A1F 7th Global Research Conference & 10th Patient Congress

April 4 - 5, 2025

Lisbon, Portugal

ALPHA-1 FOUNDATION

INNOVATION INVESTMENT INSIGHT IMPACT

30th 1995 2025

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Things to Know

Name Badges

Please wear your registration badge throughout the event. It will give you access to sessions and meal functions.

Meetings & Meals

All meetings will take place in the Coimbra Room on the second floor.
All meals will take place in the Eduardo VII Room on the second floor.

Photography & Video

Please be advised that the Alpha-1 Foundation (A1F) is taking photos and video throughout the conference. All photos and videos taken by the official photographer and videographer may be used in A1F publications or reproduced to be used for newsletters, promotional materials, inclusion on websites or any other purpose. You release A1F from any liability connected with the taking of photographs and video. If you do not wish to have your picture used in any publication, please let A1F know in writing. Personal recording of any presentations is prohibited.

Patient Confidentiality

Due to the sensitivity of health information, we encourage you to make an informed choice about whether or not to provide your personal information to the organizations exhibiting and participating at this conference.

Insurance

Congress organizers cannot accept liability for personal injuries or for loss or damage to property belonging to congress participants during or as a result of the event.

Letter from Leadership

Welcome to the Alpha-1 Foundation's (A1F) 7th Global Research Conference & 10th Patient Congress. With a deep history of international collaboration, A1F is proud to host representatives from over 25 countries who share a drive to cure Alpha-1 Antitrypsin Deficiency (Alpha-1) and to improve the lives of Alphas worldwide. A1F's Global Program works tirelessly with patients and stakeholders to increase awareness, detection, and access to care for Alphas around the world.

In 2025, A1F proudly commemorates its 30th anniversary, a milestone that celebrates and honors the legacy of the many patients and stakeholders who have made an impact and continue to make meaningful contributions to the Alpha-1 community.

For three decades, with the contributions of the Alpha-1 community, A1F has designed and launched innovative, patient-focused programs and services supporting Alphas including a solid infrastructure to promote pioneering scientific research and the establishment of collaborative partnerships to advance the development of novel therapies. The essence of A1F's work has resulted in improving the quality of life for those affected by Alpha-1.

From its inception, A1F has created an international network of scientists and experts who continue to forge critical partnerships in their commitment to applying their knowledge of all aspects of the condition and research towards a cure.

A1F's 7th Global Research Conference goal is to bring a multidisciplinary group of scientists and experts together to present and discuss novel treatment paradigms in Alpha-1 that will accelerate research towards a cure.

Similarly, A1F's 10th Patient Congress is designed to provide a platform for education, discussion, and networking. The program includes research and therapeutic updates, journeys to get augmentation therapy approved and reimbursed, medical collaboration, patient engagement in research, advocacy advancement through public policy and clinical practice, and the importance of awareness, testing, and detection. The overall goal of A1F's 10th Patient Congress is to empower patients to make informed decisions about care and take part in Alpha-1 research.

We would like to thank all attendees for joining us in A1F's 7th Global Research Conference & 10th Patient Congress. This event provides an incredible opportunity for Alpha-1 patients, family members, caregivers, researchers, and industry partners around the world to connect. We aim to provide you with the most current and relevant information about Alpha-1 and we encourage you to gain a renewed vision and purpose for the task at hand.

A special thank you to A1F's 7th Global Research Conference & 10th Patient Congress sponsors: AlphaNet, CSL, Grifols, and Takeda and the Emerging Therapy Sponsors: Beam Therapeutics, Kamada, Sanofi, Takeda, and Wave Life Sciences. We are grateful for your ongoing commitment to Alphas worldwide.

Scott Santarella
President & Chief Executive Officer
Alpha-1 Foundation

A1F 7th Global Research Conference Agenda

April 4, 2025
Lisbon, Portugal

Welcome and Introduction

08:00 am – 08:10 am	**Welcome and Introduction** Scott Santarella, Andrew Wilson, MD , Pavel Strnad, MD, Alice Turner, MD, PhD

Session 1: The Liver – Moderator, Pavel Strnad, MD

08:10 am – 08:30 am	**Cancer Risk in AATD** Morten Dahl, MD, PhD
08:30 am – 08:40 am	**Discussion**
08:40 am – 09:00 am	**AATD-Associated Liver Disease: Insights into Disease Course and Modifying Factors** Pavel Strnad, MD
09:00 am – 09:10 am	**Discussion**
09:10 am – 09:30 am	**A Unique Tool to Predict Future Severe Liver Disease in ZZ and SZ Children, and Findings from the NIH-Childhood Liver Disease Research Network** Jeff Teckman, MD
09:30 am – 09:40 am	**Discussion**
09:40 am – 10:00 am	**Selective Autophagy of the Endoplasmic Reticulum: Mechanisms, Roles, and Therapeutic Potential** Carmine Settembre, PhD
10:00 am – 10:10 am	**Discussion**
10:10 am – 10:30 am	**Break**
10:30 am – 10:50 am	**Learning From Trials Targeting Liver Disease** Mattias Mandorfer, MD, PhD

