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European Rare Diseases Partnership

Strategic Research & Innovation Agenda

Dissemination level: public consultation

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72 Vision of the European Rare Diseases Partnership

73 1.1. Missions

74 The European Rare Diseases (RD) Partnership will be organised around the following ambition, vision,
75 and mission.

76 **AMBITION:**

77 The European RD Partnership has the ambition to improve the health and well-being of the 30 million people
78 living with a rare disease in Europe, by making Europe a world leader in RD research and innovation, to
79 support concrete health benefits to rare disease patients, through better prevention, diagnosis and
80 treatment. It will support the EU commitment to UN 2030 Agenda's Sustainable Development Goals: (i) Good
81 health & wellbeing (SDG3), (ii) industries, innovation and infrastructure (SDG9), and (iii) Reduced inequalities
82 (SDG10) as well as the EU political priorities (a Europe fit for the digital age, an economy that works for people,
83 a stronger Europe in the world, Promoting our European way of life and democracy).

84 **VISION:**

85 To leave no one behind, the European RD Partnership will deliver a RD multi-stakeholder ecosystem
86 by supporting robust patient need-led research, developing new treatments and diagnostic
87 pathways, by using the power of health and research data and spearheading the digital
88 transformational change in RD research and innovation (R&I).

89 Finally, the Partnership will structure the European Research Area on RD by supporting the
90 coordination and alignment of national and regional research strategies, including the
91 establishment of public-private collaborations, through research activities all along the R&I value
92 chain, ensuring that the journey from knowledge to patient impact is expedited, thereby optimizing
93 EU innovation potential in RD.

94 This vision will be enabled by a tripartite mission to be accomplished by the end of the Partnership.

95 **MISSION:**

- 96 • Bring and share supporting R&I knowledge, resources and services from across Europe under
97 one roof so that every RD research project would benefit from cross-disciplinary expertise, goal-
98 oriented study planning and efficient execution.
- 99 • Enable every consenting patient living with a rare disease to be findable and enrolled in a
100 suitable clinical study, by boosting generation and sharing of FAIR-compliant, regulatory-quality
101 data from diversity of sources, with the ultimate goal to fasten advances in prevention,
102 diagnosis, disease knowledge and treatment.
- 103 • Make Europe a global leader on rare disease research through a significant increase in
104 investment to spur innovation, by aligning the regional, national and European research and
105 innovation priorities, leading to job creation and improving EU competitiveness in R&I.

106 1.2. Building on Lessons learned

107 The European RD Partnership stems from joint actions between the EU Members States, Associated
108 countries, European Commission and other relevant stakeholders. It builds on lessons learned from
109 the European Joint Programme on Rare Diseases, EJP RD, a major milestone that was achieved in
110 Europe to structure the RD research landscape. EJP RD was launched in 2019, as a prime example
111 of Member States and other stakeholders working together on a more integrative and cross-
112 sectorial approach to tackle health challenges. It gathered more than 130 institutions from 35
113 countries and built the foundations of RD ecosystem by integrating multinational RD funding,
114 support services and data infrastructure (virtual platform of distributed FAIR data sources and
115 services). The Partnership will benefit equally from the outputs but the ones that are key for the RDP
116 of several other key programmes and initiatives supported by the EU (to only name few) as the
117 European Reference Networks (ERN), their registries and their clinical research coordination
118 platform ERICA; IMI projects like Connect for Children (C4C) pan-European collaborative paediatric
119

123 network for high quality clinical trials in children, and Screen4Care; Orphanet, the EU-funded
124 multilingual knowledge base on rare diseases and orphan drugs including ORPHA codes ontology;
125 EU-funded research projects such as Solve-RD, accelerating RD diagnosis pathway for unsolved
126 rare diseases for which the molecular underlying cause is not yet known; RD-Connect, a European
127 genome-phenome analysis platform including directory of RD biobanks and samples; the European
128 Rare Diseases Registry Infrastructure implemented by JRC, projects such as X-eHealth and EHDEN
129 that target millions of health data records, and the 1+Million Genomes initiative targeting 1 million
130 sequenced genomes accessible in the EU including RDs as key use case.
131 Finally, the national contributions will be essential to the European RD Partnership to ensure long-
132 term commitment, integration of resources and best alignment of the national plans and/or
133 national strategies to tackle rare diseases.

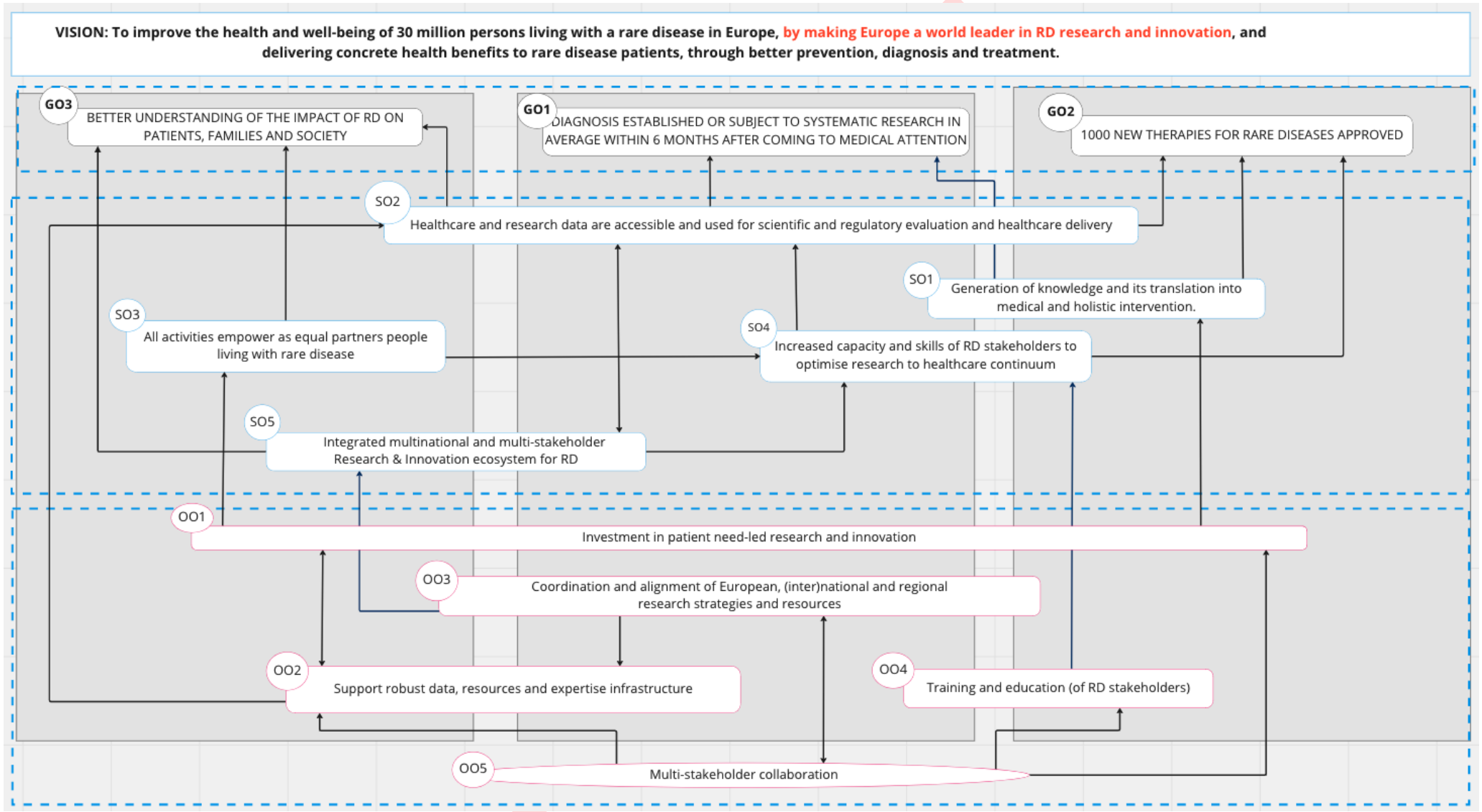
134
135 The RDP will consolidate and extend the achievements of EJP RD so that other actors can contribute
136 more easily and efficiently to the generation of evidence that leads to concrete benefits for
137 patients.

138
139 The ambition of the European RD Partnership will build on, contribute to and accelerate directly the
140 goals set by the International Rare Diseases Research Consortium¹ (IRDiRC). This is reflected in the
141 General Objectives of the Partnership. The programme aims to provide solutions for tackling the
142 identified main R&I bottlenecks that hinder the efficient development of better diagnosis, therapy
143 and care fostered by research in the RD field which are: 1/the need for further collaboration &
144 alignment of research funding and optimal integration with national rare diseases plans/strategies;
145 2/ the huge gap in translation of research results to deliver cost-effective solutions for people living
146 with a rare diseases (noting that the conduct of clinical studies is a burden that can be addressed);
147 and 3/ the fragmentation of knowledge and data, lack of holistic R&I ecosystem. Research &
148 Innovation activities on rare diseases should create value for patients by reducing suffering of
149 people living with rare diseases through better prevention, better diagnosis and better treatment
150 as a direct result of research outcomes. The European RD Partnership should drive the research cost
151 effective translation and bringing innovation to address the unmet medical needs of the rare
152 diseases community, while coordinating national research efforts and establishing a holistic
153 research and innovation ecosystem of knowledge, data, disciplines, people and sectors.

