



EU Alpha-1 Awareness Day

24 April 2018

European Parliament, Brussels

Introduction

On Tuesday 24 April 2018, Member of the European Parliament (MEP) **Marek Plura (EPP, Poland)**, in collaboration with the European Alpha-1 Community, organized an EU roundtable discussion to raise awareness for the rare and genetic disorder of Alpha-1 at the European Parliament in Brussels (Belgium). The event celebrated the first EU Alpha-1 Awareness Day, supported by **MEPs José Inácio Faria (EPP, PT), MEP Wim van de Camp (EPP, NL), MEP Seb Dance (S&D, UK), MEP Jytte Guteland (S&D, SE), and MEP Sirpa Pietikäinen (EPP, FI)**. The roundtable discussion presented an opportunity for national Alpha-1 patient organizations and MEPs to discuss the challenges of Alpha-1 patients in the individual EU Member States. Presentations were given by Alpha-1 leaders from Poland, Spain, Italy, the Netherlands, Belgium, Romania, and the UK, to create a clear image of the burden and difficulties of the Alpha-1 condition.

Alpha-1 antitrypsin deficiency is a rare genetic disorder that increases the risk of lung, liver and other diseases. Alpha-1 is present worldwide, but the prevalence of its severe form varies across Europe, affecting about 1 in 1,500 to 3,500 individuals, whereas mild forms are much more common.

The principle difficulty in managing Alpha-1 is achieving early diagnosis, something which most patients will not receive until severe symptoms have begun, despite the cheap and effective means of diagnosis that exist. Alpha-1 can be a life-threatening disease, and without proper treatment and care patients may have a decreased life expectancy.

Opening remarks



MEP Marek Plura opened the roundtable by thanking and welcoming all participants to the first ever EU Alpha-1 Awareness Day. Mr. Plura noted that like other rare conditions, Alpha-1 antitrypsin deficiency affects only a small group of the population, and for a long time the needs of such patients were neglected due to the limited knowledge about rare diseases. As a result, Alpha-1 patients often experience delays in diagnosis and have difficulty finding information about their condition and inadequate access to expert care.

Gladly, the perspective on rare diseases is currently changing in the EU, where Member States are launching national rare disease plans, the EU is funding and incentivizing research in clinical trials, and through the formation of the European Reference Networks. He further recalled the fact that Europe has an important role to play in the establishment of good practices in healthcare, as most responsibilities on organising and management diagnosis, care and treatment of the diseases belong to the Member States. Therefore, it is important to share knowledge and experience of patient advocates at a national level. To this end, Mr. Plura emphasised the importance of the Alpha-1 Awareness Day, which gave the best opportunity to create insights on key challenges for Alpha-1 and to share good practices on the implementation of the European Alpha-1 Expert Group Recommendations.

Gonny Gutierrez addressed participants on behalf of **Alpha-1 Global**. She expressed her gratitude to **MEP Marek Plura** for hosting the event, **MEP José Inácio Faria** for his support and all other MEPs who unfortunately could not be present at the roundtable discussion but contributed to the discussion via the recording of videos. These MEPs were **MEP Wim van de Camp (EPP, the Netherlands)**, **MEP Seb Dance (S&D, the United Kingdom)**, **MEP Jytte Guteland (S&D, Sweden)**, and **MEP Sirpa Pietikäinen (EPP, Finland)**.



Presentations



Robert Durlik from Alpha-1 Poland familiarised the participants with the objectives of Alpha-1 Poland which are to cooperate and spread knowledge among the public and health professionals. This is done through various methods including informational leaflets and active social media presence. Mr. Durlik noted that in Poland one of the main challenges for Alpha-1 patients is early-diagnosis, since highly experienced specialists in the field are lacking. Additionally, there is very limited access to modern treatment, such as augmentation therapy, which

unfortunately is not reimbursed. In Poland, the system of modern and professional care is still in development. There is a lack of medical centres focused on Alpha-1. **The key policy asks of Polish Alpha-1 patients are: to increase the budget for the medical aid and research & development, to reimburse the expensive and modern therapies, to educate professionals and patients, to create specialized medical centres, and to change legislation to simplify the procedures of adopting new drugs.**

Elena Goyanes from Alpha-1 Spain explained that her organisation cooperates with FEDER and FENEAR, two Spanish national associations specialized in rare diseases and respiratory diseases. Ms. Goyanes noted that Alpha-1 is not included in the rare diseases strategy in Spain. Furthermore, there are only three centres of excellence to cover seventeen regions while augmentation therapy is only available for 430-450 patients. She stressed that there is a lack of education and knowledge of prevention programs among children and adolescents. Difficulties exist in testing and genotyping. **The key policy asks for Spanish Alpha-1 patients are: ensure early diagnosis, increase awareness on how and when to test, increase knowledge among healthcare professionals, create access to augmentation therapy, ensure patient support and education, launch prevention programmes, and establish more centres of excellence.**





Cristina Barbiero from Alpha-1 Italy reflected that in Italy Alpha-1 is unknown. Nonetheless, it is expected that more than 5000 people are affected by Alpha-1. Ms. Barbiero explained that the national association promotes multiple informational Alpha-1 days every year together with an annual national assembly. Two screenings were promoted and recently a poster was developed, which will be distributed to all pneumological clinics. The biggest issue in Italy is the decentralization of the health system. **Therefore, the policy asks in Italy are to create**

reference centres in all regions with clinical knowledge and expertise on Alpha-1, to standardize diagnosis with centralized registry and protocols, to standardize oxygen devices, to have therapy continuity in all Member States, and to improve awareness.

