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Commissioner for Health and Food Safety

Helena Dalli  
Commissioner for Equality

Iliana Ivanova  
Commissioner for Innovation, Research, Culture, Education and Youth

Nicolas Schmit  
Commissioner for Jobs and Social Rights

Brussels, 29 February 2024

**Subject: Inclusion of a European strategy on rare diseases in the European Commission work programme for 2024-2029**

Dear President von der Leyen,

We are writing to you on behalf of **EURORDIS-Rare Diseases Europe and its National Alliances for Rare Diseases from over 20 EU countries.**

On Rare Disease Day 2024, we commend the European Commission's continued commitment to addressing the unique challenges faced by individuals with rare diseases. In light of the progress made and the evolving landscape of research and healthcare, **we respectfully request the inclusion of a comprehensive European Action Plan for Rare Diseases in the Commission's Work Programme for the period 2024-2029.**

We are grateful for the efforts made thus far, as reflected in various communications and initiatives since 2019. Over the past years, we have closely followed the European Commission's endeavours to improve the lives of those affected by rare diseases, and we have noted the significant strides taken. These efforts are crucial in addressing the unmet needs of patients, their families, and caregivers, and we acknowledge the Commission's dedication to fostering a more inclusive and supportive environment for individuals living with rare diseases, including rare cancers.

However, the job is not done: **the 30 million people living with rare diseases** in Europe<sup>1</sup>, their families and the clinicians and health professionals who care for them are still facing huge challenges in the EU today. **The average wait for an accurate diagnosis of a rare disease remains at about 5 years**<sup>2</sup>. **Only 5% of people living with a rare disease have received a transformative treatment approved for the entire EU. 22% of people with rare diseases could not get the treatments they needed because they were not available where they live, reflecting the fragmentation of the market across the Member States**<sup>3</sup>. **52% of people living with a rare disease or their carers say that their condition has a severe impact on everyday life**<sup>4</sup>.

The two-year Foresight Study, Rare 2030, initiated by the European Parliament and funded by the European Commission<sup>5</sup> concluded, on behalf of hundreds of key experts and thousands of people living with a rare disease, that renewed European action on rare diseases is required. **An overarching strategy on rare diseases emerged as the key recommendation of Rare 2030**. Indeed, an overarching strategy is currently needed to bridge national and European legislation, policies and programmes across the Pharmaceutical Package, the European Health Data Space, European Reference Networks, Europe's Beating Cancer Plan, EU4Health, Horizon Europe, the European Pillar on Social Rights, the European Disability Strategy, and beyond.

The adoption of such a European strategy, penned a European Action Plan for Rare Diseases, is now supported by various European bodies. It has gathered the support of **21 Member States** who endorsed a Call to Action under the Czech Presidency of the EU Council<sup>6</sup>, hereby joining the European Parliament<sup>7</sup>, the European Economic and Social Committee<sup>8</sup>, the European Court of Auditors<sup>9</sup> and the rare disease community at large. In December 2021, the General Assembly at the United Nations adopted a Resolution on 'Addressing the Challenges of Persons Living with a Rare Disease'<sup>10</sup> recognising the global impact of rare diseases.

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<sup>1</sup> Nguengang Wakap, S., Lambert, D.M., Olry, A. et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet* 28, 165–173 (2020). <https://doi.org/10.1038/s41431-019-0508-0>

<sup>2</sup> Global survey on diagnosis of Rare Diseases, a [RareBarometer](#) survey, 2023, pending publication of results.

<sup>3</sup> Access to treatment: [Unequal care for European rare disease patients](#), a RareBarometer survey, February 2017

<sup>4</sup> ['Juggling care and daily life: The balancing act of the rare disease community'](#), a RareBarometer survey, May 2017

<sup>5</sup> <https://www.rare2030.eu/>. Over a two-year period, Rare 2030 brought together more than 250 patients, practitioners and key opinion leaders to develop policy recommendations to improve the lives of people living with rare diseases and offer them a better future.

<sup>6</sup> Call to action from the expert conference on rare diseases under the Czech Presidency of the EU Council - Toward a new European policy framework on rare diseases "*Building the future together for rare diseases*", Prague, 25-26 October 2022: <https://slg.cz/documents/90/cz-pres-2022-call-to-action.pdf>

<sup>7</sup> Parliament debate on the European action plan for rare diseases in 2021: [A European Action Plan Against Rare Diseases](#) and [letter of MEPs to the President of the European Commission on Rare Diseases Day 2023](#)

<sup>8</sup> Opinion of the European Economic and Social Committee in 2022: <https://op.europa.eu/webpub/eca/special-reports/cross-border-health-care-7-2019/en/>

<sup>9</sup> European Court of Auditors special report n°7/2019: <https://op.europa.eu/webpub/eca/special-reports/cross-border-health-care-7-2019/en/>

<sup>10</sup> <https://www.rarediseasesinternational.org/un-resolution/> Prior to this Resolution, in 2019, UN Member States adopted the landmark Political Declaration on Universal Health, which includes a strengthened commitment to fight rare diseases.

**This Action Plan should build upon the foundation laid in these previous initiatives and take into account the latest advancements and insights in the rare disease field.** We believe that a dedicated Action Plan will further strengthen the European Union's commitment to addressing the challenges faced by the rare disease community. It should encompass a holistic approach, covering areas such as:

1. **Research and Innovation:** promote research initiatives focusing on rare diseases, fostering collaboration between stakeholders and leveraging the latest technologies.
2. **Access to Diagnosis and Treatment:** ensure timely and equitable access to accurate diagnoses, innovative therapies, and essential healthcare services.
3. **Patient Empowerment:** facilitate patients' access to information, education, and involvement in decision-making processes concerning their healthcare.
4. **Cross-Border Healthcare:** facilitate cross-border cooperation to improve access to expertise, share best practices, and ensure the harmonisation of healthcare services.
5. **Data Sharing and Collaboration:** encourage the development of platforms for data sharing, collaboration, and coordination among Member States to improve care, research and innovation.
6. **Holistic care and Inclusive Policies:** promote inclusivity policies, recognising the unique challenges faced by individuals with rare diseases in various aspects of life.

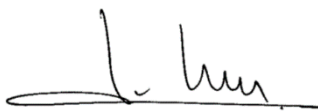
By incorporating these elements into the future Commission's Work Programme, we believe the European Union can continue to lead the way in addressing the needs of the rare disease community and contribute to a more just and equitable healthcare system. A European strategy for rare diseases would continue to demonstrate it, at a time where national governments' individualism is taking precedence over the possibility of a stronger European Union. The rare disease community knows that the EU can meet challenges that Member States cannot solve alone. You will always find the rare disease community by your side, valuing every day the significant role of the European Union and understanding better than anyone the importance of collaboration and cooperation.

Fifteen years after the last comprehensive EU strategy on rare diseases, it is time to act and propel rare diseases as an engine for a European Health Union. **This Rare Disease Day, we stand with the rare disease community and respectfully urge you to take forward the recommendation of a European Action Plan for Rare Diseases to ensure that no one is left behind.**

Yours sincerely,



Avril Daly  
President  
EURORDIS-RareDiseases Europe



Yann Le Cam  
Chief Executive Officer  
EURORDIS-RareDiseases Europe

Signatories	National Alliances	Logos
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