



COLLABORATE

Working together for adult rare diseases

2025

INITIATIVE 1:

Elevating the rights of patients with
adult-onset rare diseases



INITIATIVE 1: Background context

Initiative 1: Elevating the rights of patients with adult-onset rare diseases

LEAD: Mariano Votta



RATIONALE:

- Around 30% of people with rare diseases do not develop symptoms until adulthood.
- Yet, their voices are often missing from the conversation about rare diseases.
- The first-of-its-kind multistakeholder COLLABORATE initiative aimed to understand the challenges faced by people with adult-onset rare diseases and identify opportunities for change.



CHALLENGES & OPPORTUNITIES:

- 24 May 2025, the Resolution on Rare Diseases has officially been adopted at the 78th World Health Assembly (WHA78). This is a milestone moment for the global rare disease community. The adoption of this Resolution by WHO Member States marks a powerful step forward in ensuring equity, visibility, and inclusion for the 300 million people living with a rare disease around the world. What attention is reserved for the condition of patients with adult-onset rare diseases?
- Yesterday (12 Nov 2025), European Parliament Plenary debated on the new 2028-2034 MFF: architecture & governance. MEPs and Commission and Council representatives are discussing the architecture and governance of the EU's 2028-2034 long-term budget (multiannual financial framework - MFF). What about health-related issues? What about for people living with rare diseases (between 27 and 36 million EU people live with a rare disease)



INITIATIVE 1: What is the idea?

Initiative 1: Elevating the rights of patients with adult-onset rare diseases

LEAD: Mariano Votta



OVERVIEW: An institutional initiative to secure cross-party political champions in the European Parliament to push age-equitable access to diagnosis (including genetic testing), transition of care, and treatment in EU and national policies



OBJECTIVES:

1. To **leverage** at the EU level perspectives/requests/recommendations of the COLLABORATE constituency in the framework of the EU4HEALTH 2021-2027 priority
2. To officially **present** for the first time to the EU Institutions the COLLABORATE expert consensus paper “Challenges & Opportunities in Adult-Onset Rare Diseases: Perspectives from a Global Multi-Stakeholder Expert Group”. Read more
3. To **promote/open** a dialogue with EU Institutions focused on the implementation of policy solutions that support:
 - The needs of older adults with rare diseases must be recognized in national rare disease frameworks and policies globally, receiving equal consideration and attention to childhood rare diseases
 - Advocating for integrated, coordinated care essential for all people with a rare disease across the life course



INITIATIVE 1: From the idea to the implementation – Structure

Initiative 1: Elevating the rights of patients with adult-onset rare diseases

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- Institutional venue: European Parliament in Bruxelles (Belgium)
- Policy dialogue in a multi stakeholder format
- Event of 1 ½ / 2 hours with a journalist as a moderator
- Representatives of COLLABORATE in the panel together with independent experts & representative of the EU Institutions
- Q & A session & editorial team (to capture the main suggestions)
- Speaking language: English without translation
- Link to join remotely connected (tbc)
- Supported by (tbc)



INITIATIVE 1: From the idea to the implementation – Dissemination

Initiative 1: Elevating the rights of patients with adult-onset rare diseases

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Dissemination activity:

- Media partnership
- Articles in specialized media
- Publication of the Policy Recommendations in a peer-reviewed international journal
- video-interviews to the speakers
- Communication materials (save the date / roll-up, press release / programme, etc.)
- Social media activity
- Photo Gallery



