Paving the way for rare diseases patients: Addressing Unmet Needs for an Improved Quality of Life and Care

14 November 2023, 14:00-16:00 CET
European Parliament (room 3H1)

Event Key Highlights

- The event sought to bring together stakeholders from public, private, and civil society sectors to discuss recent research findings on patients' perspectives regarding the socio-economic impact of scleroderma and their care journey, whilst also highlighting potential benefits from current health-related legislative proposals.
- The event emphasized the vital significance of comprehensive treatment funding. This requires healthcare systems to fully finance pharmacological and non-pharmacological treatments, ensuring that patients can access comprehensive and funded healthcare solutions, thereby averting financial burdens.
- The event speakers acknowledged the importance of the European References Networks (ERNs) in enhancing access to care, improvement of the diagnostic processes and clinical research. They agreed that the EU must encourage Member States to incorporate the ERNs into their national health systems.
- Speakers also emphasized that recognizing and addressing the psychological impact of rare diseases in health policy is crucial to improving patients' quality of life. This recognition not only positively influences the individual's overall wellbeing but also plays a pivotal role in the efficacy of treatment and the recovery process.
- The discussion addressed the recent opportunities presented by the revision of pharmaceutical legislation. It was emphasized that to seize these opportunities, it is crucial to integrate the patient voice at every phase of the decision-making process.
- The discussion delved into the definition of high and unmet needs. It was observed that existing definitions are vague, potentially heightening uncertainty for drug developers and diminishing their efficacy in encouraging innovation and impact. Conversely, it was highlighted that a more specific definition might involve criteria open to interpretation by Member States, such as quality of life, which is intricately tied to healthcare costs.

Opening Plenary

On 14th November, MEP Patrizia Toia (S&D, IT) hosted an event organized by the Federation of European Scleroderma Associations (FESCA) **on "Paving the way for rare diseases patients: Addressing Unmet Needs for an Improved Quality of Life and Care".** The event's main objective was to discuss the results of FESCA's European-wide survey on unmet medical needs of the scleroderma patient community and how these can be addressed through policy initiatives. The event brought together public, private, and civil society stakeholders.

MEP Patrizia Toia (IT, S&D) emphasized policymakers' crucial role in supporting rare disease communities as people living with such conditions often face unique challenges, from timely diagnosis and access to care, to prevention and treatment. MEP Toia welcomed some of the

policy initiatives that seek to address these challenges, such as the recent comprehensive approach to mental health that has been launched by the European Commission, as well as other initiatives, such as the European Partnership on Rare Diseases, the important work undertaken by the European Reference Networks and the upcoming review of the pharmaceutical legislation. MEP Toia also highlighted that the challenges faced by patients living with rare diseases, such as scleroderma, could also be addressed through an updated cohesive strategy that would bridge national and European legislation, policies, and programs across various domains.

The event continued with a short video, released by FESCA to mark World Scleroderma Day 2023, which showcases the journey that patients living with scleroderma go through, from diagnosis to treatment and finding the light to bloom.

Following the video, **Mr. Michael Oeschger**, **Chairman of German Scleroderma Association and patient living with scleroderma**, shared his 17-year struggle with the diseases. He highlighted the challenges of symptom onset, misdiagnosis, and the crucial need for early detection to mitigate severe consequences. Advocating for patient-centered policies, Mr. Oeschger stressed the importance of access to specialist care, emphasizing multidisciplinary support and proposing the establishment of rare disease specialist centres. He emphasized the financial hurdles, such as insurance coverage for specialized treatment, and urged policymakers to take into account the economic and psychological dimensions of living with a rare disease.

Ms. Sue Farrington, President of FESCA, presented the results of the survey, which was conducted in 20 countries and 16 languages, yielding more than 1200 responses and identifying critical challenges. The report emphasises the need to empower patients through shared patient-physician decision-making, and advocates for improving outcomes through timely diagnosis, equitable access to clinical trials and approved treatment options, as well as expanded access to multiprofessional care. Moreover, the report looks into the financial implications of living with scleroderma, and calls for comprehensive healthcare and labour market policies for those affected.

MEP Aldo Patriciello (IT, EPP) shared a video message to express his support to FESCA and the scleroderma patient community. In his message, MEP Patriciello emphasized the multiple effects of scleroderma on the physical, financial, and psychological well-being of patients and underlined the importance of treating rare diseases from a socio-economic perspective. MEP Patriciello stressed the need for the European Union to invest in research and development of rare diseases therapies, to improve access to timely diagnosis and treatment, and to include socio-economic and labour market policies in health strategies.

Panel I – Unveiling the Hidden Challenges: Exploring the Social Impact of Scleroderma on Quality of Life and Holistic Well-being

- Ms Donata Meroni, Health Monitoring and Cooperation, Health Networks Head of Unit, DG SANTE, European Commission
- Ms Ilaria Galetti, Federation of European Scleroderma Associations (FESCA)
- Dr. Linda Kwakkenbos, Co-Director of the Scleroderma Patient-centered Intervention Network

Ms. Donata Meroni, Head of Unit Health Monitoring and Cooperation, Health Networks, at DG SANTE, European Commission, provided an overview of the European Union's initiatives to tackle rare diseases, including the work of the ERNs, the launch of a €18 million

action to integrate the ERNs into national healthcare systems, the creation of the EU Platform for Rare Diseases Registries, the proposal on the pharmaceutical legislation reform and the potential of the European Health Data Space. Ms. Meroni highlighted that the European Commission's objective is to always put the patient at the center. She highlighted that more than 400 clinical practice guidelines have been developed with the active involvement of patient organizations, gathering knowledge on best care pathways for patients. These joint efforts represent support a comprehensive approach to address rare diseases and improve healthcare in the European Union.

