

Improving Care for Alpha-1 Patients

IMPROVING CARE FOR ALPHA-1 PATIENTS

Co-hosted by MEPs Isabel Wiseler-Lima (EPP, Luxembourg) and Marisa Matias (GUE/NGL, Portugal)

23 April 2021
14.00 - 15.30 CET

In anticipation of the 2021 European Alpha-1 Awareness Day (25 April), Alpha-1 Global is pleased to collaborate with Members of the European Parliament in organising this political discussion.

Friday 23 April 2021, 14:00 – 15:30 CEST

Hosted by Isabel Wiseler-Lima (EPP, Luxembourg) and co-hosted by Marisa Matias (GUE/NGL, Portugal)

Introduction

In anticipation of the 2021 European Alpha-1 Awareness Day, Alpha-1 Global, a program of the Alpha-1 Foundation, organised, in collaboration with Members of the European Parliament (MEP) Isabel Wiseler-Lima (EPP, Luxembourg) and Marisa Matias (GUE/NGL, Portugal), a political discussion on the topic of Improving Care for Alpha-1 Antitrypsin Deficiency (Alpha-1) patients. The event, moderated by Yordan Aleksandrov, RPP Group, took place virtually on 23 April 2021.

A wide range of stakeholders attended the event, including, patients, caregivers, clinicians, academia, government representatives and industry. With EU institutions constantly looking into how to improve the lives of rare disease patients, participants provided valuable insights into Alpha-1, the disease itself and with regards to policy actions that can be undertaken to improve the lives of Alpha-1 patients.

The event proposed actions which could make a true change for the Alpha-1 patient community once implemented, especially considering the current environment provided by the forthcoming revision of the Cross-Border Healthcare Directive.

On the eve of the European Alpha-1 Awareness Day, Alpha-1 Global made a call to action targeting EU and national authorities on improving care and treatment for Alpha-1 patients. The call to action contained the following points:

1. Optimising the work of the European Reference Networks (ERN)
2. Improving diagnosis of Alpha-1 patients through EU and national level activities
3. Improving access to treatment via availability and reimbursement of augmentation therapies
4. Adopting a holistic policy approach to Alpha-1.

Welcome and opening remarks



By means of introduction, **MEP Isabel Wiseler-Lima** (EPP, Luxembourg) expressed that she was honoured to be part of the political discussion on Alpha-1. She congratulated Alpha-1 Global for their continuous work in raising awareness across key stakeholders and EU institutions, as awareness about Alpha-1 remains insufficient. She underlined the political momentum of the event as many healthcare topics are currently being considered. She noted that the upcoming

policy revisions and the issues posed by the COVID-19 pandemic provide a solid foundation for the event's discussions. As such, she stated that action at the EU level will be key for tackling rare diseases, as was also recently highlighted by the European Commission¹. Ms. Wiseler-Lima committed to working with the patient community to ensure that Alpha-1 remains present in political discussions and that upcoming policy reflects the needs of patients.

With the upcoming revision of the Cross-Border Healthcare Directive, MEP Wiseler-Lima underlined, once more, that it is the right time for stakeholders to share their perspectives and strive to make sure they are reflected in this process. Referring to the [Alpha-1 policy recommendations](#), she expressed her eagerness to hear more about how the ERNs can be improved in the future.

¹ https://ec.europa.eu/commission/commissioners/2019-2024/kyriakides/announcements/world-rare-disease-day-statement-commissioner-stella-kyriakides-0_en

MEP Wiseler-Lima introduced **MEP Marisa Matias** (GUE/NGL, Portugal), who likewise expressed her gratitude for being involved in this political discussion. She perceived the event as an opportunity for her and MEP Wiseler-Lima to listen to the needs of Alpha-1 stakeholders, in order to be aware which aspects should be emphasised in their political work. She agreed that indeed, health is a central part in today's policy agenda - an opportunity which should be grasped to create a coherent and holistic approach for Alpha-1 and other rare diseases. With this in mind, MEP Matias conveyed her firm support of the call for action put forward by Alpha-1 Global.