10:50 am – 11:00 am	**Discussion**
11:00 am – 11:20 am	**From Technology to Disease Mechanisms: Spatial Single-Cell Proteomics in AATD** Florian Rosenberger, PhD
11:20 am – 11:30 am	**Discussion**
11:30 am – 11:50 am	**Selection for Somatic Escape Variants in SERPINA1 in the Liver of Patients with Alpha-1 Antitrypsin Deficiency** Stefan Marciniak, MD, PhD
11:50 am – 12:00 pm	**Discussion**
12:00 pm – 01:00 pm	**Lunch Break**

Session 2: The Lung – Moderator, Alice Turner, MD, PhD

01:00 pm – 01:20 pm	**Biomarkers and Endotypes in AATD** Josh De Soyza, MD
01:20 pm – 01:30 pm	**Discussion**
01:30 pm – 01:50 pm	**Going Beyond the Z Allele** Teresa Martin, MD
01:50 pm – 02:00 pm	**Discussion**
02:00 pm – 02:20 pm	**NLRP3 Inflammasome and Interleukin-1b Activation in AATD** Emer Reeves, PhD
02:20 pm – 02:30 pm	**Discussion**
02:30 pm – 02:50 pm	**Break**
02:50 pm – 03:10 pm	**Controversies and Advances in Augmentation** Miriam Barrecheguren, MD
03:10 pm – 03:20 pm	**Discussion**
03:20 pm – 03:40 pm	**Using Real Life Data to Extend Understanding of AATD** Alice Turner, MD, PhD
03:40 pm – 03:50 pm	**Discussion**

Concluding Remarks

03:50 pm – 04:00 pm	Scott Santarella, Andrew Wilson, MD

A1F 10th Patient Congress Agenda

April 5, 2025
Lisbon, Portugal

09:40 am – 09:45 am	**Welcome and Introduction** Scott Santarella
09:45 am – 09:55 am	**Industry Sponsor Presentation** CSL
09:55 am – 10:10 am	**Emerging Therapies in Alpha-1** Beam Therapeutics
10:10 am – 10:30 am	**Research Panel: Alpha-1 Registries & Participating in Clinical Trials – EARCO, Ireland and the Alpha-1 Foundation** Jeanine D'Armiento, MD, PhD, Gerry McElvaney, MD, Marc Miravitlles, MD, FERS
10:30 am – 10:40 am	**Q&A**
10:40 am – 11:10 am	**The Augmentation Therapy Journey in Canada and Alpha-1 Norden** Angela Diano, Sylvain Grenier, PharmD, Knut Skaar
11:10 am – 11:20 am	**Q&A**
11:20 am – 11:40 am	**Break**
11:40 am – 11:50 am	**Industry Sponsor Presentation** Grifols
11:50 am – 12:05 pm	**Emerging Therapies in Alpha-1** Kamada
12:05 pm – 12:25 pm	**Progress Updates from South America** Mariano Fernandez Acquier, MD
12:25 pm – 12:35 pm	**Q&A**

12:35 pm – 12:45 pm	**Industry Sponsor Presentation** Takeda
12:45 pm – 01:45 pm	**Lunch**
01:45 pm – 02:00 pm	**Emerging Therapies in Alpha-1** Sanofi
02:00 pm – 02:20 pm	**Alpha-1 Europe Alliance – Working Collectively on Behalf of Patients** Fernanda Aspilche, Cristina Barbiero
02:20 pm – 02:30 pm	**Q&A**
02:30 pm – 02:50 pm	**Patient Empowerment – Your Voice in Advocacy & Policy** Ines Aoufi, Kinga Wojtowicz
02:50 pm – 03:00 pm	**Q&A**
03:00 pm – 03:30 pm	**Research Panel – Advancements in AATD Q & A** Mark Brantly, MD, Jeanine D'Armiento, MD, PhD, Gerry McElvaney, MD, Robert Sandhaus, MD, PhD, Pavel Strnad, MD, Jeff Teckman, MD, Alice Turner, MD, PhD, Andrew Wilson, MD
03:30 pm – 03:45 pm	**Emerging Therapies in Alpha-1** Takeda
03:45 pm – 04:05 pm	**Break**
04:05 pm – 04:35 pm	**Panel: Living with Alpha-1 - Patients' Perspective** Cristina Barbiero, Carlos Cambon, Shane Fitch, Elena Goyanes, Peg Iverson, Steven Knowles, Anabela Lemos, Frank Willersinn
04:35 pm – 04:45 pm	**Q&A**
04:45 pm – 05:00 pm	**Emerging Therapies in Alpha-1** Wave Life Sciences
05:00 pm – 05:15 pm	**Alpha-1 Foundation Global Program Update** Scott Santarella
05:15 pm – 05:25 pm	**Q&A**
05:25 pm	**Closing Remarks** Scott Santarella

Speaker Bios

Ines Aoufi

RPP Group, Belgium

Ines Aoufi joined the RPP Group in June 2022 and holds a master's degree in international health policy from the London School of Economics. With a robust background in Public Health and a profound interest in health policy and politics, she has worked diligently to support numerous patient organizations in making their voices heard within the EU policy Landscape. Ines has been instrumental in activating patient associations across EU member states, empowering them and guiding their advocacy efforts.

