



ANNUAL REPORT 2017

 Alpha One
Foundation

MISSION STATEMENT

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

CORE ACTIVITIES OF THE ALPHA ONE FOUNDATION



STAFF OF THE ALPHA ONE FOUNDATION:

Ms Geraldine Kelly, CEO

Dr Tomás Carroll, Chief Scientist

Ms Laura Fee, Clinical Research Associate

Ms Margaret Molloy, Research Nurse and Registry Coordinator

Professor Gerry McElvaney, Chairman

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 [@Alpha1_Ireland](https://twitter.com/Alpha1_Ireland)

Alpha One Foundation (Ireland) Charity Code: CHY14812

Contents

1. Executive Summary	2
2. Update from the National Alpha-1 Antitrypsin Deficiency Targeted Detection Programme.....	3
3. The New National Alpha-1 Antitrypsin Deficiency Registry.....	5
4. Meet the Alpha-1 Researchers	7
5. Recent Events	8
6. Sixth Global Alpha-1 Patient Congress	10
7. Current Research Developments in Alpha-1	11
8. Acknowledgements	13

This year's Annual Report is dedicated to the memory of John W. Walsh who passed away earlier this year. Founder and CEO of the US Alpha-1 Foundation, John was a tremendous friend to the Alpha-1 Community in Ireland and an inspiration to all those lucky enough to meet him. Ar dheis Dé go raibh a anam dílis.

Executive Summary

It has been a very busy 12 months for the Alpha One Foundation. While our main activity continues to be the National Alpha-1 Screening Programme, which has tested over 18,000 Irish people, and the National Alpha-1 Clinic, this year we have also focused on two additional priorities. Firstly the campaign for the reimbursement of augmentation therapy (Respreeza) to treat emphysema caused by the severe form of Alpha-1, secondly the re-development of the National Alpha-1 Registry.

In August of this year we received notification from the HSE that the reimbursement of Respreeza had not been approved. The refusal to reimburse the therapy for over 60 people was a huge blow for the patients, their families, the clinicians who take care of them and for the Alpha One Foundation. Thank you to everyone who helped with political briefings, demonstrations, and numerous media interviews in the campaign to date. A programme has been established in Beaumont Hospital to monitor the patients who have ceased the medication, with a view to addressing any health concerns they may experience, to help provide valuable data to the clinicians and to assist with the continuing campaign for reimbursement of the drug. We are still hopeful that a solution can be found.

We continue to develop and populate our National Alpha-1 Registry which is diligently supported and managed by Margaret Molloy. The number of patients now enrolled in our registry is in excess of 400. We will continue to grow this number over the coming 12 months. This registry is critical to providing clinicians in the National Alpha-1 Clinic with easier access to medical data and treatment outcomes.

Our awareness and advocacy efforts in the past year included presentations at the Irish Thoracic Society (ITS) conference in Dublin, UCD School of Medicine, COPD Support Groups in Ballyfermot and Sligo, as well as presentations to medical teams in St Vincent's, Drogheda, Letterkenny, Waterford, and Peamount hospitals. Our annual Alpha-1 conference was held in October 2016 in the Marino Institute of Education. Over 100 Alpha-1 patients and family members congregated in a relaxed and informal environment. We were delighted to welcome David Gillick, Olympic athlete to open the conference. Speakers included Professor Robert Sandhaus, Medical Director, US Alpha-1 Foundation, and Debby Lambert, from the National Rare Diseases Office. Dr Graham Love, CEO, Health Research Board launched our new

educational film "What is Alpha-1" which made a big impact.

The Alpha One Foundation has continued to work with the Medical Research Charities Group (MRCG), the Irish Donor Network, the Irish Platform for Patient Organisations, Science and Industry (IPPOSI), the Irish Lung Health Alliance, the Rare Disease Taskforce, the European Organisation for Rare Diseases (EURORDIS) and COPD Support Ireland. Association with such prestigious organisations is invaluable - enabling us to raise awareness of the Alpha One Foundation and to benefit from the expertise and experience of their members.

We are very thankful to everyone involved in fundraising in the last 12 months. The efforts have been amazing and varied, mini-marathons, full marathons, coffee mornings and Christmas cards. The money raised has been put to good use in our Alpha-1 screening programme.

Hopefully this brief synopsis will give an idea of the progress made by the Alpha One Foundation over the past 12 months. This work is a team effort and I wish to thank all my colleagues for their hard work and dedication throughout the year, particularly Dr Tomás Carroll, Laura Fee and Margaret Molloy.