Setting the scene



The Alpha-1 Global Director at the Alpha-1 Foundation, Randel Plant, set the scene by outlining the priorities of the international Alpha-1 patient community.

He explained Alpha-1 Global's mission, which is to develop a collaborative global network of Alpha-1 patients, patient leaders, physicians and researchers to increase awareness, detection, early diagnosis as well as optimal access to care.

In light of the prevalence of Alpha-1 in Europe lying between 1 in 1,500 and 1 in 3,500, limited treatment and delayed diagnosis are major contributors to its burden. Accordingly, Mr. Plant noted that a consolidated approach, involving policies that facilitate sharing expertise and preventive measures, such as patient care and screening, are absolutely crucial. Notably, he unveiled Alpha-1 Global's call to action, which was developed in a timely manner as associated EU policies are being revised. The call to action incorporates the patient's perspective and includes the recommendations below. These suggestions are aligned with [Alpha-1 European Expert Recommendations](#), published in 2017 and are targeted towards EU and national authorities:

- **Optimising the work of the ERNs:** In light of the upcoming revision of the Cross-Border Healthcare Directive, Mr. Plant noted that the European Commission should consider developing a project to generate minimum credentials for Alpha-1 centres of excellence.
- **Improving diagnosis** by means of:
 - Development of an Alpha-1 diagnosis programme as part of a rare disease plan by Member States;
 - Updating the 2002 CORDIS study and issue subsequent recommendations to Member States by the Commission;
 - Consideration of newborn screening for Alpha-1 by Member States.
- **Improving access to treatment** by making augmentation therapy available and reimburse it for all patients in all Member States, as well as national legislation that does not deter individuals from genetic testing.
- **Having a holistic approach towards Alpha-1, with the help of:**
 - A comprehensive strategy on indoor air quality developed by the European Commission.
 - Improvement of ambient air quality by Member States through effective implementation of EU and World Health Organisation (WHO) developed air quality standards.

Introduction to Alpha-1 Antitrypsin Deficiency

Professor of Medicine Noel G. McElvaney from the RCSI Medical School Dublin gave an overview of the history of Alpha-1, and described its clinical and medical characteristics.



Professor McElvaney noted that Alpha-1 Antitrypsin Deficiency was discovered in 1963. It can manifest itself in two different types. As a lung disease, it is a shortage of the alpha-1 antitrypsin protein, leading to loss of function. In Alphas, there isn't enough alpha-1 protein in the lungs; the enzyme then keeps on working, attacking and destroying normal lung tissue. As this damage continues over years, lung disease such as Chronic Obstructive Pulmonary Disease (COPD) can develop. Contrarily, as a liver disease, too much alpha-1 antitrypsin protein is accumulated in the liver, resulting in a gain of toxic function. The abnormal protein becomes trapped in the liver rather than being released into the blood; the blood doesn't get as much of the protein as it needs to help other parts of the body. This makes the lungs more sensitive to damage from cigarette smoke and other air pollutants. The buildup of the AAT protein in the liver can damage the liver, which may then develop scar tissue.

He explained that when it comes to potential harmful consequences, both types of Alpha-1 could lead to outcomes such as emphysema, cirrhosis, panniculitis, the need for a transplant or even death. Moreover, he presented recent findings which have shown that for some alpha-1 antitrypsin phenotypes, smoking can increase the risk of developing COPD.

In order to optimise the diagnosis of Alpha-1, Professor McElvaney emphasised that, according to the WHO and the European Respiratory Society guidelines, all of the following groups should be tested:

- All COPD patients
- All non-responsive asthmatics (adults/adolescents)
- All patients with panniculitis
- All first-degree relatives of patients/carriers with Alpha-1

Regarding the promising treatments currently discovered, namely augmentation therapy, oral neutrophil elastase inhibitors and ribonucleic acid interference medicines, Professor McElvaney emphasised that efforts need to be undertaken in order to grant access for all patients.

