

REPORT ON

European Parliament Interest Group Meeting on Rheumatic and Musculoskeletal Diseases
on

“Policy issues and challenges in the treatment and care of rare diseases”

European Parliament (Brussels), Altiero Spinelli Building, room ASP 1G2,
6 December 2016, 12:30 – 14:30

Mr Takis Hadjigeorgiou (MEP, Vice-Chair of the Interest Group) opened the meeting by thanking the distinguished speakers, in particular the representatives of the European Commission and the European Parliament. Mr Hadjigeorgiou pointed out that the meeting focused on the treatment and care of rare diseases which do not receive the attention they deserve, both publicly and politically. The term “rare diseases” denotes all conditions which affect only 1 out of 2000 persons. This number may seem small individually, but across the EU this means that up to 36 million people suffer from one of the 7.000 rare diseases already discovered. He also underlined that access to health care is magnified for those with rare diseases, given the little understanding of these illnesses and the scarcity of specialists and medical centres.

Mr Jaroslaw Waligora (DG Health and Food Safety, European Commission, Programme Management and Diseases) opened his presentation by stressing the potential for improving effectiveness and efficiency when it comes to the European strategy on rare diseases. He was satisfied with the positive development of the national plans and strategies. In 2009 only 4 EU countries had national plans in place, while there are 23 in 2016. The European Commission has also decided to set up an expert group on rare diseases, which is composed by Member States representatives, patient’s organisations, producers, scientific societies and individual experts. This group meets two or three times per year and has already made a crucial impact on the work of the Commission. The health care for rare diseases has improved and more than 120 orphan medicinal products are being authorised. “*The European Reference Networks*” links national health care providers and allows them to improve the quality and access to highly specialised healthcare for the European citizens. The key features of the Networks are that they are patient centred and clinically led, they are assessed by 3rd parties and endorsed and approved by National Authorities.

Ms Ana Rath (Coordinator of the Scientific Secretariat, International Rare Diseases Research Consortium Director) started her presentation by noting that there are specific problems related to Rare Diseases, including recruitment of health care professionals, regulatory and organisational issues and the increasing costs and time required for the development of evidence and treatments. In terms of regulatory challenges there are different legal and regulatory frameworks in the EU. This situation affects patient information and consent, clinical site activation, insurance acquisition and human and data protection compliance. In order to generate solutions in building evidence based clinical practice for Rare Diseases a multi-stakeholder involvement and academic research environment is highly desirable. Furthermore, the mapping of the international expertise and



capacity, as well as good quality systematic collection of patients would have crucial impact in the future. All stakeholders have to be involved in the process of building evidence, and the data should be validated and standardized. Not only the clinical staff, but patients as well, have to be trained and made aware of the importance of clinical research.

Prof Marta Mosca (On behalf of the ReCONNET, University of Pisa) presented the challenges ahead for research and development in rare diseases. Due to different phenotypes and heterogeneity of clinical manifestations there are long delays in the diagnosis. Furthermore, literature data show that the implementation of recommendations in clinical practice is not homogeneous and may lead to disparities in health care and outcomes. ReCONNECT network facilitates access to better and safer healthcare, defines proper organisational assessment and identifies standard and cost-effective pathways for the management of Rare Diseases. The main goals include increasing the empowerment and engagement of patients; improving knowledge on epidemiological, clinical and therapeutic issues and facilitating information sharing and circulation. Similarly, the mission and vision of a European Network of centres of Excellence in rare and complex hereditary and autoimmune connective and musculoskeletal diseases aim to optimize the availability of resources and to deliver cost effective care.

Ms Kirsten Lerstrom (Lupus Europe) stated that there are approximately 500,000 citizens already diagnosed with lupus in Europe. Lupus is a highly complex and complicated disease that affects almost any organ and body. There is only one compound in the last 50 years. There are currently more than 100 open lupus trials on, but so far only "*belimumab*" has passed the third phase. Patients are able to do more when they know more about their diseases, therefore Lupus Europe has organized two highly successful conventions: "*Tame your wolf- tame your lupus*" in 2015 and "*Kick Lupus*" in 2016. These events promoted adherence to treatment and more involvement of the patients into the projects and trials. Lupus Europe has also brought to life a task force dealing with research and clinical trials.

Ms Karin Kadenbach (MEP, Member of the Interest Group on Innovation in Health and Social Care) emphasized that rare diseases don't get enough political attention at national level. This often leads to wrong diagnoses and false treatment. For this reason, she noted, the European Reference Network is a highly important and significant tool. It is also important to mention that especially patients with lower income are not able to access health centres.

Mr Takis Hadjigeorgiou (MEP, Vice-Chair of the Interest Group) closed the meeting by stating that the European Union can provide real added value through its competences, and the Members of the European Parliament must make sure that these competences are put into practice. The establishment of European Reference Networks for rare diseases, which are to roll out in 2017, will provide support to healthcare professionals in finding better treatment for these diseases.