¹ IRDiRC goals for the decade 2017-2027 are: Goal 1 – All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline; Goal 2 – 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options; Goal 3 – Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patients (Future of Rare Diseases Research 2017–2027: An IRDiRC Perspective. C.P. Austin et al. Clin Transl Sci. 2018 Jan;11(1):21-27. [doi: 10.1111/cts.12500](https://doi.org/10.1111/cts.12500). Epub 2017 Oct 23)

156

1.3. intervention logic - Partnership Specific Impact Pathway (PSIP)



157

158 **1.4. General Objectives²**

159 The General Objectives (GOs) of the European Rare Diseases Partnership are defined
160 in line with the Partnership's vision and mission to improve the health and well-being of
161 people affected by rare diseases by delivering concrete health benefits through
162 prevention, diagnosis and treatment development. It was agreed that they should be
163 inspired by and fully aligned with the goals of IRDiRC. Moreover, they are contributing
164 to EU political priorities (cf. "Ambition") and to the Sustainable Development Goals
165 (SDGs) of the 2030 Agenda for Sustainable Development adopted by the United
166 Nations in 2015. In particular, they are affiliated with SDG3 "Ensure healthy lives and
167 promote well-being for all at all ages", SD9 "Build resilient infrastructure, promote
168 inclusive and sustainable industrialization and foster innovation", and SDG10 "Reduce
169 inequality within and among countries".

170

171 **1.4.1. GENERAL OBJECTIVE 1: DIAGNOSIS ESTABLISHED OR ENROLLMENT**
172 **IN SYSTEMATIC RESEARCH IN AVERAGE WITHIN 6 MONTHS AFTER**
173 **COMING TO MEDICAL ATTENTION**

174 Patients with undiagnosed diseases and their families often face an uncertain and
175 unpredictable journey, called a diagnostic odyssey, which is particularly complex in
176 the case of rare diseases as 50% of patients still do not have a final diagnosis, and
177 when they do, it is on average after 4 years of the diagnostic journey.

178 The European RD Partnership will contribute to shortening the diagnostic pathway for
179 patients with rare diseases. For those disorders already identified in the literature the
180 ambition is that a patient is diagnosed within a maximum of six-months after the first
181 medical appointment with a specialist. For the undiagnosed disorders efforts will be
182 made to build and/or strengthen the bridge between research and healthcare to
183 provide to every undiagnosed patient the possibility to be included in a globally
184 coordinated diagnostic and research pipeline.

185

186 **1.4.2. GENERAL OBJECTIVE 2: 1000 NEW THERAPIES FOR RARE DISEASES**
187 **APPROVED**

188 95% of RDs are still underserved in terms of research and patients with rare diseases,
189 although diagnosed, face a lack of viable long-term treatment options. To contribute
190 to IRDiRC goals, and more specifically Goal 2 – "1000 new therapies for rare diseases
191 will be approved, the majority of which will focus on diseases without approved
192 options", the European RD Partnership, will accelerate the development of new
193 therapies (especially for diseases without approved options) by providing the
194 necessary support to research projects aimed at developing new treatments and by
195 expediting clinical trial readiness of rare diseases, including contribution to regulatory
196 fitness to enable regulatory approval.

197

² General Objectives correspond to the impact aimed to be achieved by the European Rare Diseases Partnership, i.e., the wider & long term effects on society (including the environment), the economy and science, enabled by the outcomes of R&I actions.

198
199

1.4.3. GENERAL OBJECTIVE 3: BETTER UNDERSTANDING OF THE IMPACT OF RD ON PATIENTS, FAMILIES AND SOCIETY

200 Rare diseases place a significant burden on patients and their families, on caregivers
201 and on society in general. For 52% of them, rare diseases mean a serious impact on
202 their daily lives. Understanding of the impact of RDs on people lives means also better
203 evaluation of the societal and healthcare costs and capacity to implement more
204 inclusive, holistic healthcare approaches. Through required means (funding, data
205 collection) and processes (involvement of people living with rare diseases at all levels)
206 the Partnership will contribute to capturing of RD impact and comprehensive
207 understanding of patients and carers needs leading to, in the long term, improved
208 and/or new processes that will facilitate the diagnosis and care pathways and
209 translate into meaningful societal support.

210
211

1.5. Activities and resources (Operational Objectives³)

Operational Objective 1: Investment in patient need-led research and innovation

214 The European RD Partnership will implement annually competitive Joint Transnational
215 Calls (JTC) to fund patient-needs driven, multinational, research projects, including the
216 funding of dedicated support to patients' organisations. Specific measures will be
217 applied for the JTCs in order to improve the participation and visibility of under-
218 represented countries in the European RDP.

219 Other funding schemes will be used including support to expanding or establishing
220 new networks for knowledge sharing targeting underserved Rare Diseases, and fund
221 in-house research, through the Clinical Research Network (CRN)⁴. These latter funding
222 schemes will be supported by direct use of EC funds, complemented by in-kind
223 contributions of involved research performing organisations and possible in-cash
224 and/or in-kind contributions of industry. Clinical trials conduct could benefit from these
225 funding schemes; the resources expected for their implementation will need to be
226 estimated.

227

Operational Objective 2: Support robust data, resources and expertise infrastructure

228 The state-of-the-art infrastructure, services and support will be further advanced so
229 that clinical and translational RD research are highly productive.

231 The infrastructure of the clinical research network will be established by leveraging on
232 and expanding and connecting existing resources and tools (e.g., EU RD Platform, EJP
233 RD Virtual Platform, European patients' registries and biobanks, as well as other
234 national data sources & capacities). This infrastructure will comprise dedicated
235 support services that will include, but are not limited to, provision of distributed and
236 cloud computing and data exploitation facilities, innovative analysis resources, quality
237 assurance services, research guidance on coordinated diagnostic, Patient-Centred
238 Outcome Measures, biostatistical and multinational Clinical Trials. Other ad-hoc

³ Operational Objectives correspond to the actions, activities and resources that will be deployed by the European RD Partnership to achieve its Specific and General Objectives.

⁴ The Clinical Research Network has for objective to promote efficient implementation of clinical studies and preparedness for clinical trials. It connects various resources from the European RD Partnership partners and collaborators, supported by an IT infrastructure. It conducts internal research projects that are selected through internal calls and are backed by dedicated services, including but not limited to diagnostic research support, biostatistical guidance, clinical trials methodologies and operations, and Clinical Outcome Assessment support. Its IT infrastructure utilizes existing resources and platforms and extends them to allow for data exploitation for in-house research projects and piloting of the CRN.

239 support services (e.g., for identification of biomarkers and surrogate endpoints and
240 validation, mHealth expertise) can be developed according to the emerging needs
241 of the CRN. Importantly, these integrative services will be expanded, developed and
242 deployed to support all activities of the European RD Partnership (beyond CRN). They
243 will comprise data integration and coordination services and expanded mentoring
244 services to support all funded projects. Ethics, Legal, Regulatory and Societal Impact
245 support will also be implemented.

246

247 **Operational Objective 3: Coordination and alignment of European, (inter)national and**
248 **regional research strategies and resources**

249 A fully integrated strategy and coordination will support effective public, public-
250 private and civil society partnerships. National coordination and alignment will be
251 ensured through maximisation of the national in-kind contributions in advance and all
252 along the lifetime of the Partnership. The National Mirror Groups (NMGs) will be set-up
253 and supported to organise coordinated interaction between the Partnership and
254 national and regional stakeholders. They will catalyse the transfer of good practices
255 to the national and regional level, including leveraging the power of
256 national/European data sources, by making nationwide or regional RD discoverable
257 and actionable for international RD research.

258 The cooperation with international partners will be ensured through (1) the operation
259 of the IRDiRC Scientific Secretariat to provide strong links to international collaborators
260 (such as the US National Institutes of Health) as well as a joint management of research
261 and innovation strategy; (2) the maintenance of already established collaboration
262 with Associated partners (like Canada, Israel and Australia) contributing to and
263 aligning with research and training activities; (3) expanding the collaboration and
264 integration of other countries willing to join with their knowledge and resources; and
265 (4) Stimulating and supporting the development of trans-regional activities.

266

267 **Operational Objective 4: Training and education (of RD stakeholders)**

268 The Capacity building of all stakeholders will support new generations of researchers,
269 clinicians, patient representatives and policy makers, decrease knowledge and
270 competences gaps between countries, empower patients and constantly improve
271 the capacities of the experienced RD stakeholders.

272 The European RD Partnership will integrate training and capacity building components
273 as part of its support activities for funded research projects and Clinical Research
274 Network. Dedicated efforts will be made to train patients and their representatives on
275 topics of relevance to ensure and accelerate their informed engagement at all levels.
276 To support access to RD education for overall society and stakeholders, comprising
277 general student and clinician population interested in RDs, including at national level,
278 the Partnership will take advantage of already initiated by EJP RD massive open online
279 courses and expand them to accredited education programmes.

280

281 **Operational Objective 5: Multi-stakeholder collaboration**

282 All types of actors will be involved, along the health and research value chain, in
283 priority setting. These include research funders; research and innovation communities
284 across life science and technology/data disciplines; users represented by patients and
285 citizens, health care professionals and health care providers; as well as EU-wide and
286 national policy makers, regulatory authorities, Health Technology Assessment bodies,
287 and health care payers. The European RD Partnership will gradually bring on board
288 additional stakeholders. Mechanisms will be created to onboard Under-represented,

289 including EU13⁵ countries, Associated and non-EU countries. The possible inclusion of
290 industry as full beneficiaries in the partnership is considered as major gamechanger in
291 building integrative RD ecosystem and advancing European RD Partnership goals. This
292 inclusion needs to happen in full synergy with some other initiatives listed in annex 1.