Alpha-1 and the European Reference Networks



The next speaker was Professor **Joanna Chorostowska-Wynimko**, Lead of the Cross-Border Care Committee within the ERN-LUNG, as well as the Secretary General of the European Respiratory Society.

Prof. Chorostowska-Wynimko gave an overview of Alpha-1 and the ERNs. She underlined that equal healthcare provision in terms of access to diagnostics and treatment has been a longstanding issue for the EU. For Alpha-1 patients, this is unacceptable and requires urgent actions. Referring back to the call for action, she emphasised that a holistic approach is needed to address Alpha-1 issues politically. This would imply (1) a coordinated EU policy to harmonise standards on treatment and strong

rare disease plans from national governments, while (2) stimulating sharing of expertise. As part of this, she made a strong call that centres of excellence should work through networks such as the ERNs to share expertise, refer patients and educate healthcare professionals across Europe.

Prof. Chorostowska-Wynimko explained her involvement in the ERN-LUNG, a clinical care network for all rare, respiratory diseases. She is a member of the ERN-LUNG steering committee. The steering committee consists of several core groups, with one focused on Alpha-1. She stressed that the network within the Alpha-1 core group needs to be extended to other regions and additional countries, as it currently covers only nine European countries, including Spain, Germany and the UK.

In line with this, Prof. Chorostowska-Wynimko recalled the main aims of the Alpha-1 core group:

- **Group growth**, with priority for adding non-involved countries and a focus on Eastern European countries.
- **Providing highly specialised healthcare for Alpha-1 patients in Europe**, through cross-border clinical cooperation, high standards and reliability of AATD genetic diagnostics, as well as harmonisation of diagnostics.
- **Development of longitudinal European Alpha-1 registry** in close collaboration with the EARCO Clinical Research Collaboration network (which is part of the European Respiratory Society).

In the process of achieving these goals, Prof. Chorostowska-Wynimko stressed that involvement of patients is very important and necessary.

Alpha-1: My experience

Shane Fitch, CEO of the Lovexair Foundation and parent of an Alpha-1 patient, explained how life is severely affected by the condition. As a baby, her son was diagnosed with the Alpha-1 liver disease (cholestasis), which at the time was life threatening. As a consequence, he had to undergo lengthy procedures, including a risky surgical intervention to confirm his diagnosis.

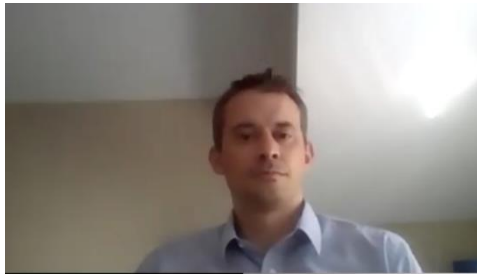


Having experienced the challenges Alpha-1 patients are facing on a daily basis, Ms. Fitch became engaged in advocacy to ultimately improve quality of life for Alpha-1 patients. She stated that education about the condition can result in better care practice, and patient networks currently in place need to be maintained and improved.

Ms. Fitch underlined the difficulties that Alpha-1 can bring to people's lives, such as, but not limited to, loss of employment, suffering social life, giving up social leisure activities and stress on close relationships. For many patients, there is little or no access to pulmonary rehabilitation in combination with limited care plans. On a positive note, she added that the COVID-19 pandemic has highlighted how many issues can be addressed and fixed with the right amount of determination.

For concrete policy suggestions, she referred to newborn screening for Alpha-1, a public policy approach to maintaining healthy lungs, treatment guidelines, e-HTA (Health Technology Assessment), and genetic counselling.

The European Reference Networks and the upcoming revision of the Cross-Border Healthcare Directive



Martin Dorazil, Deputy Head of Unit in the digital health - ERN section at DG SANTE, provided an update on the ERNs' latest developments, especially in the context of the evaluation of the Cross-Border Healthcare Directive.