Fernanda Aspilche

Belgium

Fernanda Aspilche was diagnosed with Alpha-1 Antitrypsin Deficiency (Alpha-1) in December 2019 at age 47, she is a ZZ Alpha. Shortly after her diagnosis, she met Frank Willersinn, president of Alpha-1 Plus in Belgium, and started to work with him as a volunteer. In May 2020, Fernanda joined the Board of Rare Diseases Belgium (RaDiOrg) and has become a fierce advocate for people living with a rare disease, specifically those with alpha-1. Fernanda is Spanish and has lived in Belgium for 25 years with family and works in the financial sector. In 2022, she joined an initiative called Climbing for Life as a COPD representative, and together with other 7 people who live with lung diseases, they climbed by bike the Sella Ronda in the Dolomite (Italy), taking the opportunity to create awareness around Alpha-1 and COPD.

Cristina Barbiero

Italy

Cristina Barbiero is a board member of the Alpha-1 European Alliance and a member of the Steering Committee for the Italian Patient Association. Cristina's commitment to raising awareness about Alpha-1 Antitrypsin Deficiency (Alpha-1) stems from her husband, Mario's diagnosis and experience navigating his condition. Her experience as a mother of three and a devoted wife has helped her continue the work that her beloved Mario started. Through her family's experience entering an experimental augmentation therapy program and growing a family, she has been able to bring awareness to the community.

Miriam Barrecheguren, MD

Vall d´Hebron University Hospital, Spain

Dr. Miriam Barrecheguren is a pneumologist working at Vall d´Hebron University Hospital in Barcelona. She graduated in Medicine at the University of Santiago de Compostela in 2008. After training in Respiratory Medicine, Dr. Barrecheguren obtained her PhD at the Autonomous University of Barcelona in 2016 on the diagnosis and management of COPD and AATD in Primary Care. Afterwards, she fulfilled a research fellowship at McGill University (Montreal, Canada) and obtained a Rio Hortega grant from the Health Institute Carlos III (Madrid, Spain). She was granted the honorary lecture for young researchers of the Spanish Society of Pneumology and Thoracic Surgery (SEPAR) in 2018.

Dr. Barrecheguren was the European Respiratory Society (ERS) Early Career Member (ECM) representative in the European Alpha-1 Research Collaboration (EARCO) until April 2024 and is the coordinator of the regional AATD network in Catalonia. Her primary interests include COPD, Alpha-1 Antitrypsin Deficiency (AATD) and mechanical ventilation. She has more than 90 peer-reviewed publications indexed in PubMed.

Mark Brantly, MD

University of Florida, USA

Dr. Mark Brantly is UF Emeritus Professor of Medicine and the former Alpha-1 Foundation Professor of Medicine, Molecular Genetics, and Microbiology in the Division of Pulmonary, Critical Care and Sleep Medicine and Vice Chair of Research for the Department of Medicine at the University of Florida. Professor Brantly completed his undergraduate degree at Florida State University and received his MD degree from the University of Florida. His Pulmonary and Research training was at National Institutes of Health in Bethesda, MD at the NHLBI Pulmonary Branch. Dr. Brantly was a Parker B Francis Fellow from 1986-1990. He later joined the NICHD Genetics Branch as a section head and concluded his stay at NIH back in the Pulmonary Division prior to joining the University of Florida Department of Medicine. In 2018, Dr. Brantly was honored with the John W. Walsh PAR Award for Excellence. Dr. Brantly is the former Scientific Director of the Alpha-1 Foundation, and he is currently a member of the Alpha-1 Foundation Board of Directors. Brantly has co-authored more than 200 publications and many of these publications are on the molecular basis of Alpha-1 Antitrypsin Deficiency. Much of his lab's recent research has focused on the gain of toxicity associated with misfolding of Z alpha-1-antitrypsin in liver and macrophages.

Carlos Cambón

Argentina

Carlos Cambón was diagnosed with Alpha-1 in 1999 at the age of 38. At that time, his doctors gave him a limited life expectancy unless he could access replacement therapy. After overcoming many obstacles, waiting for the therapy to be assigned to him, and ensuring that his health insurance would cover it, he was finally able to receive the treatment. In January 2025, after many challenges, he and his team successfully obtained the legal status of the Alpha-1 Foundation of Argentina. The foundation is committed to supporting patients, raising awareness, and finding better ways to diagnose Alpha-1 in Argentina and Latin America. Currently, he serves as the president of the Alpha-1 Foundation of Argentina and actively collaborates with doctors and other patients. With 25 over years of replacement therapy and 23 years of self-administering his infusions without the need for nurses or assistants, he continues to advocate for autonomy and better access to treatment for those in need.

Jeanine D'Armiento, MD, PhD

Columbia University, USA

Dr. Jeanine D'Armiento is a Professor of Medicine in Anesthesiology at Columbia University. Dr. D'Armiento is the Director of the Center for Molecular Pulmonary Disease in Anesthesiology and Physiology and Cellular Biophysics, and Director of the Center for Lymphangiomyomatosis (LAM) and Rare Lung Disease. Dr. D'Armiento's research focuses on understanding the mechanisms of lung injury and repair. Her laboratory integrates both in vitro and in vivo approaches and is uniquely situated to characterize the molecular changes in the study of lung injury and disease to identify potential therapeutic targets. Dr. D'Armiento's clinical work focuses on Rare Disease, and she is the Director of the Center for LAM and Rare Lung Diseases at Columbia University, which serves one of the largest populations of women with LAM in addition to patients with Alpha-1 Antitrypsin Deficiency (AATD). Dr. D'Armiento is also the medical liaison of A1F.