293 The multistakeholder collaboration, that is at the root of the partnership, requires an
294 effective governance framework. The Terms of Reference and guidance for the
295 governance of the partnerships under Horizon Europe, that will be provided by the EC,
296 and learnings from other initiatives such as EJP RD, will be used to set the organisational
297 and governance structure of the consortium that will comprise decision-making
298 bodies; executive bodies and advisory bodies. A central coordination and
299 management of the consortium will take advantage of experience and tools already
300 acquired through EJP RD to establish active and proficient coordination office that will
301 accompany European RD Partnership partners by providing operational and strategic
302 support. This will include the management of the monitoring of partnership
303 operational, specific and general objectives through adapted monitoring system in
304 line with the requirements of Horizon Europe.

305
306 The detailed breakdown of resources to specific activities will be decided by the RD
307 partnership decision-making bodies when adopting annual work programmes,
308 considering advice from the constituted advisory bodies. The description of specific
309 activities and allocated resources will be provided in annual activity reports. The
310 annual activity reports will also report on the Key Performance Indicators used to
311 monitor progress towards reaching the European RD Partnership objectives, with
312 specific baselines and targets.

313
314

315 **1.6. Synergies with other initiatives**

316 To reach its ambition, the European RD Partnership will leverage relevant
317 complementary activities in Europe and will conversely generate content that may
318 benefit other EU initiatives.

319 Collaborations are envisioned with (i) Horizon Europe European Partnerships, (ii)
320 European Union programmes, projects and initiatives, (iii) large European or
321 international initiatives, should they be public, public-private or private including no-
322 for-profit.

323
324 Synergies will be sought with the aim to support and enhance specific RDP actions
325 (including possible co-funding, parallel funding or subsequent funding), as well as to
326 ensure relevant dissemination and exploitation of results from the European RD
327 Partnership. For instance, regional funds can support the uptake of evidence-based
328 results from e.g., the funded research projects, the services-innovations and other
329 innovations identified through the European RD partnership.

330 For each collaboration opportunity, "opportunity topics" cover diagnosis, treatment,
331 care, research, data and infrastructures that set out the roadmap for the next decade
332 of rare disease policies.

333
334 Key collaboration opportunities have been identified with **several EU Partnerships**
335 implemented in the Horizon Europe context in three main areas: (i) the Health Cluster
336 (ii); the Digital, Industry and Space Cluster;(iii) partnerships with cross-sectoral themes.
337 A close collaboration will be initiated with other European Health Partnerships, starting

⁵ List of EU13 countries: Bulgaria, Croatia, Cyprus, Czech Republic, Estonia, Hungary, Latvia, Lithuania, Malta, Poland, Romania, Slovakia, Slovenia

338 with: (1) the Innovative Health Initiative (IHI), (2) the **ERA4Health** - Fostering a European
339 Research Area for Health Research, as well as (3) the European Partnership on
340 **Personalised Medicine** and (4) the Partnership Transforming Health and Care Systems
341 (**THCS**). Aside from the Health cluster, collaboration is also foreseen with cross-sectoral
342 Partnerships such as the **EIT Health**, **Innovative SMEs** and European Open Science
343 Cloud (**EOSC**). Finally, to ensure the best uptake and alignment in data, computing
344 and machine-learning research areas, two Partnerships lying under the Digital, Industry
345 and Space Cluster have been identified as potential candidate for partnerships, one
346 on High Performance Computing (**EuroHPC**) and one on **Artificial Intelligence, data**
347 **and robotics**. These initiatives, and others with potential for collaboration, are listed in
348 annex 1.

349
350 The European RD Partnership will also take advantage of pre-existing and to-be
351 funded **EU Programmes and EU projects** to maximise the use of resources and
352 alignment. The European RD Partnership will namely build synergies with Horizon
353 Europe initiatives, such as the European Innovation Council (**EIC**), the Marie
354 Skłodowska-Curie Actions (MSCA); and Missions, in particular the **Cancer Mission**. The
355 Partnership will also develop specific synergies with the **EU4Health** and the **Digital**
356 **Europe** Programmes. Other EU support schemes such as the European Social Fund Plus
357 (**ESF+**), the invest in education, employment and social inclusion (**InvestEU**) and the
358 European Regional Development Fund (**ERDF**) will also systematically be considered
359 to develop the best uptake and development of the European RD Partnership
360 activities. Several EU programmes and projects have been pre-identified for potential
361 collaboration (see annex 1).

362 In addition to EU-funded partnerships and programmes, collaboration will also be
363 developed with overarching European or international major initiatives such as the
364 Rare Disease Moonshot launched in December 2022 or the Together4Rare initiative.
365 Collaboration with non-for-profit organisations and charities who are paving the way
366 in RD research collaboration will also be sought.

367
368

369 **2. Specific Objectives⁶ of the European Rare Diseases** 370 **Partnership**

371 **2.1. Specific Objective 1: Generation of knowledge and its** 372 **translation into medical and holistic intervention**

373 **2.1.1. Challenge**

374 The journey from bench to bedside should be accelerated thanks to the generation
375 of knowledge and its translation into medical and holistic intervention, but still faces
376 the following challenges:

377

378 **Insufficient support of RD research**

379 More than 90% of RDs are not properly addressed in terms of research and
380 accompanying sustainable R&I funding. From the scientific perspective this includes
381 lack of knowledge of the underlying molecular disease cause, pathophysiology, lack
382 of disease models and potential therapeutic targets within a disease/ disease group
383 hampering diagnosis and development of suitable treatment options. From the

⁶ Specific Objectives correspond to the outputs (direct results of the project) and outcomes (short/medium term effect of the projects results) that the European Rare Diseases Partnership aims to achieve.

384 funding perspective, the high risk-to-investment return ratios for private companies
385 discourages their engagement in RD therapeutic development, and concomitant
386 lack of alternative R&I pathways to the patient slows down the journey from the bench
387 to bedside. A streamlined and optimized, jointly driven public-private pipeline is
388 needed, supported by powerful data management.

389 **Need for more innovative RD research models**

390 One disease - one treatment equation is not a viable option for 7000 rare diseases.
391 Standard research in common disease conditions explores cell, tissue and animal work
392 in individual disease states in comparison with health, to identify disease pathways and
393 potential therapeutic targets. In RDs very small numbers of patients affected with
394 individual rare diseases make the use of such standard clinical development
395 pathways often impracticable. Specific approaches and linked research
396 infrastructure are not currently in place to explore innovative options like studying
397 groups of RDs with common underlying pathophysiology, or decentralized studies
398 leveraging on telemedicine, remote outcome evaluation, and data science, to
399 expedite the research yield and identify new therapeutic agents or re-purpose existing
400 therapies.

401 **Dysfunctional regulatory components**

402 On one hand, the generation of regulatory-compliant research results, and thus the
403 translation and uptake of academia-driven research, is often compromised by lack of
404 timely regulatory advice and interaction beforehand with regulators. On the other
405 hand, there is a need to engage and boost regulatory science to provide a robust,
406 more digital framework and accelerate implementation of novel technologies,
407 innovative trial design or the use of Real-World Evidence (RWE) in study design and
408 development.

409

410 **2.1.2. Scope**

411 To address the above challenges and enable accelerated translation of knowledge
412 into health interventions and other services, under this specific objective the
413 partnership will enable patient-need led relevant science by providing a RD research
414 support pipeline from basic research to clinical trial readiness. To better target
415 underserved RDs activities it will investigate mechanisms underlying disease and
416 disease progression, biomarkers and identification and validation of other tools to
417 promote prevention, inform development of treatments, diagnostics, and other
418 innovative healthcare solutions. Attention will be paid to Social Sciences and
419 Humanities research to better understand the impact of rare diseases and the
420 potential benefit of new interventions. Furthermore, the partnership will explore
421 research/diagnostic/therapeutic/data science approaches for multiple diseases with
422 common aetiology/pathway/other characteristics, taking account of their impact on
423 regulatory requirements and processes.

424 The integration across value chain will be addressed by combining research financed
425 and performed by both public and private stakeholders and involving patients. This
426 coupled with effective support services including state-of-the-art data infrastructure
427 (SO2) and research pipeline coordination will directly boost innovation in rare disease
428 diagnostics, therapeutics and other interventions such as prevention. The partnership
429 will aim to unite and strengthen the research ecosystem by creating infrastructures
430 that address connectivity and maximize various public and private resources to
431 support all steps of R&D, from discovery to late development, to post-marketing
432 obligations and backtranslation. Thereby, the partnership will increase reproducibility
433 of results and accelerating discovery, translational research and development.

434 Investment in outcome-oriented research projects, actively monitored and steered
435 towards translational opportunities will ensure their outputs meet regulatory
436 requirements and patients' needs thereby reducing failure rates of therapeutic
437 developments. The Partnership will aim to support processes from preclinical to late
438 development considering regulatory requirements. It will support development,
439 regulatory acceptance, upscaling and deployment of innovative clinical trial
440 methodologies (pooled design and analysis methods, AI, use of different sources of
441 evidence, including RWE, data necessary to inform reimbursement decisions) for small
442 and very small populations. Attention will be paid to demonstrate the value of new
443 methodologies to standardize and benchmark them against existing regulatory and
444 HTA evaluation and approval processes to help adapting them to rare disease
445 specificity and engage regulatory acceptance.

446
447 Activities under this specific objective will be enabled by and will inform those of
448 Specific Objective 2 (Data). Deployment of new methodologies in research,
449 regulatory and HTA practice and health practice will rely on supportive activities under
450 Specific Objective 4 (Capacity building). The integration of public and private
451 resources into one research support pipeline will contribute to the strategy of Specific
452 Objective 5 (Integrated multinational and multi-stakeholder R&I ecosystem for RDs).

