Possible association to Horizon Europe: request for information

Rare Voices Australia Submission — October 2025

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Executive

Rare Voices Australia (RVA), the national peak body for Australians living with a rare disease, welcomes the opportunity to provide input into the consultation on Australia's potential association with Horizon Europe. This formal international research partnership represents a transformative opportunity to strengthen Australia's global health and medical research connections.

RVA strongly supports Australia becoming an Associate Member of Horizon Europe, recognising that progress in rare disease research depends on global collaboration. The inherently small and dispersed nature of rare disease populations means meaningful advances are often reliant on coordinated international efforts. Association with Horizon Europe would enable Australian rare disease researchers to join multinational consortia, access shared infrastructure and funding, and exchange knowledge at the pace of global discovery.

Importantly, association would strengthen and facilitate formal connections with leading European rare disease initiatives such as the European Reference Networks (ERNs) and Orphanet, both of which are critical enablers of high-quality care and research for rare diseases.

Association with Horizon Europe directly aligns with the Australian Government's National Strategic Action Plan for Rare Diseases[1] (the Action Plan), which calls for stronger international partnerships to drive research and innovation for all Australians living with a rare disease. Learning from, and linking with, established European models such as the ERNs would support the development and implementation of networked rare diseases centres of expertise (CoE) across Australia—one of RVA's key strategic priorities. Rare disease CoE are an internationally recognised model for best practice in rare disease care. This model improves access to specialist rare disease expertise, enabling timely diagnosis and treatment, more effective data collection, and embedding research and innovation into routine clinical care.

RVA encourages the Australian Government to leverage this opportunity, not only to strengthen international research collaboration, but also to embed rare disease priorities within Australia's engagement with Horizon Europe, thereby accelerating innovation and improving outcomes for the estimated two million Australians living with a rare disease.

About Rare Voices Australia

RVA is the national peak body for Australians living with a rare disease, providing a unified voice to advocate for policy, health, disability, and social systems that work for people living with rare diseases. Our person-centred approach brings together key stakeholders across government, research, healthcare, and industry to drive the best possible outcomes for Australians living with a rare disease.

In 2018, the Australian Government commissioned RVA to lead the collaborative development of the National Strategic Action Plan for Rare Diseases (the Action Plan).[1] The Action Plan is the first nationally coordinated effort to address rare diseases in Australia and was informed by an extensive multi-stakeholder consultation process led by RVA. The Minister for Health launched the Action Plan in February 2020, with bipartisan support. RVA continues to work with State and Federal Governments, and all other rare disease stakeholders, in leading collaborative implementation of the Action Plan.

About Rare Diseases

A disease is rare if it affects fewer than five in 10,000 people. There are approximately 7,000 different diseases and together they affect an estimated two million Australians. [1]

Approximately 80% of rare diseases have genetic causes,[1] and due to the hereditary nature of some rare diseases, multiple people within the same family can be impacted. Rare diseases are often serious and progressive, exhibiting a high degree of symptom complexity, leading to significant disability, health and psycho-social challenges.

Rare diseases have profound personal and health system impacts and are now internationally recognised as a global health emergency by the United Nations and the World Health Organization. Notably, the World Economic Forum recently moved to prioritise rare diseases as one of three disease domains critical for healthcare sustainability in the Asia Pacific region. This was driven by two key factors (1) profound health inequities; and (2) recognising rare diseases as a critical window into understanding health for both common and rare conditions.

Rare diseases are the leading cause of death in children, responsible for six in ten childhood deaths[2], and associated with a hospital mortality rate 13 times higher than that of common conditions[3]. They are also a major driver of healthcare costs, contributing more than $100 billion each year in direct and indirect mortality costs[4,5,6] — and this reflects only a portion of the more than 7,000 known rare diseases.

These sobering figures underscore an urgent need for action. For many people living with progressive rare diseases, time is critical, and access to treatment is often only possible through participation in a clinical trial. Addressing this unmet need demands global collaboration in research and innovation

Rare Voices Australia and Research

In line with the Action Plan's Research and Data Pillar, RVA supports the need for high-quality collaborative research that positively impacts the lives of Australians living with a rare disease.

RVA works closely with Australian rare disease researchers to strengthen understanding of the opportunities and challenges they face, ensuring these insights inform RVA's systemic advocacy and policy leadership. We engage with rare disease researchers through multiple avenues, including via our RVA Research Partnerships Program, the RVA Scientific and Medical Advisory Committee (SMAC), and, more recently, through the Australian Rare Disease Research Network.

RVA also plays a key role in research advocacy, maintaining strong relationships with the Health and Medical Research Office (HMRO) and contributing to national research policies and initiatives. This includes involvement in co-design of the National One Stop Shop for Clinical Trials and Human Research, and participation in the development of the National Health and Medical Research Strategy, where RVA is represented on the Technical Reference Group.

Rare Voices Australia's Position on Australia's Potential Association with Horizon Europe

In rare disease, inherently small patient numbers make global collaboration vital. International partnerships enable timely access to knowledge, treatments and research opportunities that may otherwise be unavailable in Australia.

RVA, together with RVA SMAC, strongly supports Australia becoming an Associate Member of Horizon Europe. Australia's associate membership would generate opportunities for funding and shared projects led by Australian rare disease researchers. Association would unlock access to significant international funding, foster research partnerships and consortia and strengthen global knowledge exchange. In a space when knowledge is limited and approved treatments are few, international partnerships with multinational well-funded consortia like Horizon Europe are not only beneficial but essential.

