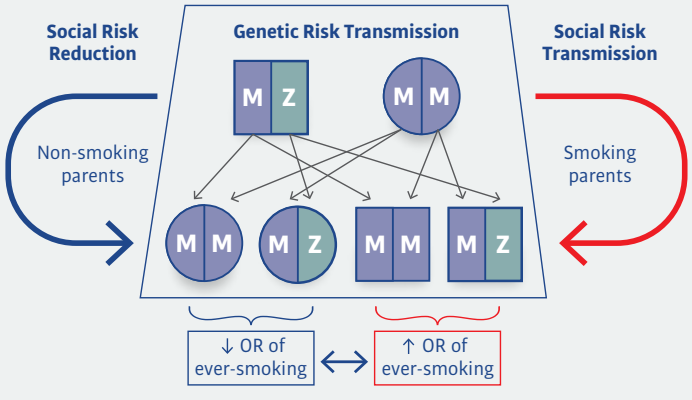
Figure 3.2. *Total number of individuals enrolled in the registry according to AATD phenotype* (**n = 788**).



Research using anonymous data from the registry improves our understanding of the health problems caused by Alpha-1. For example, an Irish study looked at cigarette smoking habits in people with Alpha-1 taking part in the registry. The study [**Alpha-1 Antitrypsin Deficiency and Tobacco Smoking: Exploring Risk Factors and Smoking Cessation in a Registry Population, Franciosi et al, Journal of COPD, 2021**] showed smokers were highly motivated to stop smoking after finding out they had Alpha-1. In addition, people with 1 or 2 parents who smoked were more likely to become smokers themselves (see Figure 3.3). This valuable information led to a greater focus on smoking cessation at the National Centre of Expertise for AATD to prevent lung disease in **current and future generations** within families affected by Alpha-1. The findings highlight the importance of an early diagnosis of all types of AATD so positive lifestyle choices (like stopping smoking and/or vaping) can be made.

Another key study made possible by the National AATD Registry was a three-country study of severe Alpha-1 published in late 2023 [**Augmentation Therapy for Severe Alpha-1 Antitrypsin Deficiency Improves Survival and Is Decoupled from Spirometric Decline-A Multinational Registry Analysis. Fraughen et al, Am J Crit Care Med, 2023**]. This study looked at the health of 615 people with severe AATD in European AATD registries in Ireland, Switzerland, and Austria; three countries where access to standard medical care is equal but access to augmentation therapy (intravenous AAT) is not. This allowed us to test the effects of augmentation therapy on survival. Our study showed people with AATD in Switzerland and Austria benefit from a clear survival advantage due to augmentation therapy. We also showed that a majority of people with AATD are being diagnosed too late by which time severe lung disease has already occurred.

Figure 3.3. *An example of the knowledge generated from a survey of smoking habits in people taking part in the National AATD Registry. The diagram shows how 2 risk factors are inherited in families with a parent who smokes; the genetic risk of Alpha-1 (the Z variant) and the habit of smoking.*



4 Living with Alpha-1 – Cáol's Story



Life can change in an instant as they say, and we never know what is around the corner. Your health is your wealth, but do we really pay attention to these sayings? Only when it comes to your own door do we suddenly pay attention and get our eyes open. Our lives can be transformed from what was the normal everyday run of the mill, monotony that we call life. Sickness, bereavement, loss of financial stability, or just downright bad luck are some of the things that can affect the course of our lives. On the upside positivity can also befall us, the perfect job bringing extra financial gain, good medical news, good luck landing at our door, or the birth of the newest addition to our families. The point is life throws so much at us as we meander our pathways from birth to death. We cannot change a lot of it but adapting to change and working at issues and problems, with support and belief we can get to where we want to be or at least better a situation. Every cloud has a silver lining; we must make the most of any situation we find ourselves in. This is the story of our situation that landed at our door, how we coped as a family, adapted and made life choices that looking back have been beneficial for us all.