Geraldine Kelly,
CEO, Alpha One Foundation



2

An Update from the National Alpha-1 Antitrypsin Deficiency Targeted Detection Programme

Alpha-1 antitrypsin deficiency (or simply Alpha-1) can be diagnosed by a simple blood test but unfortunately remains hugely under-diagnosed. A diagnosis of Alpha-1 can provide a unique opportunity for early medical intervention and in some cases the prevention of lung disease in both the affected individual and first-degree relatives. In May 2004, a national targeted detection programme for Alpha-1 was launched by the Alpha One Foundation with funding from the HSE.

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 Alpha-1. These guidelines recommend targeted testing of patients with chronic obstructive pulmonary disease (COPD), severe non-responsive asthma, cryptogenic (unexplained) liver disease and first-degree relatives of individuals with Alpha-1 (Table 2.1).

TABLE 2.1. ATS/ERS recommendations for diagnostic testing for Alpha-1 (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 necrotising 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?

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. If this is low, the second test looks at what type of alpha-1 antitrypsin protein is present by a method called isoelectric focusing. This method identifies variants of alpha-1 antitrypsin circulating in human blood, and is more commonly known as phenotyping (Figure 2.1). It is the most accurate method of diagnosing Alpha-1 and recognises not only the most common but also rare AAT variants.

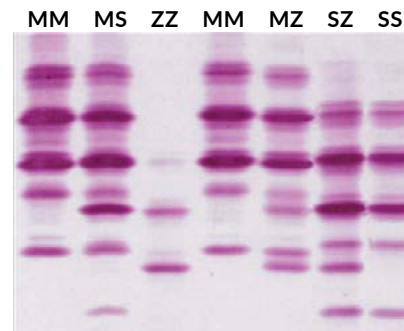


FIGURE 2.1. Typical isoelectric focusing gel for AAT phenotype identification with the most common phenotypes included.

WHAT HAVE WE FOUND IN IRELAND?

Since 2004 over 18,000 individuals with COPD, asthma, and liver disease, as well as first-degree relatives of known Alpha-1 individuals have been tested in the National AATD Targeted Detection Programme. We receive blood samples from hospitals all over Ireland as well as from GPs and primary care centres.

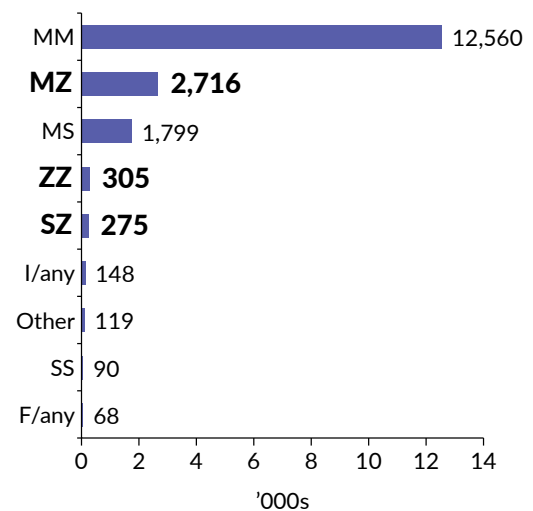


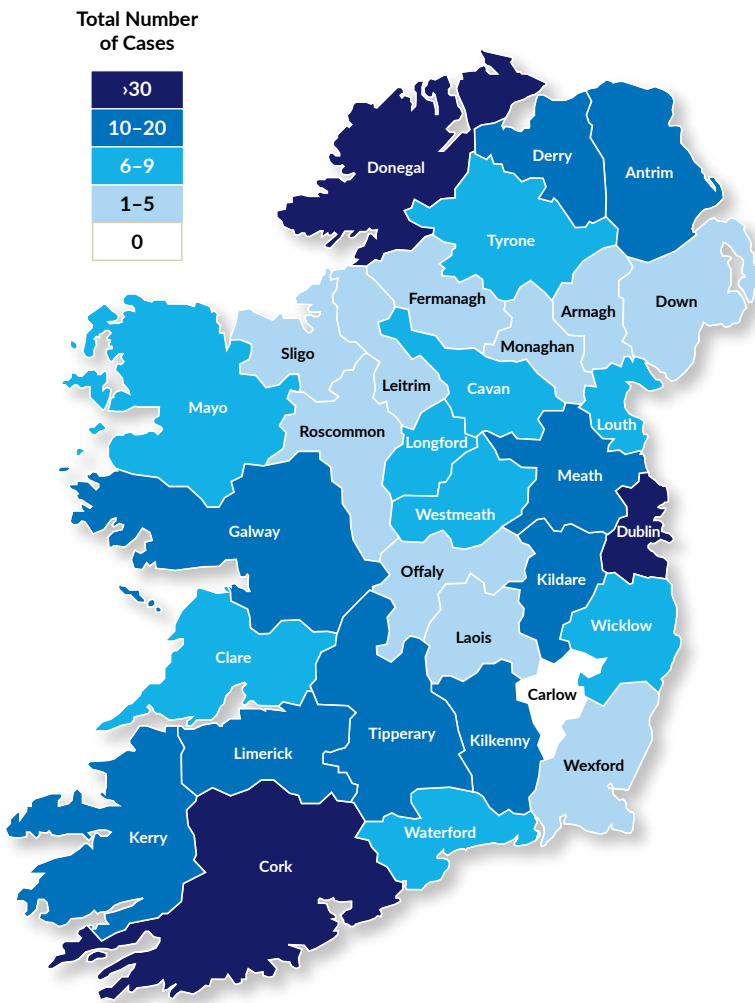
FIGURE 2.2. Results from the National AATD Targeted Detection Programme showing the alpha-1 antitrypsin phenotypes identified among the over 18,000 individuals tested.