Dr. D'Armiento serves as Chair of the Executive Committee of the Columbia University Senate. In addition, she is presently the past Chair of the Board of Directors of the Alpha-1 Foundation and serves as a Consultant to the Director of the Division of Rare Disease Research Innovations (DRDRI), NCATs.

Morten Dahl, PhD

University of Copenhagen, Denmark

Prof. Morten Dahl is a Clinical Biochemistry professor specializing in Respiratory Genetics at the University of Copenhagen, Denmark. He runs a research team whose focus is on understanding mechanisms and risk factors underlying lung diseases such as Alpha-1 Antitrypsin Deficiency (AATD). Prof. Dahl graduated from the University of Copenhagen in 1998 and studied for a PhD in "Intermediate and severe AATD", awarded in 2002. He continued his research training from 2003 to 2006 as a postdoc at Brigham & Women's Hospital and Harvard School of Public Health in Boston, USA. After returning to Denmark and completing his specialist training, Prof. Dahl became associate professor in 2008 and full professor in 2018 at the Department of Clinical Medicine, Faculty of Health and Medical Sciences, University of Copenhagen.

Josh De Soyza, MD

University of Birmingham, UK

Dr. Joshua De Soyza is a Respiratory Specialist Registrar Training in the West Midlands, UK. He has recently completed a doctoral thesis with Prof. Alice Turner and the Birmingham AATD group, exploring bronchiectasis in Alpha-1 Antitrypsin Deficiency, and including collaborative work with Thoraxklinik Heidelberg. This has involved visual and algorithmic scoring of CT scans, analysis of serum biomarkers and sputum bacterial colonization, and comparison with clinical outcomes. He has clinical and research interests in airways diseases and respiratory infection.

Angela Diano

Canada

Angela Diano is the Executive Director of AlphaNet Canada, bringing over 20 years of experience in public health and community development from Ontario's Ministry of Community and Social Services, the Ministry of Health and Long-Term Care, and the Trillium Gift of Life Network. She led a multi-year advocacy campaign to establish a new Alpha-1 Antitrypsin Deficiency (Alpha-1) class on the Canadian Blood Services' Plasma Protein & Related Products Formulary, resulting in the inclusion of an Alpha-1 proteinase inhibitor in 2024 with Canada's blood operator. This accomplishment ensures that Canadian Alpha-1 patients have equitable access to augmentation therapy at no cost to them for generations to come. In November 2024, Angela oversaw the successful merger of Alpha-1 Canada and AlphaNet Canada, and as President and Chairperson of the Network of Rare Blood Disorder Organizations (NRBDO), she remains committed to ensuring all Canadian patients living with rare blood disorders receive timely access to life-saving therapies.

Mariano Fernandez Acquier, MD

Hospital del Torax Dr. A. Cetrangolo, Argentina

Dr. Mariano Fernandez Acquier is the associate director of the Cetrangolo Hospital, a public respiratory disease center in Buenos Aires, Argentina. He specializes in pulmonology and internal medicine. He has been involved in the field of Alpha-1 Antitrypsin Deficiency (AATD) research for more than 14 years. He participated in the first screening program in Buenos Aires and took part in the guidelines for diagnosing and treating Alpha-1 patients in Argentina. Dr. Fernandez Acquier has also served the patient community as an advisor, educator, and advocate.

Shane Fitch

Portugal

Shane Fitch has been a patient-caregiver advocate for the Alpha-1 Association of Spain since her son Arran Strong was born as a ZZ Alpha 24 years ago. She has worked intensively for global networks on strategic issues concerning the main gaps that exist in chronic and minority diseases: early detection, the right to receive holistic treatment, access to medicine, innovative therapies and patient registries – together with experts and leadership, in the field of respiratory health. Shane served on the Board of Alpha-1 Foundation as the first European member 2011-2014, setting up Alpha-1 Global. In 2022, the Lovexair Foundation was established from Spain, as a patient-led umbrella organization to address key issues in respiratory by applying digital health as a driver in the development and implementation of new models of care, education and social support to patients and caregivers, empowering people in becoming active in their role for better health and well-being. Lovexair set up HappyAir as an evolving ecosystem, to manage digital health research and care programs validating a care model for COPD patients.

Elena Goyanes

Spain

Elena Goyanes is the wife of a lung-affected patient and the mother of three children with Alpha-1 Antitrypsin Deficiency (Alpha-1). Shortly after her family's diagnosis, she reached out to the Spanish Alpha-1 Patient Association for information and support. She has been a member of the association since 2008 and joined the Board of Directors. in 2010. Since 2018, Elena has served as Vice President and is responsible for international relations. In addition to her national advocacy work, she is also a Board Member of the Alpha-1 Europe Alliance, where she collaborates on international initiatives to support the Alpha-1 community across Europe. Professionally, she works as a communications consultant. Her personal experience has driven her to actively contribute to the Spanish association in multiple ways. She coordinates the support group for parents of affected children, guides newly diagnosed patients, and participates in projects aimed at raising awareness of Alpha-1. Her efforts focus on improving patients' quality of life, ensuring access to available treatments, and fostering strong personal connections among people living with Alpha-1.