INITIATIVE 1: From the idea to the implementation – Exploitation

Initiative 1: Elevating the rights of adults with adult-onset rare diseases

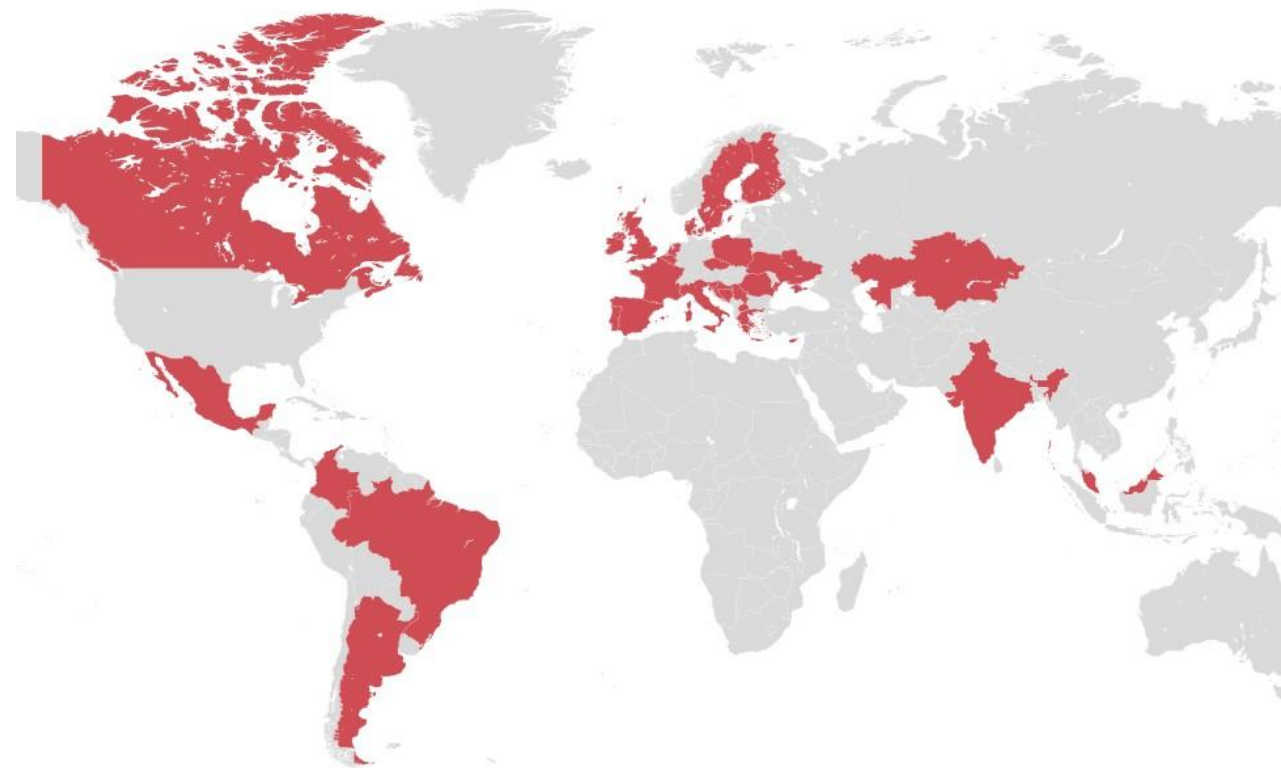
LEAD: Mariano Votta



Exploitation activity:

- Same format (official dialogue with the policy-makers, Institutional venue, multi stakeholder initiative, etc.) & template communication
- Encourage «Flagship initiatives» not only at the EU level but across the world, tailored with the specific context
- Sharing the same documents and key messages to different constituencies, stakeholder and policy-makers
- Amplify the commitment guaranteeing the visibility of the logos of all the COLLABORATE members
- From “pilot” to “standardized” initiative

Figure 1. The 32 countries represented by the COLLABORATE Community



INITIATIVE 1: Where are we now?

Initiative 1: Elevating the rights of patients with adult-onset rare diseases

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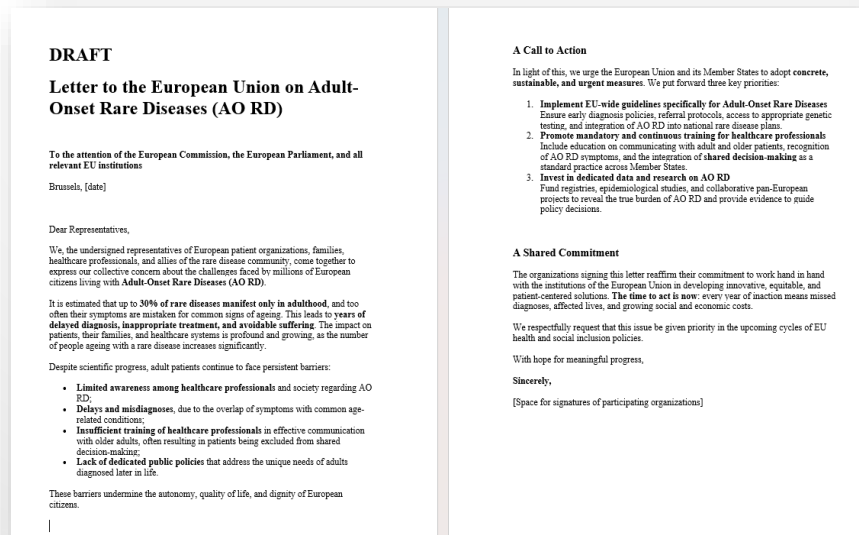