FIGURE 2.3. Mean AAT levels and ranges in the major deficient phenotypes identified in the National AATD Targeted Detection Programme.

Phenotype	Cases	Mean AAT (g/L +/- SEM)	AAT Range (g/L)
MS	1799	1.25 +/- 0.008	0.58–3.82
MZ	2716	0.91 +/- 0.004	0.44–4.08
SS	90	0.90 +/- 0.02	0.62–1.54
SZ	275	0.65 +/- 0.01	0.33–1.20
ZZ	305	0.27 +/- 0.001	0.10–0.69

A total of 305 ZZ (severe Alpha-1) individuals have been identified, as well as 275 SZ individuals, who are also at risk of developing lung and liver disease (Figures 2.2 and 2.3). In addition, a large number of other clinically significant phenotypes have been detected including 2,716 MZ, 90 SS, 21 IZ, and 12 IS phenotypes. A number of extremely rare AAT mutations have also been identified. These include M_{wurzburg}, Z_{bristol}, M_{malton} and five different Null mutations (Null_{bolton}, Null_{cork}, Null_{dublin}, Null_{porto} and Null_{amersfoort}).

FIGURE 2.4. Distribution of severe AATD cases detected to date (n = 360).



The goal of the screening programme is to ensure people with Alpha-1 are correctly diagnosed and given the opportunity to receive expert medical care. Newly diagnosed individuals can be referred to the National Centre of Expertise for AATD in Beaumont Hospital under the care of Professor Gerry McElvaney. In addition, our focus on family screening allows the identification of younger relatives with Alpha-1. These individuals benefit from lifestyle changes such as smoking cessation and closer medical observation which can help prevent or postpone the development of lung disease.

In the past 12 months we have given talks about Alpha-1 to a mixture of respiratory, liver and biochemistry teams in St Vincent’s, Drogheda, Letterkenny, Waterford, and Peamount hospitals, to COPD patient support groups in Ballyfermot and Sligo, and to 3rd year medical students in UCD. The main aim of these presentations is to increase awareness of Alpha-1 and to encourage testing. While the respiratory (and liver) teams are dealing with patient populations most at risk due to Alpha-1, many hospital Immunology, Biochemistry, and Clinical Chemistry Departments measure alpha-1 antitrypsin levels as a routine test during normal blood investigations.

Opportunities to increase the detection of AATD in the healthcare system in future include the roll-out of the National Medical Laboratory Information System (MedLIS) which is the new national IT system for laboratory tests and the new national COPD guidelines currently under development. The Alpha One Foundation has made contributions to both these initiatives.

3

The New National Alpha-1 Antitrypsin Deficiency Registry

WHAT IS THE 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, pulmonary function tests, CT scans of the chest and ultrasounds of the liver are obtained from the medical chart, entered in the registry and updated at regular intervals.

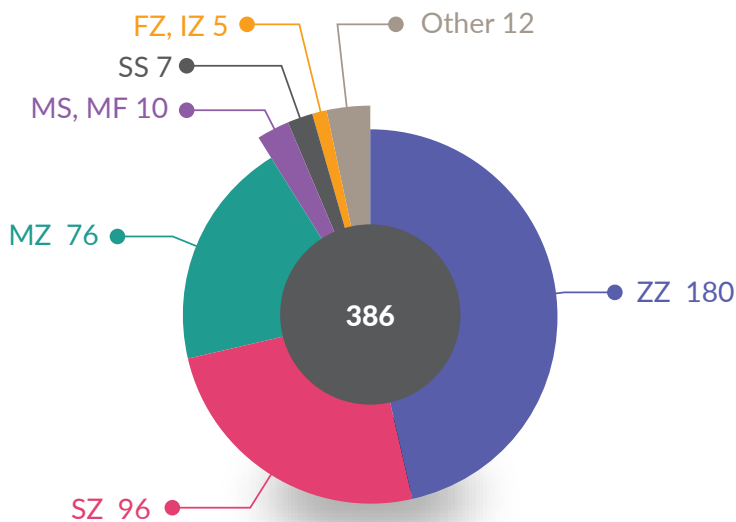


FIGURE 3.1. Total number of individuals enrolled in the registry according to AATD phenotype.