Mr. Dorazil began by explaining that the 24 existing ERNs are voluntary networks of highly specialised centres dealing with rare or complex diseases. These networks are involved in activities such as, the exchange of expertise (diagnosis &

treatment), knowledge generation, research on rare diseases, as well as education and professional training. As such, ERNs serve both patients and healthcare professionals.

As the ERNs are based on the Cross-Border Healthcare Directive, the evaluation of the latter will consider whether the legal provisions of the former are still fit for purpose. The evaluation will also look into the ERN's efficiency in providing added value for patients. Moreover, he noted that the adopted EU4Health programme provides funding for the ERNs for the period 2021-2027, as they are mentioned as one of the specific objectives to be supported under the programme.

As to next steps, Mr. Dorazil reported the following actions on the ERNs:

- Complete enlargement of the geographical scope and diseases coverage
- Integration of ERNs into national healthcare systems
- Knowledge generation actions such as Clinical Practice Guidelines and trainings
- Support ERNs' research activities and link their registries with the European Health Data Space (EHDS)
- Demonstrate the added value of the ERNs through evaluation and monitoring

To conclude, Mr. Dorazil encouraged everyone to contribute to the consultation of the evaluation of the Cross-Border Healthcare Directive.

Open Floor for Discussion



During the open floor discussion, participants were provided the opportunity to ask questions to the speakers.

The first question asked about the extent ERN-LUNG and other EU based structures involve patients to identify and solve unmet needs. This was a

question posed to Prof. Chorostowska-Wynimko. Prof. Chorostowska-Wynimko noted that indeed, patients are an important part of the network, as they are members of the core groups and involved in the leadership, representing a strong patient voice in the ERN-LUNG. She added that while patient involvement is a very important and fundamental aspect, support for collaboration centres in regions of high unmet needs should be considered. This is a consequence of the low coverage of ERNs to date, manifesting in a high concentration of centres in central Europe and considerably less on the Eastern peripheries of Europe.

Prof. McElvaney was asked about research concerning repairing and recuperating damaged lung tissue.. Prof. McElvaney explained that a lung is very fragile and does not heal well, hence concluding that regrowing a lung is not possible with the resources available to date.

The two following questions were addressed to Prof. Chorostowska-Wynimko. The first question asked if ERN-LUNG could help improve access to augmentation therapy and facilitate receiving it, if available, in a neighbouring country within the next 10 years. This practice is currently only performed in 17 out of 47 European countries. She explained that access to treatment depends very much on local policy or reimbursement policy. However, ERN-LUNG can play an important part in changing the situation by ensuring diagnostics are accessible, providing accessible clinical care and expertise. The second question inquired about the extent of disparities in access to specialised care for Alpha-1 patients. Prof. Chorostowska-Wynimko explained that disparities exist, referring to Poland as an example of a country that does not have access to treatment and diagnostics, concluding that there is much to be done by the EU, healthcare professionals as well as patient organisations.

The final question was posed to Mr. Dorazil, and asked about the role of public consultations as part of the revision of the Cross-Border Healthcare Directive, as to ensure that the views of patients are accommodated. Mr. Dorazil reported that indeed the patient's perspective and the views of other stakeholders will be key assets to this process. He noted that the public consultation will be launched in May 2021 and he encouraged everyone, once again, to contribute. He also explained that the first periodic 5-year assessment of performance of ERNs and their Members, will begin in 2022 and the final report will be published in 2023.

1. Concluding remarks



To conclude the event, MEP Wiseler-Lima thanked all panellists for their valuable contributions, and drew attention to the promising forecasts brought by the revision of the Cross-Border Healthcare Directive. She underlined that adequate implementation of the findings from this revision will be critical and will require commitment from both EU and national levels.

MEP Marisa Matias also praised all contributors and expressed that the event has reinforced her intentions to improve the lives of Alpha-1 patients by supporting the requests of stakeholders and working collaboratively with them. She likewise pointed out that the information shared by the EU Commission was very promising, but it should be ensured that patients and experts are involved in all stages of policy-making, not only for its implementation but also its development.