453

454 **2.1.3. Potential Outputs**

- 455 • RD funding programme based on long-term (*[amount to be defined]* years)
456 funding commitment and robust prioritization strategy.
- 457 • At least *[amount to be defined]* M€ invested in RD research focused on
458 underserved rare diseases., including on the impact of RD on patients, families and
459 society.
- 460 • *[amount to be defined]* M€ invested in projects using secondary use of clinical data
461 and reuse of research data for RD prevention, earlier diagnosis, treatment, and
462 mitigating impact on the life of people living with a rare disease.
- 463 • All funded projects accompanied by sustainable and integrative support services
464 to accelerate the development-ready research and to guarantee generation of
465 exploitable output.
- 466 • Functional RD research funding accelerator hub⁷ ensuring smooth transition and
467 support all along value chain to expedite research results into products. Fully
468 integrated and mutually synergistic non-clinical & clinical trial readiness RD
469 research pipeline (including Clinical Research Network).
- 470 • The capacity of relevant clinical expertise coupled to methodological excellence
471 exploited in coordination with regulators/HTA, to support evidence-based research
472 accelerating the entry into market for the patient benefit.

473

⁷ The acceleration hub aims at promoting innovation, encourage collaboration, and support the translation of scientific discoveries into real-world applications that benefit society. As a collaborative and interdisciplinary service, it brings together researchers, entrepreneurs, investors, and other stakeholders to accelerate the development and commercialization of scientific and medical innovations. It offers a range of resources and services, such as funding, mentorship, access to specialized equipment, training, networking opportunities, and regulatory guidance, that help researchers and entrepreneurs move their ideas from the laboratory to the market more quickly and efficiently. . Within the European RD Partnership, the acceleration hub will have a large scope including, but not limited to, biotechnology, drug development, medical devices and digital health, and will leverage on its public-private collaborations.

474 **2.1.4. Specific Outcomes**

- 475 • Research initiated in [*amount to be defined*] % of underserved rare diseases.
- 476 • Higher number of successful basic research projects transitioning to preclinical
477 development.
- 478 • Increased number of academic projects transitioning to industrial development in
479 the EU.
- 480 • Public Early-stage investment coordinated with later stage investment by private
481 sector and philanthropy.
- 482 • Better and faster integration of novel technologies and methodologies along the
483 RD healthcare pathway with a focus on specific subareas such as diagnosis,
484 devices, trial readiness and integrated care.
- 485 • Increase in number of RD cases with a diagnosis.
- 486 • Increased integration of RD research and care.
- 487 • Increased number of investigational medicinal products implemented into clinical
488 research and developed in Europe.
- 489
- 490

491 **2.2. Specific Objective 2: Healthcare and research data are**
492 **accessible, and used, for scientific and regulatory evaluation**
493 **and healthcare delivery**

494 **2.2.1. Challenge**

495 Projects and initiatives such as the EJP RD, JRC, ERICA, ERNs, RD-Connect, Darwin, C-
496 Path, and Solve-RD, together with ELIXIR, 1+MG, BBMRI-ERIC, EOSC, and EHDS are
497 gradually providing the foundations of a powerful, standards-based European RD
498 data ecosystem. Herein, the RD community embraced the FAIR principles [[Wilkinson](#)
499 [et al.](#)] to optimize how data can be used to reach tangible results.

500 Nevertheless, the full potential of healthcare and research data for research,
501 innovation, regulatory purposes, and healthcare delivery in the RD domain remains
502 untapped to significant extent. There are major challenges regarding the awareness
503 and integration of the accessible resources and the skills to fully exploit the data
504 ecosystem. Challenges include planning studies that use data from multiple sources,
505 analysing and interpreting data from such studies (e.g., by explainable AI and
506 interdisciplinary collaboration), and translating insights from data research into
507 actionable results e.g., treatments for individuals, clinical guidelines, development of
508 drugs and devices, increased technology readiness, and improved HTA and
509 reimbursement decisions. Increasing the capability of data producers in applying
510 standards for accessibility, quality and interoperability of data is still a challenge. Full
511 exploitation of data for the global objectives depends on widespread adoption of
512 these standards.

513

514 **2.2.2. Scope**

515 The partnership will aim at strengthening selected ongoing and new actions to harness
516 opportunities that well-managed healthcare and research data present for rare
517 diseases. Opportunities include qualifying data pertinent to innovation for regulatory
518 purposes, optimizing clinical trial readiness within the EU Clinical Research Network, RD

519 diagnosis in EU wide initiatives (e.g., EU-wide undiagnosed program), understanding
520 RD impact and burden, and exploiting patient-centred outcomes.
521 The European RD Partnership will support the generation, pooling, integration and
522 sharing of high-quality and interoperable RD data in an expanding ecosystem of
523 distributed FAIR data sources, building on existing infrastructures encompassing the
524 European Platform on Rare Disease Registration, the EJP RD Virtual Platform network,
525 RD-Connect, and services not specific for RD (e.g., BBMRI-ERIC and ELIXIR). It promotes
526 advanced data analysis and data interpretation methods and approaches that
527 exploit this ecosystem. Approaches are as federated as possible and as centralised
528 as needed to enable robust and flexible data use scenarios that promote
529 collaboration among European countries and stakeholders, facilitate research,
530 innovation and regulatory qualification of data, as well as better translate into tangible
531 healthcare benefits for RD patients, contributing thus to the SO5.
532 The partnership will also support the development of data-driven computational tools,
533 statistical and artificial intelligence methods, as well as digital solutions to understand
534 the diseases progression, to solve undiagnosed RD cases and implement new clinical
535 studies/trials designs for small populations, this will be enabled by and will inform the
536 activities of the Specific Objective 1. The involvement of RD patients and clinicians is
537 essential to ensure that advanced computational data access, analysis and
538 modelling tools are being developed, considering user needs, utility and sustained
539 exploitation early on, with patient's health outcome improvement being the key
540 driver. This will rely on supportive activities of SO3 and SO4 for patients' empowerment
541 and capacity building of RD stakeholders.
542 Advancing RD data standards, harmonising data access services and deploying high
543 performance data analysis capacities will be promoted within the partnership in
544 coordination with the activities of the SO5, through the collaboration with existing
545 national, EU and international data initiatives and infrastructures.
546

547 **2.2.3. Potential Outputs**

- 548 • Exploitation of FAIR repositories of clinical and omics RD data on a European scale,
549 extending to Patient Reported Outcome Measures (PROMs), longitudinal real-
550 world observations, streaming data, and data from wearables.
- 551 • An EU-wide undiagnosed programme based on the effective detection of
552 undiagnosed RD patients in national health systems and on an infrastructure of FAIR
553 reference data (phenomics, genomics, multi-omics, etc.).
- 554 • A comprehensive data infrastructure based on FAIR principles, existing resources,
555 data protection regulation, and quality standards including validated Patient
556 Centred Outcome Measures (PCOMs) to support patient-centred research, as well
557 as regulatory and HTA decision making.
- 558 • Promoting the regulatory qualification of RD data with the goal of accelerating the
559 development and access of therapies, and measurements or methods that aid
560 therapy development across rare diseases.
- 561 • A FAIR data-based framework exploiting patient-driven health and socioeconomic
562 studies to inform policy decisions.
- 563 • Widespread use of data-driven computational tools and models, artificial
564 intelligence methods and digital solutions that exploit the FAIR data ecosystem to
565 advance trial readiness, to solve undiagnosed RD cases, to understand disease
566 progression, develop and validate clinically meaningful trial outcomes, and
567 implement innovative clinical study/trial designs for small populations.

- 568 • Collaboratively developed knowledge platform for open access, dissemination
569 and sharing of scientific knowledge, including negative results and data.
- 570 • Data standards for making new types of data elements FAIR, including real world
571 observations and evidence, patient reported outcomes, and data required to
572 satisfy regulatory requirements.
- 573 • Demonstrations of the value of a critical mass of FAIR data for secondary use of
574 clinical data and reuse of research data for RD prevention, earlier diagnosis,
575 treatment, and mitigating impact on the life of people living with a rare disease.
576
577

578 **2.2.4. Specific Outcomes**

- 579 • Improved RD diagnosis (higher diagnostic yield, earlier diagnosis) through FAIR
580 data use.
- 581 • Support for symptomatic patients without a satisfactory diagnosis.
- 582 • Improved trial readiness and therapeutic options through FAIR data use.
- 583 • Accelerated development of therapies across rare diseases, facilitated through
584 regulatory-grade data that are FAIR for analytics supporting RD characterization.
- 585 • Increased accuracy of diagnosis and individualised treatments from clinical
586 decision support using advanced data driven methodologies/analytics.
- 587 • Reduced time-to-use of therapeutic solutions in a clinical context by advanced
588 data driven methodologies/analytics.
- 589 • Increased availability and usability of RD innovation.
- 590 • Demonstrated added value of digital health tools for RD.
- 591 • Researchers, patients and clinicians are increasingly re-using and sharing RD data
592 to implement multinational research for delivering new concepts in RD
593 pathophysiology, new diagnostics, novel drug targets, biomarkers and new
594 disruptive approaches for clinical research.
595
596

597 **2.3. Specific Objective 3: All activities empower, as equal** 598 **partners, people living with rare disease**