The second of October 2010 my wife delivered our fourth child a boy, with his older brother and two older sisters coming to visit the excitement was immense. They were allowed home that evening and he was not being let settle at all, being spoilt and surrounded with love and affection. This would soon settle down as we got adjusted to the newest family member and the reality that everyday life continues as normal. All was well, all were happy, but unknown to us something was not right, and our lives were to be turned upside down as the child became ill. The baby was not himself, crying and not feeding and his mum realised something was not right. He was only a few days old, but weaker than he should be and sick looking. I took him to our family doctor in Letterkenny about 20 kilometres from where we lived in a rural area. I had phoned the surgery beforehand and was advised to come straight in. Looking back to 2010 it was a lot easier to access a doctor or gain an appointment than it is today.

The journey did not take long, about 20 minutes, the child wrapped up snugly in his car seat, looking back he was hard to see being so small. I entered the surgery and was sent to sit outside the doctor's door. That day our family G.P. was not in so I was directed to another doctor in the practice. I was called in and started to remove blankets and clothing from the wee bundle of life that lay in my arms. The doctor examined the child and was asking me questions as she did, about his journey from birth to the surgery he was now being examined in, and I gave as much information as I could. Then her professional opinion she shared, "Alpha-1 is a possibility", what is that I enquired? And she briefly explained this new term that I never had heard of before. Our fourth child was only a few days old, losing weight due to not feeding from his mum. Jaundiced as many babies can be but the whites of his eyes were almost orange and he was very thin. He was very sick, she said he must go into the hospital immediately. I said I will go collect his mum and we will take him in, the doctor was adamant, "No, he is going straight to the hospital". His little journey into the world of Alpha-1 Antitrypsin Deficiency had begun.

I left the surgery not fully understanding the situation and made my way to Letterkenny University Hospital five minutes up the road, it seemed longer as I sat in traffic and held my son's wee hand, having our first man to man talk, "I told him all will be fine, daddy is with you". I called my wife from the hospital carpark and explained what was going on, through her tears

and panic she had a thousand questions that I could not answer, the other children were at home and the baby's mum needed to be at the hospital asap. A few phone calls to family and all was sorted, and soon a worried mum was at his bedside in a small isolation room in paediatrics. Tests were carried out and examinations took placed, a busy room as doctors and nurses went about their routines all communicating with each other, as we the parents seemed to be stuck in a vacuum. Word was soon sent from Beaumont Hospital as results were analysed there, that the Letterkenny baby must be transferred immediately to Our Lady's Hospital for Sick Children in Crumlin, Dublin. Preparations were put in place for us to go to Crumlin, as I organised things at home, but delay after delay held the transfer back a few days, with Crumlin Hospital on the phone daily demanding the child be brought down, but now on a Saturday no ambulances were available from transportation. On talking to the consultant in Crumlin he had a plan B, I was given a letter from Letterkenny, and we took the child in our own car the unknown journey to Crumlin. His worried mum nursing him in the back, trying to feed him, trying to comfort him, he was getting weaker, would he make it to Dublin. I kept reassuring her he would be fine, she was worn out with worry and lack of sleep herself, staring at the child and sometimes the blank stares out the window of the car, she did not want to stop for a break, just keep going on this unknown journey to the capital.

Crumlin was reached eventually and the child was whisked away for tests as we were shown to another small isolation ward in the hospital and reassured the child would be brought back soon. And he was soon placed back in the arms of his anxious mother, as a consultant introduced himself and tried to explain the situation a bit better for us. "This child is very ill but we endeavour to give him the best fighting chance we can", I never forgot those words as he spoke them with compassion but also with belief, I now for the first time thought honestly to myself than he will make it, as all along I was preparing for the worst, and hiding my true feelings from my wife, trying to shield her from the possibility that we may lose our boy, holding out hope that a miracle will happen and all will be good. We both settled down to relax for the first time in a few days, as the child was placed on a drip and given fluids to try and boost his strength. A bed was placed in the room for my wife to spend with the baby as I went in search of accommodation within the hospital, as parents' rooms were available. Once this was sourced, I needed to go outside, my first cigarette in hours and I had more than one as I needed to get back upstairs. I found my wife asleep on a chair nursing the child, she was exhausted. I took the baby and placed him in his small cot, put a blanket over my wife. Then I lifted the baby and sat down, we needed another man to man.

