

General Information	
Preliminary title of the European Partnerships	European Partnership on Rare Diseases
Short description of the partnership	Improving the lives of rare diseases patients
Services directly involved	DG RTD, SANTE, JRC and CNECT
Context and problem definition	<p>Almost 40 million EU citizens are affected by one of the 6000 to 8000 life threatening or chronically debilitating rare diseases. More than 90% of rare diseases lack effective treatment options; the average time for diagnosing a known disease is today more than 5 years; it is estimated that only about half of the diseases are known, which makes diagnosis impossible and the development of a treatment or cure even further away.</p> <p>Major investments have been made in research and innovation on rare diseases from the previous EU Framework Programmes. The EU activities have leveraged and brought together scattered resources and expertise on rare diseases to attain a critical mass required for tackling ambitious research objectives and provision of high quality expert care. However, despite the advances in biomedical research and in the provision of care there are still huge unmet medical needs related to rare diseases.</p> <p>The European Joint Programme on Rare Diseases (2019-2023) is a prime example for working together with Member States and other stakeholders on a more integrative and cross-sectorial approach to tackle health challenges.</p>
Objectives and expected impacts	<p><u>Objectives:</u></p> <ul style="list-style-type: none"> - Contribute towards the objectives of the International Rare Diseases Research Consortium IRDIRC to shorten the average time to correct diagnosis to 1 year, have 1000 new therapies for rare diseases and methodologies to assess the impact on patients by 2027; - Further develop the ecosystem for discovery research and development of new diagnostic tools and therapies for rare diseases providing an efficient and effective "pipeline" from research to healthcare to ensure that research and innovation results are reaching the patients as quickly as possible and that healthcare needs can better feed into research prioritisation; - Capacity to collect and share all relevant rare disease data at EU and international level efficiently utilizing data collected by the European Reference Networks; - Research collaboration in the EU, the Associated and Third Countries enabling unprecedented pooling of resources and expertise; - Provide evidence for fit-for purpose regulatory framework taking account of latest science and establishment of EU/international standards largely facilitating research and innovation; - Reinforce EU as an effective "hub" for rare disease research and innovation. <p><u>Expected impacts:</u></p> <p>Societal:</p> <ul style="list-style-type: none"> - Improve lives of rare diseases patients by providing new and optimised prevention strategies, diagnostic tools and treatment options for these diseases; - Ensure that rare diseases patients have timely access to affordable, preventive and curative health care of good quality in line with the EU Pillar of Social Rights; - Support the EU's commitment to the UN's Sustainable Development Goals calling for universal health coverage for all at all ages by 2030, leaving no one behind, and ending preventable deaths.

	<p>Economic:</p> <ul style="list-style-type: none"> - Improving health outcomes of rare diseases patients resulting in increased life expectancy, capacity to work and sustainability of health and care systems; - Building the ecosystem for rare diseases research and innovation making the EU an attractive place for industry to invest; - Creating highly skilled health and care workforce capable of using novel innovations in order to reach efficiency gains in healthcare. <p>Scientific:</p> <ul style="list-style-type: none"> - Harness the combined skills of different stakeholders including researchers, clinicians and patients to improve the scientific knowledge base on rare diseases and develop new solutions for better prevention, treatment and cure of rare diseases; - Exploiting the opportunities provided by new technologies and digitalisation of healthcare to accelerate research and innovation and improve the uptake in healthcare system. <p>Contribution to EU policies:</p> <ul style="list-style-type: none"> - Directive on the application of patients' rights in cross-border healthcare (2011/24/EU); - Commission Communication on Rare Diseases (COM(2008) 679) and the Council Recommendation on an action in the field of rare diseases (2009/C151/02); - Commission Communication on "Enabling the digital transformation of health and care in the Digital Single Market; empowering citizens and building a healthier society" (COM(2018) 233).
Necessity test: rationale for a European Partnership	<p>Even though there are almost 40 million EU citizens affected by one of the 6000 to 8000 life threatening or chronically debilitating rare diseases, there are just a very few cases for an individual rare disease in each Member State. Therefore, collaboration at the EU and international level is essential for being able to progress the development of diagnostic means, treatments and cures. Moreover, reaching meaningful impact requires a portfolio approach encompassing a broad range of activities including research funding, data sharing, training, piloting, demonstration and support for innovation. It is also crucial to facilitate access for patients and general practitioners to highly specialized health care providers (European Reference Networks) which in turn need to be linked effectively with researchers and research infrastructures across Europe. This requires coordination and concentration of efforts towards strategic priority areas at the EU and MS level.</p> <p>Through traditional Framework Programme calls for proposals, it would not be possible to achieve the level of coordination, collaboration and sharing of resources, expertise and disease cases (individual patients) needed to effectively tackle the challenges and deliver on the objectives. Maximising the impact on the patients' health and/or health systems level requires building a long-term strategy and annual programming for a wide range of activities including research, innovation, networking, training, demonstration and dissemination, which is not possible through traditional collaborative projects.</p>
Relevant for the following parts of Horizon Europe	<p>Pillar II 'Global Challenges and European Industrial Competitiveness'</p> <ul style="list-style-type: none"> <input checked="" type="checkbox"/> Cluster Health <input type="checkbox"/> Cluster Culture, creativity and inclusive society <input type="checkbox"/> Cluster Civil Security for Society <input type="checkbox"/> Cluster Digital, Industry and Space <input type="checkbox"/> Cluster Climate, Energy and Mobility <input type="checkbox"/> Cluster Food, Bioeconomy Natural Resources, Agriculture and