Sylvain Grenier, PharmD

Canadian Blood Services, Canada

Dr. Sylvain Grenier graduated from Université Laval, Québec with a Bachelor of Pharmacy. He completed a Doctor of Pharmacy degree from the University of Toronto. Dr. Grenier joined the Canadian Blood Services in 2019 as the Director of the Plasma Protein and Related Products Program (PPRP) where he has been leading various initiatives to modernize the Formulary

program and introduce pharmacy expertise in the PPRP field. Prior to this, he served 28 years in the Canadian Armed Forces (CAF), holding various roles and positions as a military pharmacist and retiring as the National Practice Leader for pharmacy, responsible for the management of the Canadian Armed Forces drug program. In addition of working in different military locations and positions in Canada, Dr. Grenier deployed in Bosnia in 1999-2000 and Afghanistan in 2010. After retiring from full-time active duty, he transferred to the Reserves where he still serves with the CAF on a part-time basis. He is the President of the Military and Emergency Pharmacy Section of the International Pharmaceutical Federation (FIP), elected in 2016. Between 2013 and 2023, he was a member of the CSHP National Awards committee, being appointed chair between 2014 and 2017. Dr. Grenier has been Adjunct Professor at the University of Ottawa since 2009, where he taught several Clinical Pharmacology courses for the Bachelor of Nursing and for the Ontario Primary Health Care Nurse Practitioner program.

Peg Iverson

USA

Peg Iverson was diagnosed with Alpha-1 Antitrypsin Deficiency (Alpha-1) in 1974 at the same time as her mother received her Alpha-1 diagnosis. She participated in the first National Institutes of Health Alpha-1 Study in 1985 after the death of her mother. She retired early, after a career in information technology. She began working as an AlphaNet Coordinator in 2011 and currently serves as an AlphaNet Program Manager. She is a past Iowa Support Group Leader, has chaired an Alpha-1 Foundation fundraiser called "Iowa Alpha-1 Get the Scoop/Walk", attends the Iowa support group meetings, participates in Alpha-1 advocacy efforts, and volunteers to speak about Alpha-1. She serves on the Foundation's Executive Committee as Secretary.

Steven Knowles

Australia

Steven Knowles established the Alpha-1 Association of Australia (AAA) in 2005 as a result of his continued interest in understanding Alpha-1 Antitrypsin Deficiency (Alpha-1) and bringing interested parties in Australia together. Steven's interest in Alpha-1 started in 2002, when he serendipitously discovered his Alpha-1 phenotype of MZ. With little clue as to what that meant, a path of inquiry led him to an invitation from the Alpha-1 Foundation to attend their inaugural International Patient Congress, held almost 20 years ago in Barcelona. He has attended all international conferences since, apart from 2015 in Barga, Italy, due to ill-health at the time. His adult children also have Alpha-1 and fortunately all are asymptomatic.

Anabela Lemos

Portugal

Anabela Lemos is a Portuguese pharmacist, with 15 years of experience in health and management-related fields. Her life took a new turn when her daughter was diagnosed with Alpha-1 Antitrypsin Deficiency (Alpha-1), after a difficult time dealing with symptoms. After the birth of her second child (MZ), she decided to leave her career as a pharmacist to focus on her family and investments. This new flexibility allowed her to engage with AA1P (Alpha-1 Portuguese Association), where she became a Board Member in 2020. Anabela is also involved in advocacy and communication activities with Alpha-1 Euro Alliance and Eurordis. She is part of the European Reference Networks for the liver and lungs. In her free time, she enjoys playing music and practicing padel, which helps her maintain a balance between her personal and professional life.

Mattias Mandorfer, MD, PhD

Medical University of Vienna, Austria

Dr. Mattias Mandorfer studied Medicine in Vienna & Heidelberg and obtained his M.D. degree in 2012. He is a consultant in Internal Medicine (since 2018) and Gastroenterology and Hepatology (since 2021), and Intensive Care (since 2023). In 2019, he stayed as a fellow at the Barcelona Hepatic Hemodynamic Lab, Liver Unit, Hospital Clínic. Dr. Mandorfer obtained his Ph.D. degree in 2017 and a junior faculty position in 2018. Since 2021, Dr. Mandorfer is a senior faculty member (Ap.Prof.; Medical University of Vienna) at the Division of Gastroenterology and Hepatology, Medical University of Vienna, and has been appointed head of the Vienna Hepatic Hemodynamic Lab. He serves as an associate editor of JHEP Rep and is/was a member of the editorial boards of Gastroenterology, J Hepatol, Hepatology, Am J Gastroenterol, and Liver Int, and Hepatol Comm. He is part of the Baveno Cooperation Steering and Research Committees and also serves as treasurer. In addition, he contributes to EASL (Educational Committee), UEG (National Society Forum), and EF-CLIF (Inspiring and Writing Group). Besides advanced chronic liver disease/cirrhosis/portal hypertension, alpha-1 antitrypsin deficiency is one of Dr. Mandorfer's key research interests.