The doctors and nurses came in at regular intervals and checked his condition throughout the night. Over the next few days several tests were carried out to get the diagnosis right, and to plan for his road ahead. We were still trying to get our heads around this Alpha-1 thing; Google did not help and there was little or no literature available. My wife was worried sick. Not taking care of herself just worried about the baby and the other children at home. I accompanied him for all his tests throughout the hospital and spoke to the doctors and consultants, I still did not understand what this new term meant but just kept reassuring my wife. Between all this I was regularly going outside for a smoke, I was at that time on about 60-80 cigarettes a day - a heavy smoker not that I realised. I met two guys from Letterkenny who had children in the hospital also, we met for a cigarette and a conversation several times a day, our little piece of comfort in the monotony of hospital life. I had an appointment with a consultant one morning, so I met one of the guys for a cigarette beforehand, I opened a new packet of 20 John Player Blue and took one and offered him one, lighting them up with my new red lighter. He said he would wait until I came back from the meeting outside the hospital, a bit of moral support if needed.

The meeting opened my eyes to the dangers of smoking that I always ignored, not just my health but the health of my now confirmed Alpha-1 baby, my wife and other children. "Smoking will kill the baby or have very serious consequences", the consultant explained. And he then went on to explain what smoking does to a lung, eroding the lining and damaging the sacs, making it difficult to breathe and causing serious issues that can be fatal. He explained the scenario as we had a coffee and he used a Chunky Aero chocolate bar as a prop. Breaking the chocolate bar in half showing me the small air holes in the bar that makes it famous, comparing these to the sacs in a lung if dissected, a simple way to explain to a non-medical person what a lung looks like if cut open. He continued about the dangers of smoking and fumes as he held his Chunky Aero over the warm cup of coffee in front of him. Then he played his Ace card he showed me the Aero again, now partially melted from the heat of the coffee, the holes almost all gone, just a few visible. He said this is what a smokers lung looks like, damaged from smoke, very few working air sacs, now try and breathe using a lung like that. I had just been given a masterclass in why I needed to kick this habit, not just for me, but for my family.

I walked down the stairs and outside to my friend who was standing waiting, he offered me a cigarette and I said "NO". I handed him the remaining 18 John Player Blue and the red lighter and said I am going off them. That was almost 16 years ago, I never smoked again, changed for the better and never looked back. I had a reason to quit, he was seriously ill upstairs in his mother's arms, I made a choice to sacrifice something to give him at least a fighting chance. I got into running and keeping fit, my wife also is a runner now, I have done 16 marathons, countless half marathons, 10 milers, 10k and 5k races. I run a couch to 5k for several years to get people fit, I got my eyes open by a melting Chunky Aero and a consultant who thought outside the box when looking to explain Alpha-1 and smoking. I had to get our other children tested, one was a carrier, one had no symptoms, the older girl like her newborn brother turned out to be ZZ. They live normal lives, diagnosed young, kept on the right path physically and with fantastic care in Letterkenny, Crumlin and now Beaumont down through the years, the future is a lot brighter than it was 16 years ago as I smoked a cigarette outside Crumlin Hospital before meeting a consultant, that not alone changed my life, but that of my family. I didn't know what was around the corner, it did land at our door, but we adapted to the necessary changes, we live with the condition, we sacrificed some small things along the way, but if I was still smoking the way I was, I would be very ill today if here at all. My little boy and I still have our man to man talks, the consultant looked up at him the last time he was in Crumlin and smiled, "Look at the size of him now, from laying ill in my hand 16 years ago, to standing looking down at me". He said he was a withered up little baby, and very ill, as he held him in one hand, he did reassure me that he would do his best, he would give him a fighting chance, he did that, and thanks to his input, we are all here today as a family. My son he will get on fine, he plays GAA, tries his best and will get on in life. He is a blessing like his siblings; we are lucky to have them and grateful every day. Many a child will never see the outside of a hospital or live a normal life. Many a parent has only a grave of their child to visit, this must be so painful. Your health is your wealth, family is everything.