599 **2.3.1. Challenge**

600 Research on RD should create value first and foremost for patients. People living with
601 a RD are often the most motivated stakeholders to make progress on their disease
602 given the number of patients living with the disease is low and that knowledge,
603 expertise and funding are scarce. At the same time, patients and carers are often a
604 significant source of expertise related to individual rare diseases. Only by harnessing
605 patient expertise, together with clinical and research expertise, can we address the
606 challenges posed by RD.
607 Although patient engagement is recognised as a cornerstone of the RD ecosystem,
608 obstacles remain to genuine and significant involvement of patients in research. More
609 specific challenges arise for the 'undiagnosed' and ultra-rare diseases, where
610 collaboration across sectors and geographic borders is indispensable but where
611 research activity lacks scale and visibility among patients who would like to
612 participate. Resources are not targeted to research on RD with the highest unmet

613 needs and access for patients to interrogate limited existing research sources is not
614 eased. Patient involvement is not systematic and/or capitalised on to generate data
615 that support decisions making by regulators or payers.
616 Furthermore, there is currently insufficient patient participation at all levels of research
617 to enable productive and sustainable partnerships between researchers and patients.
618 This includes incentives (funding, regulatory) to enable equitable inclusion of Patients
619 Living With a Rare disease (PLWRD) and/or representatives from the earliest point of
620 research or participation of patients/patient organisations as co-designers of research.
621 Coordinated cooperation in the development of the RD disease specific patient-
622 centred outcome measures (PCOMs/PROMs), consideration of patient preferences,
623 and co-development of Real-World Evidence (RWE) must also be stimulated. Thus, an
624 organized framework for patient involvement in research, building upon what has
625 been initiated by the EJP RD, is required to systematically support patient-centred
626 research and deliver new innovations.
627

628 **2.3.2. Scope**

629 The European RD Partnership will provide an inclusive pathway and adequate
630 resources to empower PLWRD and/or representatives as equal partners. PLWRD will be
631 involved at all levels of governance and execution of the European RD Partnership,
632 with training or induction as necessary. A structured, flexible and coherent framework
633 for patient engagement in research will be developed which will be adaptable at
634 national levels and will promote best practices, re-using and extending existing
635 resources (such as PARADIGM, EJP RD PENREP⁸, etc.). Patients and/or representatives
636 will be active and equal partners in planning and prioritising research activities,
637 engaging in projects and facilitating patient engagement across all research
638 activities, encompassing implementation, monitoring and dissemination of projects'
639 results.

640 Training for patients/patient representatives will be provided on a continuous basis to
641 ensure and accelerate their informed engagement at all levels. Patients will also have
642 a role in identifying training needs for researchers and clinicians working with people
643 with rare diseases, so that training on patient involvement in research will be provided
644 to funded projects.

645 Novel and more inclusive funding models will be developed to ensure sustainable
646 patients' involvement in research projects and to ensure that availability of funding is
647 not a barrier to patient participation at a national/regional level. PLWRD will be
648 engaged in decision-making on the allocation of funding to research projects
649 (including evaluation and monitoring).
650

651 The European RD Partnership also aims to reduce inequities between different types of
652 RD by targeting underserved RDs through meaningful empowerment, engagement,
653 and leadership of patients or their advocates, building new or expanded networks
654 and supporting dedicated research.

655 In developing this inclusive pathway, the European RD Partnership will take advantage
656 of the existing infrastructures like Patient Advocacy Organisations (agnostic or RD
657 specific), RD Patient National Alliances, the ERNs and their European Patient
658 Advocacy Groups (ePAGs), charities, etc.
659

⁸ The EJP RD PENREP, Patient Engagement in Biomedical Research Project, working group is composed of patients' representatives and research funders who aims to encourage fruitful, sustainable and enduring partnerships between scientists and patient organisations, co-leading the way for systematic patient-centered research.

660

661

2.3.3. Potential Outputs

662 • Patient-informed decision making, on which unmet needs to investigate and
663 prioritize in research is made.

664 • Patient representation in all governance structures within the European RD
665 Partnership.

666 • Patients/patients' representatives involved in all research applications and on
667 steering/governing committees of all funded RD studies.

668 • Effective patient partnerships enabled through dedicated funding of patient
669 organisations contributing to research projects.

670 • Agreed mechanisms to feedback research results in a consistent and systematic
671 way to relevant patient groups.

672 • Awareness and adequate signposting of the infrastructures and resources
673 available to support and guide patients in the RD Research landscape.

674 • Patient empowerment through capacity building and training activities resulting in
675 proactive patient partnerships in research.

676 • Increased knowledge within PLWRD to further understanding of rare diseases.

677 • The training on patient involvement in research is coupled to every funded
678 research project.

679 • PCOMs/PROMs co-developed by PLWRD and applied across all relevant funded
680 research and all 24 ERNs.

681 • Guidelines developed to support equitable patient inclusion to inform researchers,
682 regulators and funders at the national and European levels.

683

684

2.3.4. Specific Outcomes

685 • Increased participation of patients/patient organisations as co-designers of
686 research Innovative and disruptive approaches in funding and developing
687 patient-centred research benefitting the whole health research ecosystem.

688 • Patient voices are considered when deciding about research priorities and
689 strategies.

690 • A greater sense of shared ownership of the research process/outcomes.

691 • Trusted relationships to access resources, expertise and the support required to
692 translate research into positive health impact.

693 • A better understanding of the real needs and preferences of patients informing
694 research questions and driving new design interventions.

695 • Healthcare solutions assessed according to criteria that matter to patients and
696 public contributing to achieving people-centred healthcare.

697 • Building legal requirements for equitable inclusion in all levels of engagement in
698 research.

699

700

2.4. Specific Objective 4: Increased capacity and skills of RD stakeholders to optimise research to healthcare continuum

2.4.1. Challenge

The capacity building element is often underestimated when considering the long-term strategy for building strong rare diseases ecosystem. Despite several efforts deployed by the EJP RD, ERNs or EURORDIS to provide a wide range of knowledge sharing, training and educational activities for RD research stakeholders, there is still an unmet need for an integrated concept combining systematic and comprehensive knowledge transmission with targeted acquisition of specialized skills in order to increase the EU's RD research capacity in an efficient and sustainable manner.

Both raising new generations of RD researchers/clinical specialists/patient experts and continuous acquirement of new competences by RD stakeholders are main challenges augmented by fragmentation and lack of sustainability of existing training and education programmes. This is even more evident at national level where specialised curricula are incomplete or simply do not exist and the sharing of available knowledge is slowed down due to the language barriers. Furthermore, efficient capacity building is hampered by the absence of a central knowledge hub allowing on the visibility of existing expertise and contributing to better alignment of efforts deployed under different initiatives (including the bottom-up funding programmes of the European Commission that generate important volume of RD-related projects).

2.4.2. Scope

The partnership will incorporate capacity building activities as **integral part of the rare disease research pipeline**. Alignment with the knowledge generating actions of the initiatives (ERNs, C4C, STARS, etc.) will be sought. This will enable, on one hand, upgrading of scientific, technology (including FAIR approaches) but also regulatory knowledge of stakeholders participating in research projects financed through competitive calls but also those performing "in house" research activities as part of the Clinical Research Network of the European RD Partnership. On the other hand, new generations of RD researchers will be equipped with state-of-the-art competences. Young researchers will be given the opportunity to train during interdisciplinary liaison programmes and secondment coupling clinical and non-clinical activities.

To unlock the access to RD top-level education to all, the partnership will develop an accredited, comprehensive online education programme taking stock of highly performing pre-existing modules complemented by novel training units.

The model of "train the trainer" and innovative language AI technologies will be used to expand and deliver capacity building programmes in all countries participating in the partnership.

Finally, the partnership will provide a central platform for knowledge sharing by gathering and enabling access to relevant expertise (comprehensive catalogue & helpdesk) and ensuring connection with all existing RD projects and initiatives. This will provide novel opportunities for collaboration, improve the visibility of RD stakeholders and optimise the use of resources by enhancing the performance of previously disconnected activities.

2.4.3. Potential Outputs

- All researchers in funded projects have access to suitable training courses/certification.

- 749 • Interdisciplinary mobility programmes for early career researchers linked to and
750 optimally serving the needs of consortia supported by the partnership.
- 751 • European Master graduation programme enabling training of new generations of
752 RD researchers.
- 753 • RD stakeholders empowered and mastering methodologies required to generate
754 and use good-quality data according to European standards.
- 755 • Increased participation of researchers from under-represented countries in
756 education/training programmes.
- 757 • Train-the-trainer programmes enabling capacity building at national level,
758 including under-represented countries.
- 759 • Central knowledge hub enabling mapping and access to existing expertise,
760 resulting in improved knowledge transfer and forging new collaborations.
761

762 **2.4.4. Specific Outcomes**

- 763 • A new generation of researchers trained in transdisciplinary, patient-centric RD
764 research interconnected with clinical care.
- 765 • The EU equity among countries for RD capacity building is increased.
- 766 • National/regional training and education programmes are aligned with European
767 standards.
- 768 • Increased awareness of RD stakeholders of the needs of translational and clinical
769 RD research.
- 770 • The EU RD capacity building is increased.
771
772

773 **2.5. Specific Objective 5: Integrated multinational and multi- 774 stakeholder Research & Innovation ecosystem for RD**

775 **2.5.1. Challenge**

776 In the field of rare disease research (e.g., RD diagnostics, therapeutic development,
777 trial readiness networks) cross-national, cross-disciplinary, cross-sectoral and multi-
778 stakeholder collaboration lays the ground for scientific and technological progress
779 that translate in innovative and relevant research results and improvements of care.
780 However, the opportunities for integrating the different national, European and
781 international collaboration in the diverse areas along the healthcare pathway have
782 not been fully harnessed yet. The challenge can be divided around four main axes:
783 (1) Multi-stakeholder collaborations that still suffer from insufficient number of effective
784 public-public and public-private collaborations that are translated towards
785 application, due to lack of trust to open every tool to the most effective type of
786 collaboration, backed by lack of awareness of needs of other actors in R&I value
787 chain and persisting gaps in the funding pipeline. This includes also lack of a structured
788 and continuous dialogue among regulatory agencies, payers and developers on
789 common challenges.
790 (2) National-EU-international alignment, especially operative integration of national
791 capacities as part of a multinational ecosystem. This involves lack of suitable
792 governance models and federated solutions enabling data access/visiting across
793 different data sources in different countries or of sustainable models for the collection
794 of RWE and data on burden of disease (including societal costs), closely linked to the

795 Specific Objective 2; but also insufficiently coordinated policies and R&I funding for RD
796 in multiple countries.

797 (3) Collaboration between existing projects/programmes or initiatives that is subject to
798 fragmentation and duplication of efforts which translates into lack of sustainability and
799 innovation drop rate in EU.

