RADDIAL

RARE DISEASE DIAGNOSIS ALLIANCE

Event Report

POLICY EVENT :

JOINING FORCES FOR A BETTER DIAGNOSIS OF RARE DISEASES

Thursday, June 29th, 11:30-14:30, 2023
House of Representatives (Erasmus Room)

Takeda sanofi janssen Chiesi global rare diseases Alnylam PHARMACEUTICALS RaDiOrg RARE DISEASES BELGIUM

Members of the Rare Disease Diagnosis Alliance

On behalf of the Rare Disease Diagnosis Alliance, Stefaan Fiers offered a brief word of welcome, thanking all speakers for their willingness to participate and all audience members for their presence. He also thanked all stakeholders who were involved in the two roundtable conferences that took place in the Fall of 2022, which formed the basis of the development of the RADDIAL Memorandum.

Opening remarks

Caroline Taquin, *Member of Federal Parliament, Member of the Committee on Public Health and Equal Opportunities (MR)*

n her opening remarks, MP Taquin expressed her gratitude on behalf of the House's Health Committee to the organisers of this initiative, and argued initiatives like this are an important aid to support policymakers in prioritising policy initiatives. Key issues on which she focused in her remarks were funding issues and lack of information in the field of rare diseases.

Concerning the diagnosis of rare diseases, she emphasised the need for public services to work together more closely and to share data. In this regard, the interdependence of institutions was also highlighted. Finally, she drew attention to the importance of a continued dialogue about rare disease, since they affect up to 500.000 Belgians.

INTRODUCTION: Why should we join forces for a better diagnosis of rare diseases?

Eva Schoeters, *Director, Rare Diseases Belgium (RaDiOrg))*

The central question of Mrs. Schoeters' introduction was why a correct and timely diagnosis is an indispensable condition for optimal care. In this regard, she highlighted that, due to differences in screening policies, diagnosis of rare diseases varies greatly across European countries. Furthermore, she acknowledged that due to the large number of rare diseases, it is not possible for healthcare professionals to be knowledgeable about each individual rare disease. However, she argued, healthcare professionals and other medical stakeholders should receive the appropriate education to allow them to recognise key symptoms and red flags.

1ST PANEL: Screening and early detection

Jean-Marie Huet, *President, Association Belge contre les Maladies neuro-Musculaires (ABMM)*

François Eyskens, *Clinic Head of Paediatrics and Metabolic Disorders, Universitair Ziekenhuis Antwerpen*

n this panel, the speakers underlined the importance of improving communication and collaboration throughout the diagnosis process for rare diseases between all involved parties, including patient representatives, policy makers, multidisciplinary teams and different regional institutions, as well as the importance of keeping up with advancements in technology and testing methods. The need for nationally harmonised screening programs and for the rectification of divergences across regions was also emphasised.

Furthermore, Professor Eyskens discussed the role of policymakers in ensuring proper education on screening and genetic testing, particularly when biomarker testing is not available. He stressed the need for a thorough discussion and understanding of the importance of second-tier genetic testing when first-tier testing on biomarkers does not yield results.

2ND PANEL: Improving knowledge and awareness

Quentin Mary, *President, Société Scientifique de Médecine Générale (SSMG)*

Johan Vansintejan, *Head of Department of Family Medicine and Chronic Care, Vrije Universiteit Brussel*

The central theme of this panel discussion was the importance of improving knowledge and awareness of healthcare professionals in training, in order to support a faster and more efficient diagnosis process. Particularly general practitioners have a central role in this objective, since they know the medical history of the patient's family and follow them up over time. The speakers also stressed the importance of cross-disciplinary collaboration to improve knowledge, as well as the benefits of continuous training on rare diseases and red flags for healthcare professionals.

Next to that, the suggestion was discussed to supply medical teams with a digital toolbox, which could assist them throughout the diagnostic process. In the speakers' view, such a toolbox should be developed with the close involvement of patients. With respect to this toolbox, Professor Vansintejan suggested an integration with existing tools such as Orphanet, in order to make it as comprehensible as possible.