5 Recent Events

Thirty Years of Alpha-1 Foundation in America

On January 31st our friends in the US Alpha-1 Foundation celebrated their 30th birthday. The organization was founded by three Alpha-1 patients, including Irish-American John Walsh (RIP) who was a big supporter of Alpha-1 Foundation Ireland.



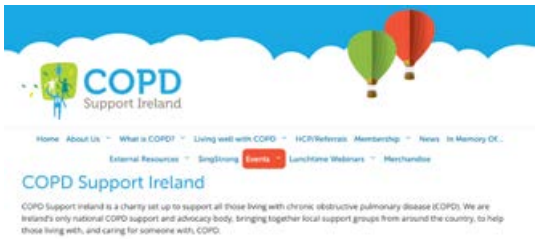
Lecture at UCD Medical School

In February we visited University College Dublin to discuss Alpha-1 with third year medical students taking the excellent rare disorders module. Running since 2012 this innovative lecture series showcases rare conditions like Alpha-1. Thanks to Paula Byrne for the invitation to raise Alpha-1 awareness among the next generation of doctors.



Teaching at Mullingar and Galway Hospitals

In March we visited Midlands Regional Hospital in Mullingar to talk about Alpha-1. Thanks to Norma Mullen and colleagues for the warm welcome at Grand Rounds. In April we met staff working at Portiuncula University Hospital (PUH). The PUH team serves Roscommon and East Galway, providing care to people with lung disease in hospital and in a community hub.



Carlow & Kilkenny COPD Support Groups

In May we visited both Carlow COPD Support and Kilkenny COPD Support groups. To find out if there is a COPD group in your area, visit *www.copd.ie*.



500th person with ZZ Alpha-1 Diagnosed

In June the 500th person with ZZ Alpha-1 was diagnosed by the national AATD targeted detection programme, which is based at RCSI Beaumont Hospital and co-ordinated by Alpha-1 Foundation Ireland.



Alpha-1 Patient Speaks at Prestigious Scientific Conference

Later in June Orla Keane spoke at the 16th Jenner Glycobiology and Medicine Symposium in Maynooth University. Orla provided a brilliant patient perspective on living with Alpha-1 since her diagnosis in 1996.



Alpha-1 visits DCU School of Nursing

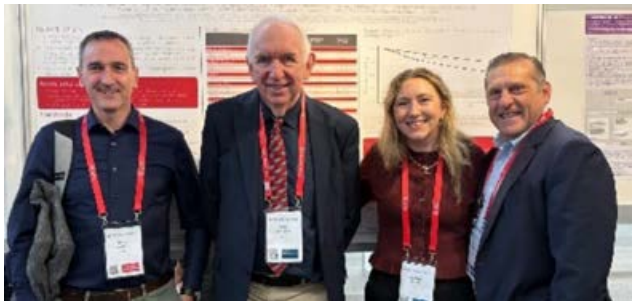
In September, Alpha-1 Foundation Ireland visited DCU to talk about Alpha-1 with the large 2nd year nursing class. Thanks to Dr. Ciara White & Dr. Hazel Ní Chonchubhair for this valuable educational opportunity.





Alpha-1 Marks World Lung Day

In late September our CEO Anne Marie O'Dowd and registry manager Laith Bedri joined the Irish Lung Health Alliance to talk to our political leaders about lung health and Alpha-1. Thanks to Senator Evanne Ní Chuilinn and Deputy Jerry Buttimer for their support.



Irish Alpha-1 Research Presented at European Respiratory Society (ERS) Conference

In late September Dr. Suzanne Roche presented her research at the ERS conference in Amsterdam, pictured here with Dr. Tomás Carroll, Professor Gerry McElvaney and Scott Santarella. Thanks to Scott Santarella and Alpha-1 Foundation USA for their work in driving awareness of and research into Alpha-1 for the benefit of Alphas all around the world. Ní neart go cur le chéile!



Alpha-1 Patient and Family Conference 2025

In October the Alpha-1 Patient and Family Conference in Dublin saw over 60 people with Alpha-1 gather to learn all about the latest developments in Alpha-1.