800 (4) Participation and visibility of under-represented countries.

801

802 **2.5.2. Scope**

803 To address the above-mentioned challenges the Partnership will break the silos
804 between communities by consolidating the already existing strong community,
805 currently mostly consisting of public sector researchers, research infrastructures as well
806 as RD patients and representatives, and stepping-up the integration of
807 underrepresented perspectives, namely the industry, regulatory bodies and payers.
808 This will be reflected by relevant governance and advisory structures but also overall
809 Partnership organisation to ensure coherence and maximise impact of all actions.
810 Contribution to RD Moonshot objectives will be essential. Furthermore, through
811 dedicated onboarding mechanisms, the European RD Partnership will gradually bring
812 in additional players to attract and increase the critical mass of resources, know how,
813 talents and excellence, but also to erase white spots on the RD research map and
814 offer equal opportunities to patients across Europe and beyond. The integration of the
815 Scientific Secretariat of IRDiRC will be key to provide strong links to international
816 collaborators as well as a joint management of research and innovation strategy. This
817 will be particularly relevant to drive and support the participation of members from the
818 US National Institutes of Health who are also members of the IRDiRC Consortium
819 Assembly and participate in its activities. These interactions will stimulate the European
820 added value in the field of international collaboration to advance faster toward the
821 vision and goals defined by IRDiRC.

822 The proposed European RD Partnership will also catalyse the transfer of good practices
823 to the national and regional level, including leveraging the power of
824 national/European resources, making them discoverable and actionable for
825 international RD research. In this regard, the role of **National Mirror Groups** will be
826 extremely important to ensure meaningful collaboration with and between countries,
827 since they will bring together the national representatives of the European RD
828 Partnership and other relevant RD stakeholders.

829

830 By default, the Partnership will build on previous and currently operating actions in the
831 RD field such as EJP RD, Solve-RD, ERICA, 1+MG, EHDS or the forthcoming JA on ERNs
832 to help leverage the existing capacities. It will also ensure close alignment and (when
833 possible) joint activities with other Horizon Europe partnerships (e.g., IHI, EIT Health,
834 Innovative SMEs, ERA4Health and Partnerships on Personalised Medicine and
835 Healthcare Systems) as specified in the Synergies with other initiatives section.

836

837 **2.5.3. Potential Outputs**

838 • Structured and enabling environment for multistakeholder and multinational
839 governance and consultation upstream (researchers, industry, patients,
840 regulators), to define common and concerted objectives, considering the
841 constraints of each and aligned with the needs of patients.

842 • RDP used as multistakeholder platform for dialogue to support technical questions,
843 but also social challenges and policy debates linked to RD research (drug
844 regulation, diagnostics, medical devices).

- 845 • By end of the Partnership all partner countries have an active National Mirror Group
846 supporting alignment of goals, strategies and shared best practices.
- 847 • Efficient mechanisms to identify, onboard and deploy high value (national)
848 resources, services and tools that are valuable to the RD community.
- 849 • Effective transcontinental collaboration through integration of IRDiRC
850 recommendations, accessibility to European RD Partnership resources and shared
851 research, clinical and development opportunities.
- 852 • Set-up complementarities and synergies with other relevant programmes and
853 initiatives.
- 854 • Integrative solutions and research pipelines for RD subareas such as diagnosis or
855 trial readiness that integrate and leverage the existing European and national RD
856 research actions.
- 857 • Structural involvement of regulatory bodies (medicines, diagnostics,
858 reimbursement agencies) in all actions involving research.
- 859 • Enable novel collaborations between funders, regulators, payers, and other sectors
860 through provision of frameworks and models for multi-stakeholder collaboration.
- 861 • Improved trial readiness of clinical research sites.

862

863 **2.5.4. Specific Outcomes**

- 864 • RD patient benefits from research results that were enabled through the multi-
865 national and multi-stakeholder Research & Innovation ecosystem for RD.
- 866 • National resources and capacities are supported, optimised and fully integrated in
867 the overall RD ecosystem and their use maximised for the benefit of people living
868 with rare diseases.
- 869 • Sustainable national RD research strategies, aligned with and benefiting from EU
870 and international collaborations in all participating countries.
- 871 • Successful implementation of transcontinental collaboration.
- 872 • Improved coordination of EU initiatives and enhanced EU leadership in RD field.

873

874

875 **3. Performance Indicators**

876 **Under development**

877 *These Performance indicators are designed to measure the outputs, the outcomes of*
878 *the European Rare Diseases Partnership Objectives (General Objectives and Specific*
879 *Objectives). This is work in progress; the final KPIs may be different from what is listed*
880 *below.*

881

882 **General Objective 1** "DIAGNOSIS ESTABLISHED OR ENROLLMENT IN SYSTEMATIC
883 RESEARCH IN AVERAGE WITHIN 6 MONTHS AFTER COMING TO MEDICAL ATTENTION":

- 884 • Number of undiagnosed patients who receive a confirmed diagnosis or
885 enrolled in systematic research within 6 months after first medical examination
886 at secondary care level, facilitated by the partnership.

- 887 • Number of improvements on the time to diagnose patients seeking medical
888 attention for an unknown condition.
- 889 • Number of improvements (efficiency, quality) in all steps underlying diagnosis,
890 from gains in fundamental research (e.g., biomarkers) to the clinical journey of
891 a patient.
- 892 • Number of countries having undiagnosed programmes/activities.
- 893 • Best practices developed within diagnosis-translational pipelines disseminated
894 or adopted or implemented, by diagnostic centres.
895
896

897 **General Objective 2** "1000 NEW THERAPIES FOR RARE DISEASES APPROVED":

- 898 • Number of new therapies approved for rare diseases per year.
- 899 • Number of clinical trials conducted for new therapies for rare diseases
- 900 • Number of partnerships between industry, academia, and government to
901 develop new therapies for rare diseases
- 902 • Time to approval for new therapies for rare diseases
903
904

905 **General Objective 3** "BETTER UNDERSTANDING OF THE IMPACT OF RD ON PATIENTS,
906 FAMILIES AND SOCIETY":

- 907 • Number of publications and presentations on the impact of rare diseases in
908 scientific conferences, policy briefings, and media outlets
- 909 • Number of research studies conducted on the impact of rare diseases on
910 patients, families, and society
- 911 • Number of policy changes or initiatives at local, national, and international
912 levels aimed at addressing the impact of rare diseases on patients, families, and
913 society
- 914 • Number of collaborations between patient groups, academic researchers,
915 industry, and government to address the impact of rare diseases
- 916 • Increase in funding for research on the impact of rare diseases on patients,
917 families, and society
918
919

920 **Specific Objective 1** "Generation of knowledge and its translation into medical and
921 holistic intervention":

- 922 • Number of funded RD projects
- 923 • Number of transitions from one phase in the value chain to the next
- 924 • Number of collaborations between research institutions and healthcare
925 providers
- 926 • Number of publications resulting from RD research projects supported by the
927 Partnership
- 928 • Number of RD projects funded using secondary use of clinical data

- 929 • Number of collaborations between academic researchers, industry, and
930 patient advocacy organizations to develop and implement medical and
931 holistic interventions for RD
- 932 • Funding for RD research and development of medical and holistic interventions
933 within the Partnership (per year)
- 934 • Number of research projects initiated within the Partnership in collaboration
935 with patients / patient organisations
- 936 • Number of RD research projects supported by the Partnership (or a previous co-
937 fund on Rare Diseases) resulting in drugs approved by EMA/FDA, patents and
938 new companies
- 939 • Number of new RD cases with a diagnosis facilitated by the Partnership
- 940 • Number of businesses spinning off and/or direct benefiting from funded project
941 results

942
943

944 **Specific Objective 2** "Healthcare and research data are accessible, and used, for
945 scientific and regulatory evaluation and healthcare delivery":

- 946 • Number of researchers, patients, and clinicians who are re-using and sharing
947 rare disease data to implement multinational research, as evidenced by
948 published research papers, patents, and collaboration agreements.
- 949 • Number of healthcare and research data sources that are made available for
950 scientific and regulatory evaluation and healthcare delivery
- 951 • Number of cases where healthcare and research data use led to a clinically or
952 biomedically relevant outcome (interventions, diagnosing an undiagnosed
953 case, new biomarker, new candidate drug for repurposing)'
- 954 • Number of validated Patient-Centred Outcome Measures (PCOMs) included in
955 the comprehensive data infrastructure based on FAIR principles.
- 956 • Number of clinical trials that are initiated or have progressed due to improved
957 trial readiness and therapeutic options through FAIR data use

958
959

960 **Specific Objective 3** "All activities empower, as equal partners, people living with rare
961 disease":

- 962 • Number of patients empowered, within the Partnership, through capacity-
963 building and training activities related to research.
- 964 • Number of funded research projects that involve patients/patient organisations
965 as co-designers.
- 966 • Percentage of research studies that have involved patient representatives in
967 their governance and decision-making structures.
- 968 • Number of guidelines developed with patients or patient organisations (when
969 they exist) to support equitable patient inclusion in research, and their adoption
970 by relevant stakeholders.
- 971 • Adoption of patient-informed decision-making processes for prioritizing
972 research questions and agendas.