3RD PANEL: An integrated care trajectory

Greet Van Kersschaever, *Adviser on rare diseases, Cabinet of Deputy Prime Minister and Minister of Public Health and Social Affairs Frank Vandenbroucke*

Jean-François Cambier, *Coordinator Rare Disease Function, Grand Hôpital de Charleroi*

In the final panel discussion, the speakers underlined the necessity to enhance integrated care for patients with a rare disease. In this respect, Dr. Cambier drew attention to the important role of rare disease functions, since they aim to support patients with interdisciplinary teams in order to arrive at an accurate diagnosis. In his view, the expertise of these functions should be further developed in order to improve diagnoses and treatments.

Next to that, Mrs. Van Kersschaever emphasized that patients' needs should be closely considered in order to provide integrated care, and that case managers should be assigned to coordinate these needs. To this end, a pilot project will soon be organised in specific hospitals; the project will help identify best practices for integrated care and how they can be implemented nationally. The speakers also referred to the Inter-federal Plan Integrated Care. In their view, this plan offers the opportunity to further expand integrated care with the aim of achieving a broader view of the patient experience.

Q&A WITH AUDIENCE

During the Q&A session, audience members highlighted several key points. They stressed the urgent need for increased cooperation among all healthcare actors and institutions, emphasizing the importance of breaking down barriers between prevention and care. The diverse nature of diseases must also be considered when crafting policy choices. Placing the patient and their needs at the core of healthcare policies was a recurring theme.

Additionally, the call was made for policymakers and healthcare decision-makers to draw inspiration from successful developments in other countries to further advance healthcare policy and practices. Finally, the audience expressed appreciation for the collaborative efforts by multiple healthcare companies that gave rise to the initiative.

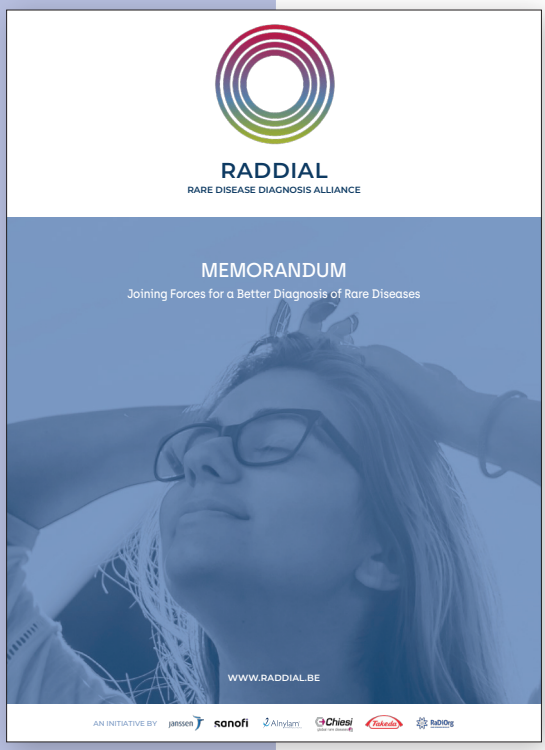
CLOSING REMARKS

Robby De Caluwé, *Member of Federal Parliament, Member of the Committee on Public Health and Equal Opportunities (Open Vld)*

In his closing remarks, MP De Caluwé announced his intention to draft a proposal for a resolution in collaboration with MP Taquin, which would be based on the central elements of the policy recommendations of the RADDIAL Memorandum. He also spoke out in favour of closer collaboration among all involved stakeholders to improve the diagnosis process and access to treatment for patients with a rare disease in Belgium. Next to that, he called on stakeholders to enhance communications among themselves, and to employ the latest advancements in digital technology as much as possible.

ABOUT RADDIAL

RADDIAL (Rare Disease Diagnosis Alliance) is an initiative of five pharmaceutical companies: Takeda, Janssen, Sanofi, Chiesi and Alnylam. The alliance was founded in 2022, with the aim of establishing a broad dialogue between different stakeholders aimed at developing policy recommendations to accelerate the diagnosis pathway for rare disease patients.



RaDiOrg, the Belgian umbrella association for people with rare diseases, is closely involved in the initiative and, as an independent partner, ensures that the patient perspective remains central to the dialogue.

Between September and December 2022, RADDIAL brought patient representatives, academic experts, doctors, healthcare providers and health insurers together for a joint dialogue. This dialogue resulted in a political memorandum with twelve policy recommendations, which was presented at a conference in the House of Representatives on 29 June 2023.

The Memorandum can be downloaded from the RADDIAL website, which you can visit via **raddial.be** or the **QR code** below.

WWW.RADDIAL.BE

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