Stefan Marciniak, MD, PhD

University of Cambridge School of Medicine, UK

Prof. Stefan Marciniak studied medicine at the University of Cambridge as part of its MD/PhD program. After training in pulmonology in Cambridge, London and Edinburgh, he undertook post-doctoral research in New York University for three years and then again in Cambridge. In 2012, he established his lab in the Cambridge Institute for Medical Research (CIMR) to study diseases caused by abnormal protein folding, including Alpha1-Antitrypsin Deficiency (AATD). In 2016, he became Professor of Respiratory Science at the University of Cambridge. He is also an active pulmonologist at Cambridge University Hospitals and Royal Papworth Hospital. In Cambridge, he directs the MD/PhD program, while nationally he directs the NHS England Rare Disease Collaborative Network (RDCN) in Familial Pneumothorax and co-leads the LifeArc Rare Respiratory Disease Centre.

Teresa Martin, MD

Hospital Beatriz Ângelo, Portugal

Dr. Teresa Martín is a pulmonologist based in Lisbon, Portugal, specializing in chronic obstructive pulmonary disease (COPD) and Alpha-1 Antitrypsin Deficiency (AATD). As of 2025, she serves as the Coordinator of the AATD Study Group of the Portuguese Pulmonology Society (SPP). She established the AATD consultation at Hospital Beatriz Ângelo, where she implemented augmentation therapy, and currently coordinates the hospital's Severe COPD consultation. Her research primarily focuses on AATD, COPD, and interstitial lung diseases. She has participated in international clinical trials and real-world registries, including EARCO (European Alpha-1 Research Collaboration), where she has been an Early Career Member for several years. Within EARCO, she has published research on the Pi*SS genotype and its association with pulmonary disease risk. She is also the first author of peer-reviewed publications in high-impact journals, such as Respiratory Research and Pulmonology. Dr. Martín has lectured at national and international conferences, actively engaging in scientific societies and contributing as a reviewer for major conferences and journals. Her dedication extends beyond academia to public awareness initiatives, including patient-focused talks for Rare Disease Day.

Noel G. McElvaney, MD

Royal College of Surgeons in Ireland, Ireland

Prof. Gerry McElvaney is Chairman of the Department of Medicine and Professor of Medicine at the Royal College of Surgeons in Ireland, Dublin, Ireland. Prof. McElvaney received his medical education at University College Dublin and completed his postgraduate internal medicine training at the Mater Misericordiae Hospital, St. Laurence Hospital and Jervis Street Hospital in Dublin before pursuing a pulmonary Fellowship in Vancouver, Canada. Following that, Prof. McElvaney worked in the Pulmonary Branch, NHLBI, NIH, Bethesda, and Cornell University-Rockefeller University Hospital, New York. He returned to Ireland in 1996. Prof. McElvaney has a well-established track record in research in Alpha-1 Antitrypsin Deficiency (AATD) and Cystic Fibrosis with substantial funding from National and International bodies. His work on inflammation and lung defenses has led to significant interactions with pharmaceutical companies interested in translational research. In 2003, Prof. McElvaney founded the Alpha-1 Foundation of Ireland and subsequently received funding from the Irish Department of Health and Children to set up an AATD research unit to further research into the condition. He also established the first National targeted detection program in Europe.

Marc Miravitlles, MD, FERS

Vall d'Hebron Research Institute, Spain

Dr. Marc Miravitlles is a senior researcher and consultant at Vall d'Hebron University Hospital in Barcelona. He was Chair of the Respiratory Infections Group of the European Respiratory Society (ERS) and Guidelines Director of the ERS. He has acted as a consultant for the development of international guidelines of COPD, including the American Thoracic Society (ATS)/ERS guidelines on exacerbations of COPD and the ERS statement on management of respiratory disease in Alpha-1 Antitrypsin Deficiency (AATD). He is also a consultant of the Spanish Ministry of Health for the development of the National Strategy Against COPD. He is the coordinator of the Spanish National Guidelines for COPD since 2011. He was the founder of the Spanish Registry for AATD in 1992 and is the co-chair of EARCO, the European Alpha-1 Antitrypsin Deficiency Research Collaboration (EARCO) Registry of the ERS since 2018.

Emer Reeves, PhD

Royal College of Surgeons in Ireland, Ireland

Prof. Emer Reeves is an Associate Professor in the Department of Anaesthesia and Critical Care Medicine, Royal College of Surgeons in Ireland (RCSI). She obtained her PhD in Biochemistry/Immunology from University College London and studied Data Protection Law at The Honorable Society of King's Inns, School of Law, Ireland. Her current research interests are centered on cell and molecular mechanisms involved in driving lung inflammation, with emphasis on the role of neutrophils and monocytes. This research is based within the clinical setting of chronic obstructive pulmonary disease, with emphasis on Alpha-1 Antitrypsin Deficiency (AATD). Prof. Reeves has attracted substantial national and international funding, and her research reaches a broad audience as demonstrated by successful publications in journals including Nature, Science Translational Medicine, BLOOD and Thorax.