COLLABORATE WORKING GROUP ACHIEVEMENTS:

- Agreement on joint statement and drafted an official letter to the European Institutions
- Hypothesis of title: *“Addressing the unmet needs of Adult-Onset Rare Diseases Patients in Europe”*
- Discussion on the key policy asks: (1) Access to genetic testing, (2) Equal treatment access, (3) Recognition in EU/national plans

To be sent to the following high level EU key-target:

- The cabinet of Olivér Várhelyi, Commissioner Health and Animal Welfare (2024-2029).
- The cabinet of the President of the EU Parliament Roberta Metsola + all the MEPs of SANT, the new permanent Committee on Public Health at the EU Parliament, established on Dec. 18, 2024 and officially took effect on January 1, 2025.
- The presidency of the Council of the EU (Denmark: July-December 2025 + Cyprus: January-June 2026).



INITIATIVE 1: Pending issues / practicalities to be done

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OVERALL ORGANIZATION AND MANAGEMENT OF THE EVENT

- Liaison with EU Institutions
- Selection of the hosting MEP
- Agreement with moderator/videomaker, etc.
- Template communication & production of documents to be shared
- Invitation/registration for attending the event
- Defining the final agenda
- Briefing with panelist
- Stakeholder engagement & outreach
- Etc.



COLLABORATE
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for adult rare diseases

**CHALLENGES & OPPORTUNITIES
IN ADULT-ONSET RARE DISEASES:
PERSPECTIVES FROM
A GLOBAL MULTI-STAKEHOLDER
EXPERT GROUP**

ENSURING INTEGRATED, COORDINATED CARE FOR ALL PEOPLE LIVING WITH A RARE DISEASE ACROSS THEIR ENTIRE LIFE COURSE

2024

What is care coordination? Coordinated care is the deliberate organization of patient care activities and sharing of information across all stakeholders involved to achieve safer and more effective care.

INTRODUCTION

People are diagnosed and live with rare disease across their entire life course. Rare diseases are complex, requiring coordination of care between multiple healthcare professionals and specialties. As they age, people with rare diseases who require a coordinated approach for the duration of their care from pediatrics to adult care, and then into older age. Despite this, most people living with rare diseases experience significant care coordination gaps, and the ability to live better for the patient or their caregiver to coordinate their own care, which often results in compromised health.

RESEARCH

- Increasing numbers of people's rare disease patients are transitioning into adulthood ("many report issues with care during this transition")
- Caregivers of people with rare diseases are usually "partners or spouses" (they care most of the physical and emotional burden of care, usually receive no financial compensation for their role)
- In Europe, 60% of rare disease patients and caregivers report having to visit multiple different services to obtain services of their own and their loved ones hard to manage
- Around two thirds of people with rare diseases in Europe believe healthcare providers communicate disease related information poorly (Fig. 2) which causes burden for patients and caregivers
- An people with rare diseases progress through the life course, the complexity of integrated care and the financial and emotional burden on caregivers can increase

RECOMMENDATIONS

INTEGRATED, COORDINATED CARE IS ESSENTIAL FOR ALL PEOPLE LIVING WITH A RARE DISEASE ACROSS THE ENTIRE LIFE COURSE

We call on the rare disease community to:

- Educate & enhance understanding
- Leverage technology
- Connect services & stakeholders

EDUCATE & ENHANCE UNDERSTANDING

- Improve understanding of the importance of coordinated rare disease care across all HCPs, including allied health professionals
- Educate and provide resources of pediatric to adult care transition and leverage key findings across rare diseases

LEVERAGE TECHNOLOGY

- Develop digital tools that support health systems and healthcare professionals to deliver coordinated rare disease care (patients, caregivers, patient research, and care team) to ensure the approach is always evidence-based, giving patients health services to include them

CONNECT SERVICES & STAKEHOLDERS

- Integrate holistic care services into standard hospital care, and enhance awareness of, and funding for, this comprehensive care approach
- Connect caregivers with healthcare professionals to support coordination of rare disease care where the care coordinator role is not established