973 • Number/percentage of research questions/interventions that have been
974 informed by patient needs and preferences

975 • Percentage increase in the number of people living with rare diseases who are
976 trained to become advocates and participate in advocacy and awareness-
977 raising campaigns.

978
979 **Specific Objective 4** "Increased capacity and skills of RD stakeholders to optimise
980 research to healthcare continuum":

981 • Number of national/regional training and education programs aligned with
982 RDP

983 • Number of RD stakeholders who participate in training programs to enhance
984 their research skills and capacity

985 • Number of train-the-trainer programmes implemented for capacity building at
986 national level, including under-represented countries

987 • Number of researchers from under-represented countries who have
988 participated in education/training programmes

989 • Number of transdisciplinary research training programs developed and
990 implemented at the European level.

991
992
993 **Specific Objective 5** "Integrated multinational and multi-stakeholder Research &
994 Innovation ecosystem for RD":

995 • Number of countries with sustainable national RD research strategies aligned
996 with EU and international collaborations.

997 • Number of active National Mirror Groups by the end of the Partnership.

998 • Increase in the number of funding programs and initiatives dedicated to RD
999 research and innovation, at both national and European levels. (indicating
1000 improved political commitment and public awareness of the RD challenge)

1001 • Number of complementarities and synergies established with other relevant
1002 programmes and initiatives.

1003 • Increase in the number of clinical trials conducted in multiple countries.
1004 (indicating improved harmonization of regulatory frameworks and ethical
1005 standards, and increased cross-border collaboration among researchers and
1006 institutions)

1007

1008 **4. Conclusions**

1009 Under development

1010

1011

5. Annexes

1012

5.1. Annex 1 - European Partnerships, EU Missions, EU Programmes, Projects and Organisations of potential relevance

1013

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<p>ERA4Health <i>[EU Partnership _Health Cluster]</i></p>	<p>The partnership aims to establish and implement a strategic research agenda and joint funding strategy between major European public funders to advance health research and develop innovation. As well as to develop new approaches that overcome known challenges in multinational clinical research. This will be achieved in close collaboration with ongoing initiatives to support the conduct of multinational non-commercial studies. This would lead to establishing appropriate mechanism(s) for identifying topics and funding sources, and for launching (joint) calls for large, multinational Investigator Initiated Clinical Studies on various health interventions addressing important public health needs.</p>	<ul style="list-style-type: none"> • The model for establishment and financing of multinational clinical trials. • Possible joint funding activities on transversal topics
<p>Innovative Health Initiative (IHI) <i>[EU Partnership _Health Cluster]</i></p>	<p>A collaborative platform bringing the several industry sectors (pharmaceuticals including vaccines, diagnostics, medical devices, imaging and digital sectors) together with academic partners for precompetitive research and innovation in areas of unmet public health need, to accelerate the development and uptake of people-centred health care innovations. Since some projects under the Innovative Medicines Initiative (IMI), predecessor of IHI, are still running / will deliver a legacy useful for the RD Partnership, synergies will be sought with them too.</p>	<ul style="list-style-type: none"> • Joint activity on Accelerator Hub • Alignment with IHI projects related to RD or relevant platforms (e.g., clinical trials, use of data, regulatory aspects)

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
Personalised Medicine <i>[EU Partnership _ Health Cluster]</i>	To align national research strategies, promote excellence, reinforce the competitiveness of European players in Personalised Medicine and enhance the collaboration with non-EU countries.	<ul style="list-style-type: none"> • Data infrastructure • Possible joint calls • Personalised treatment approaches
Transforming Health and Care Systems (THCS) <i>[EU Partnership _ Health Cluster]</i>	Improving health and care models in an ageing, data-driven and digital society, shifting to holistic health promotion and person-centred care approaches through health policy and health systems research (including guidance on how to transform health systems; developing new solutions for health and care; strengthening innovation and its successful transfer to health care systems).	<ul style="list-style-type: none"> • Innovative solutions and their integration in healthcare systems • Models for research to healthcare pathway
Artificial Intelligence, data and robotics <i>[EU Partnership _ Digital, Industry and Space Cluster Cluster]</i>	The partnership on AI will help structuring the European AI community, develop a strategic research agenda and federate efforts around a topic that holds great potential to benefit our society and economy.	<ul style="list-style-type: none"> • Optimisation of data use through AI technologies (e.g., diagnostics)
High Performance Computing <i>[EU Partnership _ Digital, Industry and Space Cluster Cluster]</i>	The EuroHPC will establish an integrated world-class supercomputing and data infrastructure and support a highly competitive and innovative HPC and Big Data ecosystem.	<ul style="list-style-type: none"> • Optimising RD data infrastructures

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
Innovative SMEs <i>[EU Partnership _ Other Partnerships (across other themes)]</i>	The initiative aims to support the transnational market- oriented research projects initiated and driven by innovative SMEs. Innovative SMEs shall take the lead and exploit commercially the project results, thus improving their competitive position. Research organisations, universities, other SMEs, large companies and other actors of the innovation chain can also participate.	<ul style="list-style-type: none"> • Joint funding models • Public-private collaboration (Proof of Concepts for RDs) • Optimisation of support for innovative SMEs in the space of RDs
European Institute of Innovation & Technology Health (EIT Health) <i>[EU Partnership _ Other Partnerships (across other themes)]</i>	Backed by the European Union EIT Health will be delivering solutions to enable European citizens to live longer, healthier lives by promoting innovation, improving health care for citizens and strengthen the health economy in Europe.	<ul style="list-style-type: none"> • Joint training activities • Accelerator hub
European Open Science Cloud (EOSC) <i>[EU Partnership _ Other Partnerships (across other themes)]</i>	The co-programmed partnership aims to improve the storing, sharing and especially the combining and reusing of research data across borders and scientific disciplines. The Partnership brings together institutional, national and European initiatives and engages all relevant stakeholders to co-design and deploy a European Research Data Commons where data are Findable, Accessible, Interoperable, Reusable (FAIR).	<ul style="list-style-type: none"> • Optimisation and integration of RD data infrastructure • Expansion of data sources for the benefit of RDs
<u>EU Mission: Cancer</u>	New initiative rooted in Horizon Europe's research and innovation programme to improve the lives of more than 3 million people by 2030 through prevention, cures, and for those affected by cancer and their families, to live longer and better with 4 key objectives: understand cancer and its risk factors; Prevent what is preventable; Optimise diagnostics and treatments; Support the quality of life of people.	<ul style="list-style-type: none"> • Innovative and holistic research to healthcare pathway models • Possible joint activities (including funding) fostering rare cancers

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<u>Digital Europe Programme</u> [EU Programme]	A new EU funding programme focused on bringing digital technology to businesses, citizens and public administrations.	<ul style="list-style-type: none"> • Digital tools for the benefit of RD community (diagnosis, RWE, PCOMs, etc.)
<u>European Innovation Council – (EIC)</u> [EU Programme]	It aims to identify and support breakthrough technologies and game changing innovations to create new markets and scale up internationally.	<ul style="list-style-type: none"> • Accelerator hub
<u>EU4Health</u> [EU Programme]	<p>EU programme of €5.3 billion complementing EU countries' policies with four main goals: 1) to improve and foster health in the EU, 2) to tackle cross-border health threats, 3) to improve medicinal products, medical devices, and crisis-relevant products, 4) to strengthen health systems, their resilience and resource efficiency. Under these 4 general goals, 10 specific objectives are pursued and several of them are relevant for the RD Partnership for example:</p> <ul style="list-style-type: none"> • Action grants for developing a pilot project for an EU infrastructure ecosystem for the secondary use of health data for research, policy-making and regulatory purposes. • Action grants supporting training activities, implementation, and best practices. • Action grants to organise and collect data to understand the safety, quality and efficacy of therapies applied in the field of assisted reproduction and based on haematopoietic stem cells. 	<ul style="list-style-type: none"> • Maximized alignment of funding and activities supporting healthcare (especially ERNs)