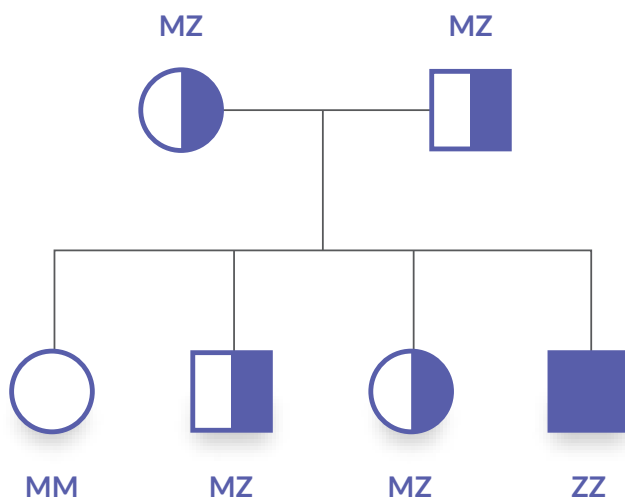


FIGURE 3.2. An example of the new family tree tool on the National AATD Registry.

WHAT IS THE PURPOSE?

Once data is entered in the registry it is analysed and used for research purposes to increase our knowledge of the condition which will lead to better treatments in the future. A registry also allows us to quickly identify who might benefit from new treatments or from participating in clinical trials in the future.

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 the Alpha One Foundation team will provide individuals with an information leaflet and answer any questions at the time of enrolment.

QUESTIONS?

If you or your family are interested in enrolling or have any questions regarding the registry please contact Margaret Molloy on 01-809 3749.

CURRENT REGISTRY

The Irish National AATD Registry was re-developed and launched on July 31st 2017 with the help of OpenApp an Irish company specialising in the use of technology in healthcare. The re-development has allowed the Alpha One Foundation to expand data collection and enhance data analysis. The re-developed registry has many new features including a pedigree tool to highlight the impact of the condition within a family.

In 2016, a total of 106 new patients were consented for the registry to give a total of 386 individuals enrolled.

The most severe form of AATD is the ZZ phenotype. Currently, there are a total of 180 ZZ individuals enrolled in the registry, of which 56.4% are male and 43.6% are female. The

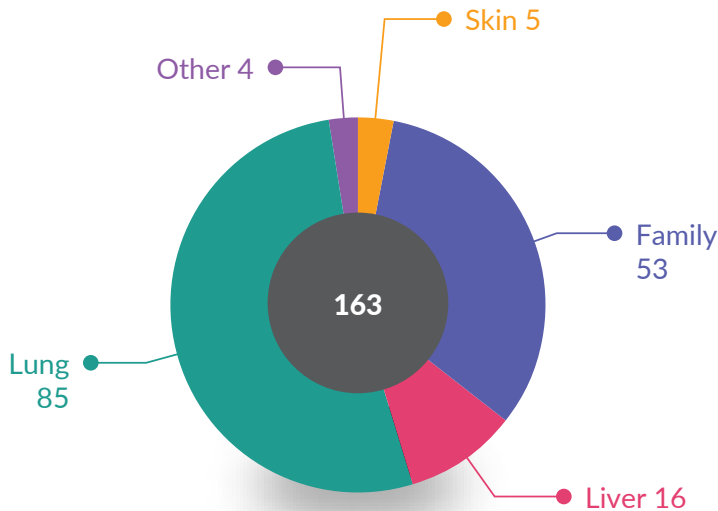


FIGURE 3.3. Reasons for Diagnosis Among ZZ AATD Registry Participants.

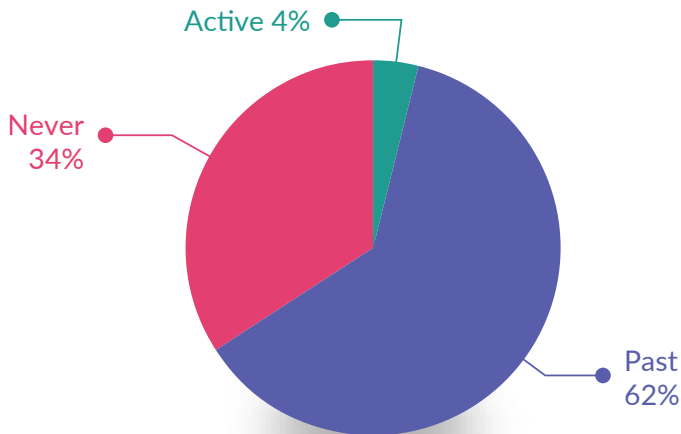


FIGURE 3.4. Smoking Status Among ZZ AATD Registry Participants.

average age is 54 years and ranges from 17–86 years. The majority of enrollees were diagnosed with AATD due to lung symptoms (51.5%), followed by family screening (32.5%), liver symptoms (9.8%), skin manifestations (3.1%) and other incidental findings (2.5%).

