



REPORT

EU Roundtable on Alpha-1 Antitrypsin Deficiency (AATD)

Marking the first European Alpha-1 Awareness Day

Wednesday 25th of April 2018, 9.30-12.00

Martin's Brussels EU Hotel, Brussels

CSL Behring
Biotherapies for Life™



On Wednesday 25th of April 2018, specialists and patient representatives met to discuss how to share best practices in the care of Alpha 1 Antitrypsin Deficiency (AATD). The roundtable was organised by CSL Behring. The meeting was held **in support of the first European Alpha-1 Awareness Day**, and in conjunction with the launch event at the European Parliament, hosted by Polish Member of European Parliament, Marek Plura. The roundtable was moderated by Alpha-1 Global; with an active participation from the Alpha-1 patient community across Europe, experts in AATD, as well as EU organisations representing the liver and COPD communities.

The meeting set two main objectives:

- facilitating the dialogue between experts and patient representatives and
- encouraging collaborations between stakeholders at EU and national levels.

The first session focused on the presentation of the disease, burden and challenges to testing and treatment, and on the work of the [European Reference Networks \(ERNs\) LUNG](#) and [ERN RARE-LIVER](#). Alpha-1 Antitrypsin Deficiency (AATD) is a genetic inherited condition occurring in people with a deficiency in the Alpha-1 antitrypsin protein (AAT) in the blood, which can cause liver disease and lung disease and which is potentially life-shortening. While there is currently no cure, treatments are available to better manage the disease and slow down the progression of the organs damage, such as plasma-derived therapy for Alpha-1 lung and skin affected patients. **Professor Robert Stockley**, from the Lung Investigation Unit Medicine of the University Hospitals NHS Foundation Trust and Queen Elizabeth Hospitals in Birmingham (UK) stressed the **importance of centres of excellence and multidisciplinary care** in the management of this complex disorder, as well as of **early diagnosis and access to treatment** before the patient's health starts deteriorating and becomes debilitating. He also referred to the importance of organising national and international registries to improve knowledge about the disease and optimal management.

Dr. Frank Willersinn, from Alpha-1 Plus Belgium, presented the process that led to the development and adoption of the Alpha-1 [European Expert Group Recommendations on AATD](#) in 2017. While AATD remains a rare disease which largely goes underdiagnosed and access to AAT treatment continues to be challenging in some EU countries, **low awareness of recommended health and lifestyle guidelines** in Alpha-1 patients (e.g. smoking cessation, avoiding sources of indoor and outdoor pollution, or alcohol consumption) before they receive a correct diagnosis is often challenging. The **translation of the Expert Recommendations in 10 European languages** will help improve knowledge and awareness. The information will be made available to specialised healthcare professionals, including lung physician associations and medical schools in various EU countries, through the work of the national Alpha-1 patient associations.

P.D. Pavel Strnad, from the University Hospital of Aachen (Germany), presented on the AATD related liver disease, which is not well defined and commonly overlooked because liver enzymes are usually within normal limits, thus both physicians and patients are not aware of their liver affection. He provided insight on the work of the ERN RARE-LIVER and on the EASL study group on AATD-related liver disease, which **stimulates awareness at an EU level and involves a growing number of recruitment centres**, like in Portugal and Denmark. Preliminary results demonstrate that homozygous Pi*ZZ genotype is a frequent cause of fibrosis and steatosis and is a relevant disease modifier; obesity and alcohol consumption are additive risk factors. He further **invited patients and experts to interact and collaborate with the ERN RARE-LIVER group** to ensure the best possible outcomes of the study.



The Q&A session brought in the discussion some further points for consideration when reflecting on improved data collection systems. **It was noted that existing data could be better exploited, and that an EU protocol to collect information homogeneously throughout Europe could help step uniform standards for interoperable data.** An insufficient number of specialist multidisciplinary centres in certain countries can be observed, while funding and administrative formalities remain barriers to improve data collection.

The second session of the meeting aimed to brainstorm ideas on **how to increase awareness and education among general practitioners and specialists and how to best utilize the ERNs to benefit the Alpha-1 community.**

Dr. Miriam Barrecheguren, from the Department of Pneumology of the University Hospital Vall D'Hebron in Barcelona (Spain) introduced the session, outlining the **long delays in diagnosis despite the 1997 WHO recommendations to screen all patients with COPD for AATD.** Reasons for under-diagnosis include the limited awareness of the disease and methods of testing, misconception on the inefficacy of existing treatments and complex testing algorithms.

Education, testing rates and awareness amongst physicians can be improved via:

- systematic screening for certain categories of patients (e.g. all COPD patients);
- detection studies and reference centres;
- media campaigns or instructional programs via web;
- free diagnostic kits;
- and presentations at international and national meetings.

Options explored during the discussion also included:

- targeted education for specialist nurses;
- improving communication with and via other patient organisations at EU level and national level, such as the liver and respiratory patient groups;
- leveraging science from congresses to increase knowledge amongst healthcare professionals, patients and policy makers;
- building on national best practices in other rare diseases to drive awareness and education.

Karen O'Hara, Chair of the Alpha-1 UK Support Group, facilitated the final discussion on the promotion of the ERNs to healthcare providers and patients. To introduce the exchange, **Professor Joanna Chorostowska-Wynimko**, from the National Institute of Tuberculosis and Lung Diseases in Warsaw (Poland), further elaborated on the ERN LUNG and the AATD Core Network, with insight on the main centres and participating centres involved in the Network. ERNs have just completed their first year, but **not all EU countries are represented in ERN-LUNG, and there is an uneven representation across the regions of Europe particularly in the AATD core network.**

Although the core value of the ERNs lies on patients, awareness about the **ERNs amongst patients remains low**, noted **Karen O'Hara**. More strategies to target patients are expected to increase the exchange of information via platforms, software, social media patient groups, through the work of the European Patients Advocacy Groups (EPAGs). Participants discussed several suggestions and opportunities, such as:

- improved communication between the AATD core network of ERN-LUNG and the patient community;
- exploring the potential of field visits/fellowship programmes for specialists between the ERN reference centres;
- diversifying target groups;
- leveraging other rare disease awareness days and synergies with other relevant initiatives.

Concluding the discussion, **Gonny Gutierrez**, Director of Alpha-1 Global, pointed out that EU policy makers can further stimulate actions at national and EU levels and contribute to respond to this under-diagnosed and under-treated disease. Several good practices in education and awareness already exist, and the Council of Europe and other EU institutions can contribute to lead and harmonise standards of care. **Dialogue and clinical research collaborations are to be continued towards the establishment of a uniform patient register, greater quality standards for diagnostics, greater involvement of countries in the ERNs and improved dissemination of information amongst patients in local languages.** Alpha-1 Global and partners remain committed to improving Alpha-1 diagnosis, management, treatment and care throughout Europe.