KEY HIGHLIGHTS

- An increasing number of young people with rare diseases are starting into adulthood, often requiring increased complexity of care
- Rare diseases are a growing burden requiring coordination of care from multiple different healthcare providers and services
- A lack of coordinated care through the life course transition into adulthood adds to the patient and caregiver burden
- Usually, rare disease patients and their caregivers manage the coordination of care to be more continuous and efficient by managing
- Better coordination of care is needed during the transition of rare disease patients from childhood to adulthood and their caregivers

Figure 1: Patient perceived quality of rare disease related communication from healthcare providers



Figure 2: Patient perceived quality of rare disease related communication from healthcare providers



ENSURING THE EQUAL CONSIDERATION OF AND ATTENTION TO OLDER ADULTS WITH RARE DISEASES IN POLICIES WORLDWIDE

2024

INTRODUCTION

Despite a substantial proportion of people with rare diseases first experience symptoms in adulthood, public and healthcare professional (HCP) awareness and understanding of adult-onset rare diseases remains limited, often resulting in suboptimal disease management. Furthermore, the healthcare experiences of many older rare disease patients is negatively impacted by poor HCP communication and a lack of relevant information for those whose rare disease emerges in adulthood. Adult-onset rare diseases are under-represented in rare disease plans and policies, hindering crucial research and funding support for these rare disease patients, and the value of the older rare disease patients often goes unmet.

RESEARCH

- Around one third of rare diseases onset in adulthood (Fig. 1) (rare pediatric, rare adult, and 2/3 exclusively adult)
- Fig. 1: Includes 218 diseases, Adult-onset 50%, Pediatric 50%
- The number of people living with a rare disease is increasing globally - in 2018, a three-fold increase in rare disease patients transitioning from childhood to adulthood was recently reported
- Life expectancy for certain rare diseases has significantly increased in recent decades and the mean age of rare disease patients in the US and Europe is now 50-60 years
- Experts report that at least 20 common signs of aging are also symptoms of adult-onset rare diseases and patients have reported being confused between the two, as well as being dismissed by HCPs who attribute their symptoms to aging "This slows time to diagnosis"
- Genetic testing is a valuable tool underexplored for supporting the timely diagnosis of certain adult-onset rare diseases, with HCPs being far less likely to offer genetic testing to patients over the age of 50
- Older people with rare diseases report experiencing greater care coordination, and healthcare provision of relevant information from their healthcare professional, which can help to regularly impact treatment outcomes

RECOMMENDATIONS

THE NEEDS OF OLDER ADULTS WITH RARE DISEASES MUST BE RECOGNIZED IN NATIONAL RARE DISEASE FRAMEWORKS AND POLICIES, GLOBALLY, RECEIVING EQUAL CONSIDERATION AND ATTENTION TO CHILDHOOD RARE DISEASES

We call on the rare disease community to:

- Educate & enhance understanding
- Improve programs & policies

EDUCATE & ENHANCE UNDERSTANDING

- Accurate laws to diagnosis of adult-onset rare diseases by increasing understanding of the symptoms and how they overlap with common signs of aging (e.g. for adult-onset osteoporosis, cognitive decline)
- Ensure the inclusion of adult-onset rare diseases in medical school curriculum and professional training for primary care physicians, as well as education on communicating with older adults
- Advocate for increased awareness of adult-onset rare diseases and the specific challenges faced by people living with them

IMPROVE PROGRAMS & POLICIES

- Include adult-onset rare diseases with a genetic component in national genetic testing programs and ensure older people are not excluded from genetic testing and counseling
- Ensure the professional impact of adult-onset rare diseases on both patients and caregivers is recognized in policies and support the development of collaborative disease management strategies
- Provide adequate and equal funding for research into adult-onset rare diseases, as well as the impact of aging on rare diseases

KEY HIGHLIGHTS

- Adult-onset rare diseases are often reported later and underfunded, and do not receive equal consideration in national rare disease plans globally, resulting in low awareness
- The symptoms of certain adult-onset rare diseases can be confused with common signs of aging by both HCPs and patients, further delaying diagnosis or resulting in misdiagnosis
- While genetic testing can be used to support timely diagnosis, it is not routinely performed for older patients
- Older rare disease patients report experiencing greater care coordination and healthcare provision
- There is a lack of patient information on rare diseases tailored to the needs of older adults

Figure 1: Rare disease onset



INITIATIVE 1: Hypothesis of Hosting MEP & EU political champions

Initiative 1: Elevating the rights of patients with adult-onset rare diseases

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