Heleen Groen from Alpha-1 the Netherlands explained that in the Netherlands through Facebook and other social media platforms, awareness is being raised on Alpha-1. Additionally, there is a lot of work to be done when it comes to spreading the knowledge on Alpha-1 among healthcare professionals. In the Netherlands, only a few pulmonologists have the proper knowledge on Alpha-1. Therefore, one of the efforts taken by Alpha-1 the Netherlands is to ensure proper education for general practitioners. Family doctors are taught to recognize the symptoms for Alpha-1 so that they can refer them to the specialists. Another challenging issue in the Netherlands is the differentiation of reimbursement of augmentation therapy, depending on the genotype. In addition, she touched upon the relation between organ donation and Alpha-1. Since Alpha-1 can cause lung-, liver-, and skin diseases, they might be perceived eligible to receive new organs. Recently, a policy change has been made in the organ donation registration process, which might influence organ donation and consequently Alpha-1 patients in the Netherlands. **The key policy ask for the Netherlands is to have equal and easier access to treatment for all patients.**





Frank Willersinn from Alpha-1 Belgium, elaborated on the composition of the Belgian patient organisation for Alpha-1, which consists of 15 members who represent 32,000 patients in 54 regions. He informed participants that in Belgium the health commission has decided to not reimburse treatment post 2010 diagnosis. Therefore, a situation exists in which patients are denied access to treatment depending on when they were diagnosed. Furthermore, Mr. Willersinn noted that there is no French speaking patient organisation for COPD patients, which is a

common consequence of Alpha-1. **The Belgian policy asks are to reimburse augmentation therapy, to establish a French speaking COPD association, to enable home treatment, to respect guidelines, and at EU level to allow and ensure qualified cross border health care.**

Simona Olteanu from Alpha-1 Romania announced that Romania has officially established their national Alpha-1 patient association. The small Alpha-1 community consists of specialized Alpha-1 physicians, less than 10 severe Alpha-1 patients, relatives of patients who are carriers, and patients with rare mutations. In Romania, the diagnostics can be determined by genetic testing, which is only available in private practice and from which the costs need to be covered by the patient. Unfortunately, there is no treatment available in Romania: augmentation therapy is not reimbursed in the public insurance health system. Key policy asks are in development and currently Alpha-1 in Romania is mainly occupied with introducing their patient association in the country. Once this has happened, the first ask shall be on reimbursement and standardization of treatment.



Karen O'Hara from the Alpha-1 UK Support Group explained that her group was established in 1997. The support group currently has over 600 members and provides emotional and practical support, which is done through social media, and communication through telephone or email. Furthermore, events are organised with the objective of giving insights on developments in research. In the UK a specialized process is currently under development to improve the standardized care for Alpha-1. One of the problems in the UK is incorrect diagnosis. Alpha-1 is

often not recognized by healthcare professionals. Therefore, it is important to spread education and knowledge on Alpha-1 among medical specialists. She emphasized that even though the European Reference Networks are very much welcomed, patient voice and consultation should be taken even further into consideration.

Professor Rob Stockley, Alpha-1 medical specialist from the UK, highlighted that it is extremely important that healthcare specialist and decision-makers work together with patient groups.



Discussion and concluding remarks



Participants noted the importance of hearing the rare disease patients' voices in the European Parliament. **MEP José Inácio Faria (EPP, Portugal)** emphasised the need for continuous engagement between patients and policy-makers. Fully supportive of the Alpha-1 rare disease community, Mr. Faria stressed furthermore the crucial position of patients in sharing with European politicians their specific needs and challenges. As the shadow rapporteur of the European Parliament Own-Initiative Report on EU Options for Improving Access to Medicines, Mr. Faria congratulated the Alpha-1 community on their first EU Alpha-1 Awareness Day and expressed his willingness to continue meeting patients on the occasion of such crucial events, which can build a strong basis for political engagement with the European Parliament. Strong political relationships are a cornerstone part in improving the quality of life of rare disease patients all around Europe.

MEP Marek Plura concluded the event by thanking all participants. He noted that he will continue to support the rare disease community and informed participants about his crucial work on the establishment of a rare disease committee at the national level in Poland.





List of participants

European Parliament

- Marek Plura (EPP, Poland)
- José Inácio Faria (EPP, Portugal)
- Luigi Daniele, Office of Dr. Elena Gentile MEP
- Elvira Hernández Toledo, Advisor European Parliament (Spain)
- Dalia Jadowska, Office of Marek Plura MEP
- Lukas Kotwica, Office of Marek Plura MEP
- Andreea Malael, Office of Daciana Sarbu MEP (Romania)
- Magdalena Ryalska, Office of Marek Plura MEP

External participants

- Yordan Aleksandrov, RPP Group
- Cristina Barbiero, Alpha-1 Italy
- Sophie Böger, RPP Group
- Robert Durlík, Alpha-1 Poland
- Fran Franco, Grifols
- Ella Geiblinger, Alpha-1 Austria
- Elena Goyanes, Alpha-1 Spain
- Kit Greenop, RPP Group
- Heleen Groen, Alpha-1 the Netherlands
- Esperanza Guisado, Grifols
- Gonny Gutierrez, Alpha-1 Global
- Sabina Janciauskiene, Researcher Germany
- Neele Jongen, RPP Group
- Joelle Khraiche, CSL Behring
- Sabrina Lannazzone, Interel
- Frederike Ludwig, RPP Group
- Jelena Malinina, RPP Group
- Karen O'Hara, Alpha-1 UK
- Simona Olteanu, Alpha-1 Romania
- Karl Petrovsky, PPTA
- Renate Shashoua, Alpha-1 Switzerland
- Robert Stockley, Physician UK
- Pavel Strnad, Alpha-1 Global
- Charles Waller, RPP Group
- Marion Wilkens, Alpha-1 Germany
- Frank Willersinn, Alpha-1 Belgium