Formal association with Horizon Europe would also enable timely and effective horizon scanning for new and novel health technologies—an activity that aligns closely with several existing Australian policy frameworks and strategies, including Genomics Australia and the forthcoming National Health and Medical Research Strategy. Horizon scanning is particularly critical in rare disease given fewer than 5% of rare diseases currently have an approved treatment[7], yet the global pace of innovation, especially in advanced therapies, is accelerating.

RVA encourages a strategic and considered approach to association with Horizon Europe that ensures Australian participation advances the priorities of Australian stakeholders and delivers meaningful benefits for all Australians, including those living with a rare disease.

Alignment with the National Strategic Action Plan for Rare Diseases

One of the three main pillars of the Action Plan is research and data. For many rare diseases, there are several barriers to effective research and no active research programs. One of the biggest challenges is that rare diseases have small patient numbers and are often very complex. Depending on the specific rare disease, research priorities can be different. For example, while funding for translational research may be important for many rare diseases, some rare diseases are not yet in the position to prioritise translational research. For some rare diseases, the unmet research needs are basic discovery research or investment into data collection and natural history studies.[1]

Strong state, national and international partnerships is one of four critical enablers of the Action Plan.[1] The Action Plan further highlights the need for improved policy settings and collaborations across borders to drive research and innovation, and improve diagnosis, treatment and care for all rare diseases.[1]

Overall, this Australian government policy position on rare diseases, and the mechanisms required for implementation of its key priorities, support the decision for Australia to become a formal associate member of Horizon Europe.

Opportunities for Australians Living with Rare Diseases

Formalising connections with the European Reference Networks

A strategic priority for RVA and the broader rare disease community is the development and implementation of networked rare disease Centres of Expertise (CoE), which connect existing specialist services and expertise across jurisdictions. There is mounting evidence and acknowledgment across the rare disease sector, both in Australia and overseas, for CoE as a solution to the multifaceted challenges faced by people living with rare diseases. The European Reference Networks (ERNs) provide a proven framework for this model.

The ERNs have achieved global recognition in facilitating the delivery of expert care, despite geographical boundaries and distances. The ERNs' objective is to tackle complex or rare diseases and conditions that require highly specialised treatment and a concentration of knowledge and resources. By facilitating cross-border consultation and virtual collaboration, ERNs ensure individuals can access the expertise they need regardless of where they live.

In 2013, Rare Disease United Kingdom's (RDUK) Centres for Excellence for Rare Diseases report, identified several key characteristics to define Centres of Excellence, one of which included: ''Membership of international networks of excellence – work collaboratively to facilitate best practice.'' The UK are also part of the European Reference Networks (ERNs).

Partnering with Horizon Europe would provide Australia with a valuable opportunity to formally learn from and engage with the ERN model. This collaboration could support the adaptation of a comparable network within Australia—one designed to overcome the challenges of distance, workforce limitations, and the uneven distribution of expertise across our vast geography—helping to eliminate barriers to timely, high-quality care for all Australians living with a rare disease.

Strengthening and Formalising Existing Relationships with Europe

Historically, RVA and members of RVA SMAC have engaged with both the National Health and Medical Research Council (NHMRC) and the Medical Research Future Fund (MRFF) to explore stronger alignment with Europe. These efforts reflect Europe's consistent interest in closer research ties with Australia.

In 2020, RVA together with RVA SMAC facilitated Australian collaboration between the European Joint Program for Rare Diseases (EJ PRD) and the Australian Government's Health and Medical Research Office. These discussions led to the 2022 MRFF Joint Transnational

Call Grant Opportunity. The EJP RD brings together the research efforts of over 130 institutions from 35 countries in the field of rare diseases. These international organisations joined forces to carry out a joint call for proposals for transnational cooperative research projects in the field of rare diseases, with one successful Australian recipient.

Australian researchers have successfully participated in other EU-funded rare disease projects—including RD-Connect, Neuromics, and Rare Best Practices—demonstrating our capability and the mutual benefit of collaboration. Formal association with Horizon Europe would remove current barriers to participation and leadership in such programs and increase access to research funding in Australia.

More recently, Australia joined the European Rare Diseases Research Alliance (ERDERA), and RVA, together with members of SMAC, is supporting the formation of the Australian National Mirror Group for ERDERA, further underscoring Australia's growing integration into the global rare disease research community.

Australia's association with Horizon Europe would significantly strengthen national rare disease research capacity by expanding access to international funding, enabling formal participation in multinational research consortia, and opening pathways to shared data infrastructure, and leading initiatives such as Orphanet and other pan-European programs.

Concluding Remarks

RVA strongly encourages the Australian Government to pursue a thoughtful association with Horizon Europe and to ensure that rare disease research is explicitly recognised and prioritised within Australia's participation strategy. This will not only advance the objectives of the Action Plan but also ensure Australians living with rare diseases can benefit from, and contribute to global innovation in diagnosis, treatment and care.

Small and dispersed rare disease patient populations demand international research collaboration, and the sharing of knowledge and expertise, to ensure timely and equitable access to the best possible treatments and care. A stronger relationship with Horizon Europe would give Australian rare disease researchers and clinicians access to world-leading networks, infrastructure, and funding, accelerating research discovery and translation for tangible outcomes for Australians living with rare diseases.

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