A major risk factor for developing lung disease in AATD is smoking. Fortunately, only a small percentage of ZZ individuals are active smokers (3.9%). The majority are past smokers (61.6%) with an average pack year smoking history of 19.3 (0 - 80) while 34.5% of individuals were never smokers. Over half (65%) of individuals displayed evidence of obstructive lung disease (COPD) on pulmonary function tests with 61.3% of these demonstrating severe or very severe disease. Of those diagnosed with COPD, 96% also demonstrated evidence of emphysema on CT of the chest. However, only 3.1% have required a lung transplant due to emphysema caused by AATD.

In terms of liver disease, 21.1% of ZZ individuals had abnormal liver function tests. A review of the abdominal ultrasounds revealed that 22.7% had evidence of fatty liver disease or cirrhosis. A small percentage (3.7%) required a liver transplant due to liver disease caused by AATD.

The lung and liver are the most commonly known organs affected in AATD. However, some evidence is emerging that other organs could be affected by AATD and the registry will be beneficial in investigating this further.

4

Meet the Alpha-1 Researchers

**THOMAS MCENERY**

I am undertaking research into alpha-1 antitrypsin deficiency (AATD) with Professor McElvaney as part of a two-year MD programme with the Royal College of Surgeons in Ireland. I am a medical doctor and previously worked on Professor McElvaney's team in Beaumont Hospital before moving to the lab; all part of my training towards becoming a Respiratory Consultant.

My research is focused on neutrophils in AATD. Neutrophils are a type of white blood cell that normally fight infection in the body. They are very effective at this but can also accidentally damage the body's tissues if their action is not controlled. For instance, neutrophils produce neutrophil elastase, the main substance that damages the lungs if not enough alpha-1 antitrypsin (AAT) is present to guard against it.

Professor McElvaney's research team have done a lot of work in showing that AAT plays an important role in controlling how neutrophils function. My research builds on this, looking specifically at how infusions of AAT ('augmentation therapy') affect the outer wall (cell membrane) of neutrophils in patients with AATD. In order to do this I take blood samples

from our patients immediately before they get their infusion and then two days later. I first isolate the neutrophils and then split them apart to detach the cell membranes. With the help of collaborators in Dublin City University we then use a device called a mass spectrometer to tell us exactly what proteins are present on the cell membrane. Comparing these proteins allows us to map exactly how the neutrophils have been affected by AAT.

This research will help us to better understand the anti-inflammatory effects of AAT. This is a very exciting area of research that may have widespread implications; for instance in broadening the applications of AAT as a treatment for other conditions like inflammatory arthritis.

**LAURA FEE**

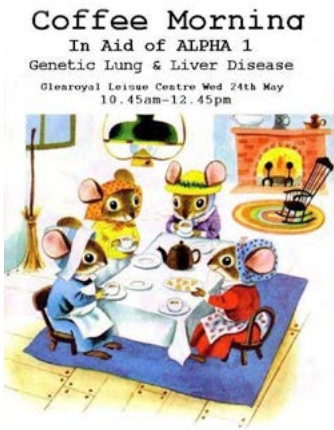
I have worked with the Alpha One Foundation since 2012 after I completed a degree in Biomedical Science in NUI Galway and a master's degree in Immunology and Global Health in NUI Maynooth. I help run the National AATD Targeted Detection Programme which tests approximately 200 samples each month from 30 hospitals and GP practices around Ireland. To date we have screened over 18,000 individuals for Alpha-1 from all over the country.

As well as my roles in the Alpha One Foundation I also began a 3 year respiratory research PhD carrying out research into Alpha-1 in April 2016 under Professor McElvaney. My project looks at how the immune system of people homozygous (ZZ) and heterozygous (MZ) for Alpha-1 acts differently compared to those without the deficiency. I have shown that a certain part of our immune system called the 'complement system' becomes 'turned on' or activated more quickly in people with Alpha-1. The complement system is a network of proteins which are important for fighting

infection in the body. This over activation can have negative effects including activation of white blood cells such as the neutrophil. These white blood cells when over-activated can release proteins such as neutrophil elastase which can attack lung tissue. All of this can result in the prolonged infections and lung tissue damage which contribute to the often early onset COPD seen in those with Alpha-1. In my future research we hope to identify why this over-activation of the immune system occurs and find approaches or treatments which can prevent this from happening.