Florian Rosenberger, PhD

Max Planck Institute of Biochemistry, Germany

Dr. Florian Rosenberger is currently an EMBO postdoctoral fellow with Prof. Matthias Mann at the Max Planck Institute of Biochemistry in Munich, Germany. His research has focused on developing methods that integrate laser microdissection and mass spectrometry to map the proteome of pathobanked tissue specimens at single-cell resolution. In collaboration, with Prof. Pavel Strnad and Prof. Aleksander Krag, he has recently characterized the proteomic events in hepatocytes that accumulate Alpha-1 Antitrypsin Deficiency (AATD), precisely delineating the molecular events of aggregate formation over time. Dr. Rosenberger is now transitioning to an independent assistant professor position at the Karolinska Institute, Sweden, where he will apply systems biology approaches to investigate metabolic diseases.

Robert Sandhaus, MD, PhD

Alpha-1 Foundation, USA

Dr. Robert Sandhaus is a Professor of Medicine in the Division of Pulmonary, Critical Care, and Sleep Medicine at the University of Colorado School of Medicine and is based at National Jewish Health in Denver, where he founded the Alpha-1 Antitrypsin Deficiency Program over 40 years ago. His early research focused on the role of white blood cell proteases in the prevention and promotion of lung disease. This work led to research and clinical interest in patients with the genetic deficiency of Alpha-1 Antitrypsin Deficiency (AATD). More recently he has been involved in evaluating the lung dysfunction of patients with Osteogenesis Imperfecta. Dr. Sandhaus has served on the Boards of Directors of the Alpha-1 Association, the Alpha-1 Foundation, AlphaNet, the Alpha-1 Project (TAP), the Association for the Accreditation of Human Research Protection Programs, Global Implementation Solutions, and the Osteogenesis Imperfecta Foundation. For more than 20 years, he has been the Executive Vice President and Senior Medical Director of AlphaNet and the Clinical Director of the Alpha-1 Foundation. More recently, he is also the Medical Director of AlphaNet Canada.

Scott Santarella

Alpha-1 Foundation, USA

Scott Santarella is President and CEO of the Alpha-1 Foundation (A1F), leading strategic initiatives to find a cure for Alpha-1 Antitrypsin Deficiency (AATD) and improve the lives of people affected by AATD worldwide. Before A1F, Scott was CEO of Global Lyme Alliance (GLA) directing business strategy, operations, revenue-generation, educational, programming, and raising awareness to advance treatments for patients suffering from tick-borne illnesses. Prior to joining GLA, Scott was President and CEO of the Bonnie J. Addario Lung Cancer Foundation (ALCF), now the GO2 Foundation for Lung Cancer, where he led the expansion from a regional lung cancer research/patient services organization to a globally recognized leader in the lung cancer community. Prior to ALCF, he was President and CEO of the American Lung Association of New York (ALANY) and spent 10 years as the Executive Director and Chief Operating Officer of the Multiple Myeloma Research Foundation (MMRF). A 30-year senior executive with experience in strategic planning, creative messaging and marketing, industry partnership development, clinical research innovation, and raising public awareness for neglected, underfunded, and often stigmatized diseases, he received an Executive Leadership Certification from Harvard Business School and holds a Bachelor of Arts in Journalism from the University of Massachusetts, Amherst.

Carmine Settembre, PhD

Telethon Institute of Genetics and Medicine, Italy

Dr. Carmine Settembre is a group leader of the cell biology and disease program at TIGEM institute and professor of histology at Federico II University of Naples. He received his PhD in Molecular Medicine from the Federico II University, Naples, Italy, and did postdoctoral work at both Columbia University and Baylor College of Medicine. The main interest of his laboratory is to understand the regulation and role of the lysosomal-autophagy pathway in both physiological and disease processes and to develop novel therapeutic approaches based on autophagy modulation.

Knut Skaar

Norway

Knut Skaar is a retired aviation professional from the Royal Norwegian Air Force and a Norwegian representative on the Council of the UN Agency for aviation, the International Civil Air Traffic Organization (ICAO) in Montreal, Canada. After retirement, his focus has been on sports, politics and inevitably with Alpha-1 Antitrypsin Deficiency (Alpha-1) as he himself is a patient. His work in this field includes being a member of the Alpha-1 Working Group of LHL where they have built awareness and knowledge about Alpha-1 among patients and their next of kin, in the national health system, and among the public and politicians. Knut contributed to establishing a new Alpha-1 Association at the Nordic level, the Alpha-1 Norden which launched its platform in 2024. His interests have led to involvement in Medicines Research Development, through participating at Eurordis Open Academy, both digital and on-site in Barcelona.