	<p>Environment</p> <p><input type="checkbox"/> Cross-cluster</p> <p><input type="checkbox"/> Pillar III ‘Innovative Europe’</p>
Currently identified links with other partnership candidates / Union programmes	<p>Explore opportunities for building links with the other partnerships and EU programmes:</p> <ul style="list-style-type: none"> ▪ Future partnership “Innovative Health Initiative” to accelerate the development and uptake of health care innovations; ▪ Future partnership “Personalised medicine” as rare diseases can be seen as a forerunner in the field, to ensure synergy and complementarity. ▪ Digital Europe Programme for deploying common digital solutions and reinforcing the digital infrastructure and skills; ▪ Connecting Europe Facility for supporting the connectivity between hospitals, medical centres and research centres; ▪ InvestEU to attract private investment in research, innovation and digitisation in the area; ▪ European Regional Development Fund for investments in research and innovation, human capital and innovative technologies and new care delivery models; ▪ European Social Fund + for investing in people in terms of education and training and improving accessibility of healthcare systems.
Does the proposed partnership build on currently active ones?	<p>E-Rare-3 ERANET until end of November 2019.</p> <p>European Joint Programme on Rare Diseases EJP RD until end of 2023.</p>
Expected type and composition of partners	<ul style="list-style-type: none"> ▪ National and regional research funders in the EU, Associated and Third Countries: long-term funding collaboration in ERA-NETs since 2006 is expected to be expanded from the current partnership in the European Joint Programme on Rare Diseases involving more than 130 beneficiaries and third parties from 35 countries. In particular, several Third Countries members of the IRDIRC are interested in funding collaboration; ▪ Universities, research organisations and pan-European research infrastructures possessing the critical skills and providing specialised services for rare diseases community; ▪ European Reference Networks as end-users of the innovative solutions in healthcare and carrying out clinical research; ▪ Patient organisations as active partners in research and innovation; ▪ Private foundations and philanthropy as important funding sources for research in the area of rare diseases; ▪ Possibilities to co-sponsor projects with pharmaceutical, diagnostic and ICT industry to be explored.
Contributions and commitments expected from partners	<p>Financial contribution from research funders required for Joint Transnational Calls</p> <p>In-kind contribution in terms of person months, use of research infrastructures, services, databases, registries etc. from other partners.</p>
Currently envisaged implementation mode(s).	<p><input type="checkbox"/> Co-programmed European Partnership</p> <p><input checked="" type="checkbox"/> Co-funded European Partnership</p> <p><input type="checkbox"/> Institutionalised European Partnership</p> <p style="margin-left: 40px;"><input type="checkbox"/> Article 185</p> <p style="margin-left: 40px;"><input type="checkbox"/> Article 187</p> <p style="margin-left: 40px;"><input type="checkbox"/> EIT-KIC</p>
Justification of the implementation mode	<p>A co-programmed European Partnership (option 1) would allow partners to align their research agendas. However, it would not deliver the close strategic collaboration and integration of activities that is needed to tackle</p>

	<p>the common challenges in the area of rare diseases.</p> <p>A co-funded European Partnership (option 2) with an annual programming provides the flexibility for funding a broad range of activities enabling a pipeline approach supporting translation of research results into clinical application and uptake in healthcare. This option is more appropriate as it will facilitate activities covering research, data sharing and knowledge translation, capacity building and technical support. Open calls for proposals address the large community of rare disease researchers. Support for demonstration activities and pilots involving ERNs ensure that end-users can test and validate research results, which is crucial to deploy and scale-up validated approaches in the clinical practice. Training activities enhance the spread of this knowledge to larger stakeholder groups. Regarding the governance model, lessons are to be learned from the current EJP RD to come up with an optimal model, which would ensure further integration and concentration of efforts across the EU and beyond.</p> <p>An institutionalized European Partnership (option 3) would not be fit for purpose, as more demanding, rigid and complex in terms of governance and management.</p>
Proposed starting year	2024