Both of these projects would not be possible without a very supportive laboratory research team in RCSI Beaumont, funding from the Health Research Board, and of course the Alpha-1 patient community who kindly donate the blood samples we need.

5 Recent Events



COFFEE MORNING

A huge thank you to everyone who took part in a coffee morning in aid of Alpha-1 in the Glenroyal Hotel in Maynooth in May. A special thanks to Orla Keane and her merry band of helpers. Thank you also to Colette Stears who donated a Dublin GAA jersey signed by the Dublin footballers and to Stephen Smith who donated an Ireland rugby jersey signed by the squad. Both jerseys were auctioned on the day.

WOMEN'S MINI MARATHON 2017

Congratulations to Helen Stephens, Elaine Greally and Sinéad Nolan who ran the VHI Women's Mini-Marathon in June 2017 in aid of Alpha-1. Thank you for flying the flag for Alpha-1 and for your generous donation.



ANNUAL ALPHA-1 CONFERENCE

Our Annual Alpha-1 Conference in October 2016 was our biggest to date with over 100 in attendance.

Special guest speakers were Irish athlete and Olympian David Gillick and Professor Sandy Sandhaus from the US Alpha-1 Foundation, pictured here (below) with Kitty O'Connor and Professor Gerry McElvaney.



Many thanks to Josephine McGuirk for organising the sale of Christmas cards on the day. The cards are available again this year, please contact the Alpha One Foundation on 01-8093876 or email alpha1@rcsi.ie.



RESPREEZA REIMBURSEMENT CAMPAIGN

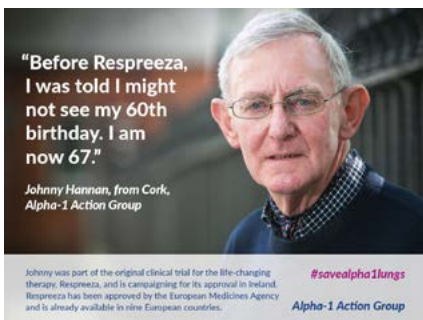
The campaign to reimburse Respreeza started in early December 2016 following a recommendation by the National Centre for Pharmacoeconomics (NCPE) to the HSE not to reimburse the treatment. Over the following nine months a number of events were organised by the Alpha One Foundation and the Alpha-1 Action Group in an effort to have the decision reversed.

February 2017: Demonstration outside the HSE Head Office. A letter was handed in for the attention of John Hennessy, National Director for Primary Care and Tony O'Brien, Director General of the HSE.



April 2017: An information briefing took place in Leinster House on April 12th involving members of the Dáil, Alpha-1 patients and the Alpha One Foundation. All TDs and Senators were invited to this meeting. We are grateful to Billy Kelleher TD for hosting the event.

May 2017: The Alpha-1 Action Group organised a march outside Dáil Éireann on May 31st 2017 where patients and their families highlighted the Respreeza issue. This was covered widely in the media on TV, radio and in newspapers.



June 2017: Social media campaign which was organised by the Alpha-1 Action Group. Over a couple of weeks six patient stories were featured to raise awareness of Alpha-1 and the campaign.

July 2017: A Joint Committee on Health hearing on how drugs for rare diseases are assessed and funded. The Alpha One Foundation were invited to contribute. After the Oireachtas hearing members of the Alpha One Foundation and the Alpha-1 Action Group met several concerned members of Dáil Éireann.

August 2017: Following a final decision by the HSE not to reimburse Respreeza a silent protest took place outside the HSE and letters of appeal were handed in to John Hennessy and Tony O'Brien of the HSE. A letter was also handed in to the Department of Health for the attention of Minister Simon Harris.

6 Sixth Global Alpha-1 Patient Congress

The Sixth Alpha-1 Global Patient Congress was held from April 7th to 8th in Lisbon, Portugal. Over 200 people from 34 countries heard world-famous Alpha-1 scientists and doctors, as well as patients, caregivers and family members discuss topics including the latest research

and detection efforts around the world. The status of patient registries and strategies to strengthen the Alpha-1 message globally were also discussed. Three Irish delegates attended representing the Irish Alpha-1 community and the Alpha One Foundation of Ireland.



Irish Delegation pictured with Henry Moehring, CEO and President, US Alpha-1 Foundation at the Global Patient Congress in Lisbon.