Pavel Strnad, MD

University Hospital Aachen, Germany

Dr. Pavel Strnad is a leading physician and professor at the University Hospital Aachen, Germany. Since 2008, he has been the head of his own research lab focusing on translational gastroenterology. His interests extend from transgenic animal studies to analyses of large biobanks. His aim is to identify pathways instrumental for the development of digestive disorders. In 2015, he started the European initiative for study of Alpha-1 Antitrypsin Deficiency (AATD) associated liver disease that combines experimental work with a systematic collection of human data and samples. In 2016, he was named one of the Rising Stars of United European Gastroenterology. Dr. Strnad's scientific interests include (but are not limited to) metabolic liver disease, liver cirrhosis and complications, keratins and iron metabolism. His clinical focus is on novel treatments, end-stage liver disease and liver transplantation. As a founding member of the European Reference Network on hepatologic diseases, Dr. Strnad is actively promoting the development of new therapies for rare liver disorders including AATD.

Jeff Teckman, MD

Saint Louis University, USA

Dr. Jeff Teckman is the Drs. James and Patricia Monteleone Endowed Chair, Vice Chair of the Department of Pediatrics, Professor of Pediatrics and Biochemistry at the Saint Louis University School of Medicine and Cardinal Glennon Children's Hospital. He has been involved in research on liver disease for more than 30 years in both basic science and clinical studies. Although his studies have ranged from viral hepatitis to liver failure in short bowel syndrome, his primary basic science focus has been the liver disease associated with Alpha-1 Antitrypsin Deficiency (AATD).

His laboratory was the first to recognize the link between AATD mutant Z protein in the liver and autophagy, he was the first to recognize that mitochondrial injury played a role in liver injury, and he published the first unified explanation of the mechanism of liver injury in AATD. Dr. Teckman has led several seminal studies of both pediatric and adult AATD liver disease and has consulted with pharma industry partners and the FDA. Dr. Teckman has also served the patient community as an advisor, educator, and advocate.

Alice Turner, MD, PhD

University of Birmingham, UK

Dr. Alice Turner graduated from the University of Leicester and has done postgraduate training via the Universities of Dundee and Birmingham, and Ashridge-Hult business school, completing a PhD focused on COPD and Alpha-1 Antitrypsin Deficiency (AATD) and postgraduate qualifications in medical education, leadership, and quality improvement (QI). She is now a professor in respiratory medicine at the University of Birmingham and works as a consultant in respiratory medicine at Heartlands and Queen Elizabeth hospitals, where she is lead for COPD and AATD services respectively. In addition, Dr. Turner is a member of the NIHR research prioritisation committee and has experience on a NICE health technology appraisal committee. She has published widely in COPD and AATD, and has ongoing research projects, mainly clinical trials, and observational clinical studies, in AATD and COPD funded by the NIHR and others.

Frank Willersinn

Belgium

Dr. Frank Willersinn received his medical degree from the Free University of Brussels in 1979 and was a military doctor for the German Army. In the eighties, Frank opened a Life Quality Shop in Brussels called NATURELLEMENT where people could find ecological products for their homes. After losing his arm to an infection, Frank was diagnosed in 1997 with Alpha-1 Antitrypsin Deficiency (Alpha-1). In 2008 he started to get augmentation therapy to stabilize his lungs, then in 2013, he launched the Belgian Patient association Alpha-1 Plus. That same year John Walsh initiated Frank to the Alpha-1 Community. He became a board member of the Alpha-1 Foundation from 2015-2021. Frank loves traveling. His passion today is to travel around Europe, looking for Alphas in countries like Romania, Bulgaria, and Greece and bringing them together.

Andrew Wilson, MD

Boston University, USA

Dr. Andrew Wilson is a Professor of Medicine at the Boston University Chobanian & Avedisian School of Medicine. He is a Pulmonary and Critical Care physician-scientist with a focus on regenerative medicine and stem cell biology. The goal of his research is to advance understanding of and treatment for genetic causes of chronic obstructive pulmonary disease (COPD) and the lung and liver diseases associated with the Alpha-1 Antitrypsin Deficiency (AATD), largely through the platform of patient-derived stem cells. In association with his research efforts, he has compiled the world's largest repository of AATD patient-specific induced pluripotent stem cells (iPSCs) and has shared these cells widely with researchers around the world. In addition to laboratory-based science, he also participates in epidemiological studies to define the incidence of and risk factors for both liver and lung disease associated with AATD. He likewise serves as the Scientific Director of the Alpha-1 Foundation. Finally, he is the founding director of the Alpha-1 Center at Boston University and Boston Medical Center, one of the largest clinical centers for patients with AATD in the Northeast, where he cares for patients with AATD.

Kinga Wojtowicz

RPP Group, Belgium

Kinga Wójtowicz is Director of EU Healthcare at RPP Brussels, a public affairs consultancy specializing in healthcare policies. Within her work, she has been working with oncology patient organizations and medical societies, supporting them in channeling their messages in the EU political sphere. Having a wealth of experience in creating and driving public affairs strategies, Ms. Wójtowicz has been supporting Alpha-1 Foundation in its advocacy activities at the European level.

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INNOVATION INVESTMENT INSIGHT IMPACT 30th 1995 2025

A1F 7th Global Research Conference & 10th Patient Congress

April 4 - 5, 2025

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The Alpha-1 Foundation (A1F) is committed to finding a cure for Alpha-1 Antitrypsin Deficiency (Alpha-1) and to improving the lives of people affected by Alpha-1 worldwide.

A1F has invested over $100 million to support Alpha-1 research and programs in 130 institutions in North America, South America, Europe, the Middle East and Australia.

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