Alpha-1 Foundation Ireland Attends the National Lung Meeting in Galway

In November, we attended the Irish Thoracic Society annual conference in Galway and hosted an Alpha-1 information table. We welcomed Dr. Gianluca Imeri, an Alpha-1 specialist from Bergamo in Italy, who won a prestigious Maurizio Luisetti fellowship to visit the National Centre of Expertise for Alpha-1 at RCSI Beaumont Hospital. Gianluca is pictured here with Dr. Tomás Carroll, Anne Marie O'Dowd and Dr. Laith Bedri from Alpha-1 Foundation Ireland.

6 Report on the National Alpha-1 Patient and Family Conference 2025

The 2025 Alpha-1 Patient & Family Conference took place on Saturday 18th October in the Marino Institute of Education in Glasnevin in Dublin.



Anne Marie O'Dowd, CEO of Alpha-1 Foundation Ireland welcomed everyone before Alpha-1 Board Member Michelle Hughes introduced the first speaker, Professor Gerry McElvaney. He talked about the importance of smoking cessation, flu vaccines, pulmonary rehabilitation, standard COPD medications, and monitoring for signs of liver disease. Gerry described the exciting new gene therapies being developed with potential to cure Alpha-1 and how some would be tested in clinical trials at Beaumont Hospital.

Dr. Emma Farrell was the winner of the 2024 Alpha-1 Prize for Best Communication of Research to a Lay Audience. Emma spoke about her unique ZZ Family Study which aims to learn how much more likely a person is to develop lung or liver disease with Alpha-1; the risk smoking poses; and whether stopping smoking slows or stops the decline of lung function.



Next, five young researchers working on Alpha-1 at the Irish Centre for Genetic Lung Disease in RCSI Beaumont Hospital spoke about their work in 3 minute "lightning" talks chaired by Dr. Mark Murphy. The audience voted Dr. Suzanne Roche and her talk on genetic discrimination in Alpha-1 as the winner of this young researcher competition. The last session before lunch saw Dr. Tomás Carroll give an update on the National Targeted Detection Programme and the National Alpha-1 Registry.



After lunch, Rosaleen Carroll gave a presentation about her role as the specialist nurse for Alpha-1. Putting the person with Alpha-1 at the centre, the role involves co-ordinating care, education, addressing physical and psychological need, and supporting the wider family. Rosie noted that Alpha-1 Foundation was developing a leaflet in relation to mental health which would provide signposting to resources.



To end the day Orla Keane gave a powerful talk about her journey with Alpha-1 over the past 35 years and how she tries "to live well with Alpha-1". She described the shock felt when first diagnosed, and her journey since then including advocacy, fundraising, multiple Alpha-1 awareness events, presentations at scientific conferences, and participation in an important international clinical trial. Orla has been a great supporter of Alpha-1 Foundation Ireland since her diagnosis – and is always willing to help others affected by Alpha-1. Her uplifting talk received a standing ovation.

The final talk was by Dr Jennifer Wilson O'Raghallaigh, Clinical Psychologist, HSE and Carol Conway. They described a new HSE programme which provides classes for adults with chronic illnesses to live well and self-manage their condition, with another programme for care givers. In closing remarks, Anne Marie O'Dowd thanked everyone for coming and contributing to such an excellent conference.

7 Alpha-1 Research Highlights in 2025



Kate Walley, RCSI medical student

Evaluating Lung and Liver Health in ZZ Alpha-1 Antitrypsin Deficiency

Kate Walley, an RCSI medical student, carried out a 6-week research project with Alpha-1 Foundation Ireland in early 2025. Kate looked at the prevalence and severity of lung and liver disease in ZZ Alpha-1 by analysing health information stored on the National AATD Registry. When people with Alpha-1 join the registry, they complete a questionnaire that captures important information such as occupation and smoking or vaping history, as these are risk factors for lung disease.