L-R. Henry Moehring, Ann O'Rourke, Gerard Greally, and Bernie Nolan

Bernie Nolan and Gerard Greally

I would like to thank the Alpha One Foundation for giving me the opportunity to attend the Alpha-1 Global Patient Congress which I attended with my brother Gerard. On the first evening we attended a memorial service for John W. Walsh a co-founder of the Alpha-1 Foundation who died on 7th March 2017. Many tributes were paid to the late Mr Walsh including one from Professor Gerard McElvaney. The service was followed by a reception and dinner at which we were accompanied by the other Irish attendees. We also met people from many other countries with whom we were able to discuss and share our experiences of many aspects of Alpha-1. Next day we attended a number of interesting presentations covering the following topics of particular relevance; the efficacy of intravenous augmentation therapy, diet for Alpha-1 patients, sleep apnea and insomnia, and lung and liver transplantation. The Congress concluded on Saturday with advice on good exercise, the importance of technology in health management, and effective patient engagement. It was apparent from our discussions with people from other countries that the research being carried out in Ireland is of particular importance and significance. We are fortunate to have such a wonderful medical and research team working within the Alpha One Foundation in Ireland whose aim is to improve the health and wellbeing of us, the Alpha-1 patients.

Ann O'Rourke

I attended the Alpha-1 global conference connecting our community in Lisbon in April earlier this year. I enjoyed meeting so many people with the same illness. I also enjoyed listening to all the international speakers and hearing their experiences with the disease. I found it very interesting, it gave me an insight into how the treatment works for people and how it is handled differently outside Ireland. It made me more self-aware about my illness and how I feel on a day to day basis. I learned a lot from the trip to Lisbon and I would like to thank the Alpha One Foundation for giving me the opportunity to attend the conference. It was a really enjoyable experience.

Current Research Developments in Alpha-1



Awards for Alpha-1 research

Earlier this year RCSI Researchers in Beaumont Hospital teamed up with collaborators from the Rheumatology Departments at St Vincent's Hospital and the Mater Hospital, and discovered a link between Alpha-1 and the severity of rheumatoid arthritis. Of major importance, their new research has shown that people with rheumatoid arthritis and one defective AAT gene (i.e. MZ carriers), have increased severity of rheumatoid arthritis disease. The research is published in the prestigious journal, *Arthritis & Rheumatology*, which is ranked 2nd of all rheumatology journals.

In the last 12 months the McElvaney lab has also established an industrial collaboration with an international company called Chiesi, which is based in Italy. The lab is working with Chiesi in testing new neutrophil elastase inhibitors in airway samples donated by Alpha-1 patients. These new compounds could substitute for a lack of the alpha-1 protein and provide the barrier that is needed to prevent tissue damage and lung disease in Alpha-1.

These research projects give a greater understanding of Alpha-1 and will allow us to develop new therapies, improve the care of patients, and ultimately lead to better health outcomes for Alpha-1 patients. This work is being carried out by an active and dynamic group of scientists and postgraduate students registered for the degree of PhD or MD who have received a number of awards for their studies over the last year. In March, Oliver McElvaney was announced winner of the postgraduate category of the RCSI Research Day. In May, Laura Fee and Danielle Dunlea presented their research findings at the American Thoracic Society International Conference, and Noreen Lacey gave an oral presentation of her project at the Irish Thoracic Society Annual Scientific Meeting. Aside from the scientific achievements and outputs of the McElvaney lab, the team also strives to part-take in community outreach programmes. Members of the research group took part in the recent BT Young Scientist exhibit at the RDS to encourage young people, especially girls, that a career in science is a rewarding possibility for everyone. The lab also hosts a number of transition year students from local secondary schools to give them a taste for what a scientist's life in a research lab is really like.



Research investigating exercise as a treatment for Alpha-1

We are now 2 years into this 4 year project. To date 30 participants from the National Alpha-1 Clinic in Beaumont Hospital have participated in our research with 9 having completed a 12 week programme of pulmonary rehabilitation. We are continuing to recruit potential participants from the weekly Alpha-1 clinic and have expanded our testing regime to include some additional measurements.

A development that we are really excited about is the launch of the MedEx Home Exercise Programme. From the outset one of the limitations of our Alpha-1 study was that we could only offer pulmonary rehabilitation to patients who lived in the north Dublin area, as all exercise classes took place in Dublin City University (DCU). The MedEx Home Exercise Programme takes advantage of the wealth of experience that MedEx Wellness, The School of Health and Human Performance in DCU and DCU Sport have built up over the last 10 years in the provision of exercise to treat a range of chronic conditions. We have designed a structured exercise programme that can be completed by patients at home.