Dr. Linda Kwakkenbos, Radboud University, introduced the Scleroderma Patient-centered Intervention Network (SPIN) initiative, which involves patient organizations, partners, researchers and healthcare professionals. The main objective of the project is to develop, adapt and test new and existing programs to help people with scleroderma cope with their illness and manage their daily lives. Dr. Kwakkenbos highlighted that many people living with scleroderma do not have access to important treatments, including psychological therapy, self-management programs and physical and occupational therapy interventions. She pointed out that SPIN has responded to this gap by developing e-health programs to help the scleroderma community with disease management and overall healthcare management. Dr. Kwakkenbos also acknowledged the ERNs' positive commitment to improving diagnosis and treatment, and conducting disease research, but expressed concerns about their predominant biomedical focus. She therefore emphasized the need for a more holistic approach and advocated for a broader focus on education and psychosocial wellbeing for people living with rare diseases, such as scleroderma.

As a representative of the scleroderma community, **Ms. Ilaria Galetti**, **Vice President of FESCA**, acknowledge the current challenges experienced by patients and the need to integrate the ERNs into national healthcare systems to ensure equal access to treatment. Ms. Galetti also called for increased psychological support and access to non-pharmacological therapies, as well as more recognition of the crucial role of family carers. She emphasized the need for patient-centered legislation and the inclusion of proactive strategies, especially for long-term conditions such as systemic sclerosis. Ms. Galetti also highlighted that, regarding employment, the emphasis should be on facilitating patients' access to work and fostering a mindset where those affected learn to manage the disease actively rather than passively awaiting a cure.

Panel II: Breaking Barriers in Scleroderma Care: Addressing Unmet Medical Needs

- Ms. Sue Farrington, Federation of European Scleroderma Associations (FESCA)
- Ms. Izabela Taborska, Medicines: Policy, Authorisation and Monitoring Unit, DG SANTE, European Commission
- Mr. Victor Maertens, Government Affairs Director, EUCOPE

Ms. Izabela Taborska, Legal Officer in Policy, Authorisation and Monitoring Unit, at DG SANTE, European Commission, started with an overview of the most important provisions of the Commission's proposal to revise the pharmaceutical legislation. She explained that incentives, particularly in the form of regulatory data protection and market exclusivity, could play a crucial role in the development of new therapies that can improve patient outcomes. Moreover, she highlighted that the proposal provides for a nuanced approach to defining unmet medical needs, which could allow for a six-month extension of regulatory data

protection. For cases where there is a high unmet medical need, a greater incentive is proposed, which provides for a 12-month extension of regulatory data protection and market exclusivity. Ms. Taborska underlined that the two definitions may seem broad due to the absence of societal elements. However, it is the intention of the European Commission to provide definitions that are not based on the specificities of the Member States. For this reason, quality of life is not considered, as this is related to the costs of healthcare that are specific to each national healthcare system. Instead, the definitions are centered on the intrinsic needs of the patient. This targeted strategy is intended to encourage pharmaceutical companies to invest in areas where there is significant unmet medical need.

Ms. Sue Farrington, **President of FESCA,** welcomed the pharmaceutical reform proposal and emphasised its potential to drive research and innovation, particularly in underserved areas of rare diseases, such as scleroderma. However, she emphasized the importance of refining the definition of unmet medical needs and high unmet medical needs. For Ms. Farrington, the unclear definition poses a challenge in assessing drug development, rendering it more intricate and less predictable. This heightened complexity introduces increased uncertainty for medicine developers, thereby limiting the impact of incentives in spurring innovation. Moreover, she strongly advocated for the involvement of patients and patient organisations in decision-making processes that impact their lives, and stressed that collaboration and engagement of all stakeholders from the outset is essential to achieve equitable access to treatment and care.

Mr. Victor Maertens, Government Affairs Director at European Confederation of Pharmaceutical Entrepreneurs (EUCOPE), emphasized the need to build on the successes of the pharmaceutical industry in the treatment of rare diseases. He stressed the importance of a nuanced approach, especially with regard to the Commission's proposed concept of modulation for high unmet medical needs. Mr. Maertens expressed concern about the lack of clarity around the definition of high unmet medical need and pointed to the different interpretations of the concept by stakeholders. Nevertheless, Mr. Maertens raised the question of the practical feasibility of a simultaneous market launch of orphan therapies in all 27 Member States, especially for smaller companies. He argued in favour of exploring best practices beyond legislation by working together with the ERNs and the National Competent Authorities on Pricing and Reimbursement (NCAPR). Mr. Maertens concluded by underlining the need for constant dialogue and collaboration between research, industry, patients, and healthcare professionals to find innovative solutions for rare diseases.

Wrap-up and Closing Remarks

Ms Farrington, President of FESCA, concluded the event by thanking all the participants and speakers for their attendance, and summarized FESCA’s report with three key points. First, she highlighted the importance of sustaining funding for European Reference Networks (ERNs) to enhance diagnostic procedures. Second, she emphasized the need to integrate the patient perspective in all stages of drug development, regulation, and policy to drive research and innovation. Lastly, Ms Farrington underscored the necessity of inclusive funding for treatments to broaden access and ensure diverse patient groups can benefit from comprehensive healthcare solutions.