279 baseline lung function (breathing) tests and 141 liver scans from 279 individuals with severe AATD (majority ZZ) were studied. The average lung function (a measure known as FEV1 %) was 56% in ever smokers, compared to 95% in never smokers. Notably, the small number of active smokers (**10/279 or just 3.6%**) shows a strong quit smoking effect. This registry analysis reveals the serious impact of smoking on the lungs of people with severe Alpha-1. Kate also found many with significant liver disease in this cohort, underscoring the need for regular liver as well as lung checks in AATD. Kate presented her work at the prestigious ICHAMS student conference and won the prize for best oral presentation!

Irish Alpha-1 Research Published in 2025

1. Advancing the understanding and treatment of lung pathologies associated with alpha 1 antitrypsin deficiency. Turner AM, Ficker JH, Vianello A, Clarenbach CF, Janciauskiene S, Chorostowska-Wynimko J, Stolk J, **McElvaney NG**. Ther Adv Respir Dis. 2025 Jan-Dec;19:17534666251318841. PMID: 39980299.
2. Longitudinal Evaluation of Individuals With Severe Alpha-1 Antitrypsin Deficiency (Pi*ZZ Genotype). Fromme M, Turner AM, **McElvaney NG**, Strnad P, et al. Gastroenterology. 2025 Feb;168(2):367-381. doi: 10.1053/j.gastro.2024.10.010. Epub 2024 Oct 15. PMID: 39414159.
3. Alpha-1 Biomarker Consortium Study Group. Rationale and Design of the Alpha-1 Biomarkers Consortium Study. Goldklang MP, Pirozzi C, Barjaktarevic I, Bhatt SP, Bodduluri S, Drummond MB, Fonseca L, Hogarth DK, Keaveny A, Liu Z, **McElvaney NG, McElvaney OJ**, Nuchovich N, Palumbo S, Plant R, Sandhaus R, Wells JM, Wilson A, Strange C, D'Armiento JM. Chronic Obstr Pulm Dis. 2025 Jul 30;12(4):274-284. PMID: 40273316.
4. Mendelian causes of early-onset emphysema: a review of the current literature. Karabatic A, van den Berge M, Carroll TP, Guryev V, Faiz A. Eur Respir Rev. 2025 Nov 12;34(178):250125. PMID: 41224373.
5. Seasonal Changes in Circulating Alpha-1 Antitrypsin Are Altered by Sex and Genetic Deficiency. **Leacy EJ, Fraughen DD, Heeney RC, Casey M, Sherlock M, O'Reilly MW, Murphy MP, Curtis AM, Carroll TP, McElvaney NG**. Am J Respir Crit Care Med. 2025 Dec;211(12):2408-2410. PMID: 40961267.

8

Financial Report

Alpha-1 Foundation Ireland
(A Company Limited by Guarantee and not having Share Capital)
Financial Statement
Financial year ended 31 December 2025

	2025	2024
	€	€
TURNOVER		
Northern Area HSE	136,654	119,568
Donations	11,722	8,424
Other Funding	10,167	2,361
Charity VAT Compensation scheme	127	651
Gross profit	**158,670**	**131,004**
Gross profit percentage	**100.0%**	**100.0%**
OVERHEADS		
Administrative expenses	(161,586)	(156,043)
Operating (loss)/profit	(2,916)	(25,039)
Operating (loss)/profit percentage	1.8%	19.1%
(Loss)/profit before taxation	(2,916)	(25,039)
Overheads		
Administrative expenses		
Wages and salaries	(88,981)	(78,705)
Lab costs	(14,260)	(11,827)
Conference costs	(2,807)	(2,594)
Printing, postage and stationery	-	(42)
Annual Report	(2,988)	(2,988)
Health Professional Education Programmes	-	(4,000)
Telephone	(180)	(180)
Registry and detection programme	(39,207)	(41,694)
Education and outreach	(1,380)	(1,564)
Accountancy fees	(6,150)	(6,150)
Bank charges	(111)	(99)
General expenses	1	(197)
Charitable Donations – Type 1	(5)	-
Subscriptions	(1,455)	(1,940)
Depreciation of tangible assets	(4,063)	(4,063)
	(161,586)	(156,043)

NOTE: Detailed financial statements are available on *www.alpha1.ie*



Acknowledgements

We would like to thank the following:

- Orla Cahalane, Ella Howard, Jack Kennedy, and Sharon Madziyaand in the Beaumont Hospital Chemical Pathology Department for their continued support and assistance
- Angela McBride, Scott Santarella and colleagues from the Alpha-1 Foundation (USA) for their continued support
- Dr. Ilaria Ferrarotti and Dr. Stefania Ottaviani, Centre for Diagnosis of Inherited Alpha-1 Antitrypsin Deficiency, University of Pavia, Italy
- Louise Clarke, Lucinda Feeney and colleagues in the Pulmonary Function Laboratory in Beaumont Hospital
- Jean Kirwan and Linda Phoenix, members of the Beaumont Hospital Respiratory Clinic Administration Team
- RCSI University of Medicine and Health Sciences for their continued support
- Kate Walley, RCSI medical student who carried out a research project with Alpha-1 Foundation Ireland in 2025
- Health Research Charities Ireland (HRCI), the Irish Platform for Patients' Organisations, Science & Industry (IPPOSI), the Irish Lung Health Alliance, and the Irish Thoracic Society (ITS)
- Austin Butler for graphic design
- A special thank you to everyone who took part in or organised awareness and fundraising events throughout the year

We would also like to thank the Department of Health and Children and the Health Service Executive for their continued financial support.

We would also like to acknowledge the participation of the following hospitals;

- Beaumont Hospital
- Blackrock Clinic
- Bon Secours Hospital Tralee
- Bon Secours Hospital Dublin
- Cavan General Hospital
- Children's University Hospital, Temple Street, Dublin
- Coombe Women and Infants University Hospital
- Cork University Hospital
- James Connolly Memorial Hospital Blanchardstown
- Letterkenny University Hospital
- Mater Misericordiae University Hospital, Dublin
- Mayo University Hospital
- Midland Regional Hospitals Tullamore, Mullingar, and Portlaoise
- Naas General Hospital
- Our Lady's Children's Hospital, Crumlin
- Our Lady of Lourdes Hospital, Drogheda
- Our Lady's Hospital, Navan
- Peamount Hospital, Dublin
- Rotunda Hospital, Dublin
- Sligo University Hospital
- St. James's Hospital, Dublin
- St. Luke's General Hospital Carlow/Kilkenny
- St. Vincent's University Hospital, Dublin
- South Tipperary General Hospital, Clonmel
- Tallaght University Hospital
- University Hospital Galway
- University Hospital Kerry
- University Hospital Limerick
- University Hospital Waterford
- Wexford General Hospital



Téarmaí Tábhachtacha as Gaeilge

Toitín	cigarette
Scamhóga	lungs
Galar scamhóige	lung disease
Galar scamhóige toirmeascach ainsealach	COPD
Broincíteas	bronchitis
Ae	liver
Dhúchasach	hereditary
Uireasa	deficiency
Uireasa antaitripsin alfa-1	AATD
Cóir leighis	treatments
Plúchadh	asthma
Feadán anála	airways
Géineolaíocht	genetics
Géinte	gene
Géiniteach	genetic
Scagthástáil gaolta	family screening
Éagsúlacht ghéinteach	genetic variation
Triail chliniciúil	clinical trial
Buíochán	jaundice
Caitheamh fulangach	passive smoking
Cársán	wheezing
Casacht	cough
Airí	symptom
Eimfiséime	emphysema

TEL: 01-809 3871 **EMAIL:** alpha1@rcsi.ie **WEB:** www.alpha1.ie

@Alpha1_Ireland

Find us on Facebook

Alpha-1 Foundation Ireland Charity Code: CHY22304

Alpha-1 is the most common genetic cause of COPD



1 in 25 people in Ireland carry the faulty Z Alpha-1 gene



Smokers with a single faulty Z Alpha-1 gene have a **10 fold** increased risk of developing a lung disease called COPD



The Irish Thoracic Society estimates approximately **380,000** people have COPD in Ireland



26,000+ people tested for Alpha-1 to date in a National Targeted Detection Programme



1,200+ people with Alpha-1 attend the National Centre of Expertise for Alpha-1 at Beaumont Hospital



900+ people with Alpha-1 take part in the National Alpha-1 Registry