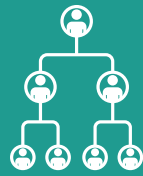
In order to take part in the home exercise programme we are asking that potential recruits would still come to DCU for testing in advance of beginning the programme and again after 10- 12 weeks. We can offer encouragement, advice and support while you are engaging in the programme and all participants will be free to attend the structured exercise classes in DCU to supplement the Home Exercise Programme, if they so wish.

Recruitment to the programme is ongoing. Over the next 12 months we are hoping to recruit up to 60 more participants on a rolling basis. All exercise testing will be taking place in DCU on a weekly basis.

Want to take part?

...If so, please contact:

Eóin Durkan at Tel: 01-700 8470 or
Email: eoin.durkan@dcu.ie



Family Study to Investigate Lung Risk in SZ Alpha-1

The purpose of this 3 year research study is to improve our knowledge about individuals who carry both the abnormal S and the abnormal Z types of the alpha-1 antitrypsin (AAT) gene, more commonly referred to as SZ.

We want to clarify whether SZ individuals are at increased risk of developing lung disease when compared to MM, MS and MZ individuals.

Our aim is to enrol 100 SZ individuals, and by testing their family members we hope to subsequently enrol 400 of their siblings and parents into this study. The strength of this study is in the unique family based design.

Participants will be offered lung function testing, routine blood tests and blood test to

confirm alpha 1 status. Each participant will then complete a questionnaire.

We will compare lung function, blood results, smoking history and other environmental exposures between these groups to identify any differences which might suggest a presence or risk of disease. We also hope to further clarify what level of AAT in the blood is protective against lung damage. We believe this level may vary depending on the type AAT each individual produces.

Want
to take
part?

...If so, please contact:

Dr Alessandro Franciosi, MB, BCh, BAO,
Clinical Researcher, Alpha One Foundation,
RCSI Building, Beaumont Hospital, Dublin 9.

Tel: 01-809 3876

Email: alessandrofranciosi@rcsi.ie

8

Acknowledgements

We would like to thank the following:

- Pat O'Brien, Emma Pentony, Helen Moore, Dr Bill Tormey and the Beaumont Hospital Chemical Pathology Department for their continued support and advice
- Angela McBride of the Alpha-1 Foundation (USA), and Gonny Gutierrez of Alpha-1 Global
- Dr Ilaria Ferrarotti and Dr Stefania Ottaviani, Centre for Diagnosis of Inherited Alpha-1 Antitrypsin Deficiency, University of Pavia, Italy
- Professor Dermot Kenny and the RCSI Clinical Research Centre (www.rcsicrc.ie)
- Jing Er Low and Kim Chuan Kok, RCSI medical students who carried out research projects with the Alpha One Foundation in 2017
- The Health Research Board (HRB), The Medical Research Charities Group (MRCG), the Irish Platform for Patients' Organisations, Science & Industry (IPPOSI), the Irish Lung Health Alliance, and the Irish Thoracic Society (ITS)
- President Michael D. Higgins for his continued support as patron of the Alpha One Foundation
- A special thank you to everyone who took part in or organised fundraising events throughout the year

The late John Walsh (far right) pictured with the Irish delegation at the 4th International Alpha-1 Patient Congress in 2013.



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

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

- Adelaide and Meath Hospitals, including National Children's Hospital Tallaght
- 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 Hospitals
- Galway University Hospitals
- James Connolly Memorial Hospital Blanchardstown
- Kerry General Hospital
- Letterkenny General Hospital
- Mater Misericordiae University Hospital, Dublin
- Mayo General Hospital
- Midland Regional Hospitals: Tullamore, Mullingar, and Portlaoise
- Midwestern Regional Hospital, Limerick
- Naas General Hospital, Co. Kildare
- Our Lady's Children's Hospital, Crumlin
- Our Lady of Lourdes Hospital, Drogheda
- Our Lady's Hospital, Navan
- Peamount Hospital, Dublin
- Roscommon County Hospital
- Rotunda Hospital, Dublin
- Sligo General Hospital
- St James's Hospital, Dublin
- St Luke's General Hospital Carlow/Kilkenny
- St Vincent's University Hospital, Dublin
- South Tipperary General Hospital, Clonmel
- Waterford Regional Hospital
- Wexford General Hospital



Alpha-1 is the most common genetic cause of COPD



Smokers who carry just a single defective Alpha-1 gene have a **5 fold increased risk** of developing lung disease like COPD



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



A study by the Irish Thoracic Society estimates that approximately **440,000 people** have COPD in Ireland



200 blood tests for Alpha-1 are performed by the Alpha One Foundation per month



18,000 individuals tested for Alpha-1 to date in the National Targeted Detection Programme



>700 Alpha-1 patients attending National Centre of Expertise for Alpha-1 at Beaumont Hospital