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<u>European Regional Development Fund (ERDF)</u> [EU Programme]	It aims to strengthen economic, social and territorial cohesion in the European Union by correcting imbalances between its regions. It will enable investments in a smarter, greener, more connected and more social Europe that is closer to its citizens.	<ul style="list-style-type: none"> • Use of structural funds to support research funding and Clinical Research Network (including facilities/ infrastructure)
<u>European Social Fund Plus (ESF+)</u> [EU Programme]	The main EU instrument for investing in people and supporting the implementation of the <u>European Pillar of Social Rights</u> . With a budget of almost EUR 99.3 billion for the period 2021-2027, the ESF+ will continue to provide an important contribution to the EU's employment, social, education and skills policies, including structural reforms in these areas.	<ul style="list-style-type: none"> • Use of structural funds to support research funding and Clinical Research Network (including facilities/ infrastructure)
<u>Horizon Europe</u> [EU Programme]	The EU's key funding programme for research and innovation with a budget of €95.5 billion. The programme facilitates collaboration and strengthens the impact of research and innovation in developing, supporting and implementing EU policies while tackling global challenges. It supports creating and better dispersing of excellent knowledge and technologies.	<ul style="list-style-type: none"> • RD knowledge hub (sharing of competences and outputs generated by HE funded projects) • Complementary funding
<u>InvestEU</u> [EU Programme]	It will provide the EU with crucial long-term funding by leveraging substantial private and public funds in support of a sustainable recovery. It will also help mobilise private investments for the EU's policy priorities, such as the European Green Deal and the digital transition. The programme consists of three components: the <u>InvestEU Fund</u> , the InvestEU Advisory Hub, and the <u>InvestEU Portal</u> . The InvestEU Fund will be implemented through financial partners who will invest in projects using the EU budget guarantee of €26.2 billion. The entire budgetary guarantee will back the investment projects of the implementing partners, increase their risk-bearing capacity and thus mobilise at least €372 billion in additional investment.	<ul style="list-style-type: none"> • RD knowledge hub (sharing of competences and outputs generated by HE funded projects) • Complementary funding • Accelerator hub

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<p><u>Accelerating research & development for advanced therapies (ARDAT)</u> <u>(IMI project, 2020-2025)</u> [Project or Organisation]</p>	<p>IMI project which aims at delivering the knowledge, tools and standards needed to speed up the development of Advanced Therapy Medicinal Products (ATMPs).</p>	<ul style="list-style-type: none"> • Outputs to be integrated into the CRN research strategies
<p><u>conect4children - Collaborative network for European clinical trials for children (c4c)</u> <u>(IMI project (2018-2024) that will be replaced by a sustainable legal entity from 2023)</u> [Project or Organisation]</p>	<p>Large collaborative European network that aims to facilitate the development of new drugs and other therapies for the entire paediatric population. It is builds capacity for the implementation of multinational paediatric clinical trials whilst ensuring the needs of babies, children, young people and their families are met. It is committed to meeting the needs of paediatric patients thanks to a novel collaboration between the academic and the private sectors.</p> <p>c4c endeavours to provide a sustainable, integrated platform for the efficient and swift delivery of high-quality clinical trials in children and young people across all conditions and phases of the drug development process.</p>	<ul style="list-style-type: none"> • Contribution to CRN

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<p><u>The Rare Disease Cures Accelerator-Data and Analytics Platform (RDCA-DAP®)</u> [Project or Organisation]</p>	<p>An FDA-funded initiative that provides a centralized and standardized infrastructure to support and accelerate rare disease characterization, with the goal of accelerating therapy development across rare diseases.</p> <p>RDCA-DAP promotes the sharing of existing patient-level data and encourages the standardization of new data collection. By integrating such data in a regulatory-grade format suitable for analytics, RDCA-DAP accelerates the understanding of disease progression (including sources of variability to optimize the characterization of subpopulations), clinical outcome measures and biomarkers, and facilitates the development of mathematical models of disease and innovative clinical trial designs.</p>	<ul style="list-style-type: none"> • Alignment/contribution to RD data infrastructure
<p><u>Data Analysis and Real World Interrogation Network (DARWIN EU)</u> [Project or Organisation]</p>	<p>EMA coordination centre to provide timely and reliable evidence on the use, safety and effectiveness of medicines for human use, from real world health care databases across the EU</p>	<ul style="list-style-type: none"> • Optimisation of RD data infrastructure, especially generation and use of RWE
<p><u>European Genomic Data Infrastructure</u> [Project or Organisation]</p>	<p>The Genomic Data Infrastructure (GDI) project is enabling access to genomic and related phenotypic and clinical data across Europe. It is doing this by establishing a federated, sustainable and secure infrastructure to access the data. It builds on the outputs of the Beyond 1 Million Genomes (B1MG) project and is realising the ambition of the 1+Million Genomes (1+MG) initiative.</p>	<ul style="list-style-type: none"> • Alignment/integration with RD data infrastructure • Re-use of genomic data for diagnosis

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<p><u>European Health Data & Evidence Network (EHDEN)</u> [Project or Organisation]</p>	<p>IMI project that aims to build a large-scale federated network of data sources standardised to a Common Data Model.</p>	<ul style="list-style-type: none"> • Optimisation of RD data infrastructure
<ul style="list-style-type: none"> • <u>European Health Data Space (EHDS)</u> [Project or Organisation] 	<ul style="list-style-type: none"> • Initiative by the EC to promote better exchange and access to different types of health data, to support health care delivery, health research and health policy making purposes. 	<ul style="list-style-type: none"> • Alignment and integration of RD data infrastructure as part of the EHDS
<ul style="list-style-type: none"> • <u>European Platform on Rare Disease Registration (EU RD Platform)</u> [Project or Organisation] 	<ul style="list-style-type: none"> • To cope with the fragmentation of RD patients' data contained in hundreds of registries across Europe. • To act as a knowledge generation centre benefiting healthcare providers including European Reference Networks, researchers, patients, and policy makers in the common effort to improve diagnosis and treatment for patients living with a rare disease. 	<ul style="list-style-type: none"> • Alignment/ integration with RD data infrastructure • Optimisation of the ERN registries
<ul style="list-style-type: none"> • <u>ERICA (Coordination and Support Action under Horizon Europe, 2021-2025)</u> [Project or Organisation] 	<ul style="list-style-type: none"> • Builds on the strength of the individual ERNs and create a platform that integrates all ERNs research and innovation capacity. 	<ul style="list-style-type: none"> • Strategic alignment to optimise ERNs research activities

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<u>GenoMed4ALL</u> [Project or Organisation]	The European initiative to transform the response to Haematological Diseases by seizing the power of Artificial Intelligence, pooling genomic/ '-omics' health data through a secure and trustworthy Federated Learning platform. This stakeholder-driven and self-governed initiative aims to support implementation of the <u>FAIR data principles</u> via Global and Open FAIR implementation networks.	<ul style="list-style-type: none"> • Strategic alignment with RD data infrastructure • Support of FAIR services
<u>Global Alliance for Genomics and Health (GA4GH)</u> [Project or Organisation]	The Global Alliance for Genomics and Health fosters common technical standards, seeking to enable responsible genomic data sharing within a human rights framework.	<ul style="list-style-type: none"> • Two-way alignment for data standards
<u>Gaia-X</u> [Project or Organisation]	Gaia-X represents the next generation of data infrastructure: an open, transparent and secure digital ecosystem, where data and services can be made available, collated and shared in an environment of trust.	<ul style="list-style-type: none"> • Optimisation of the whole RD Partnership structural models and processes
<u>Orphanet Data for rare Diseases (OD4RD) – Direct Grant</u> [Project or Organisation]	Contribute to standardized RD data generation by the maintenance and implementation of ORPHAcodes in Health Care Providers hosting ERNs, RD codification best practices, assistance and tools optimising data for primary and secondary use	<ul style="list-style-type: none"> • Alignment/integration with CRN activities
<u>Patient Focused Medicine Development (PFMD)</u> [Project or Organisation]	Not-for-profit collaborative initiative benefiting patients and health stakeholders by designing a patient-centred health care system with patients and all stakeholders.	<ul style="list-style-type: none"> • Contribution to CRN activities

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<p><u>Rare Disease Moonshot</u> <i>[Project or Organisation]</i></p>	<p>A coalition of public and private partners joining forces to accelerate scientific discovery and drug development in rare and paediatric diseases for which currently there is no therapeutic option. By fostering greater collaboration and improving the sharing of data and knowledge, they aim to accelerate clinical development of new solutions for adults and children living with rare conditions by developing novel clinical trials designs, enhancing data infrastructures and trial networks and defining processes adapted to very small populations.</p>	<ul style="list-style-type: none"> • Strategic alignment • Public-private partnerships
<p><u>Screen4care: Shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies (IMI project, 2021-2026)</u> <i>[Project or Organisation]</i></p>	<p>IMI project that aims at shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies</p>	<ul style="list-style-type: none"> • Integration of outputs into the diagnostic pathway models of CRN
<p><u>Together4RD</u> <i>[Project or Organisation]</i></p>	<p>A multi-stakeholder alliance supporting ERNs to collaborate with stakeholders, particularly with the pharmaceutical industry, to pursue opportunities that will address unmet medical needs of people living with rare diseases, in areas such as basic to translational research, clinical trials for rare & ultra-rare conditions, testing and accelerating innovative approaches to diagnosis, development and implementation of data/evidence generation initiatives.</p>	<ul style="list-style-type: none"> • Strategic alignment for public-private collaboration with ERNs

Initiative [Type of initiative]	Objectives	Pre-identified synergies (non-exhaustive)
<p><u>Towards the European Health Data Space - Joint Action (TEHDAS JA)</u> <i>[Project or Organisation]</i></p>	<p>TEHDAS JA, funded under the EU Health Programme, helps EU MS and the EC to develop and promote concepts for the secondary use of health data to benefit public health and health research and innovation in Europe. It aims at enabling European citizens, communities and companies to benefit from secure and seamless access to health data regardless of where it is stored</p>	<ul style="list-style-type: none"> • Use of outputs to improve RD data (use and reuse) models
<p><u>X-eHealth</u> <i>[Project or Organisation]</i></p>	<p>EU-funded project that aims at developing the basis for a workable, interoperable, secure and cross border Electronic Health Record exchange Format in order to lay the foundation for the advance of eHealth sector.</p>	<ul style="list-style-type: none"> • Alignment with CRN activities

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5.2. List of abbreviations

AI	Artificial Intelligence
c4c	connect 4 children
CRN	Clinical Research Network
EC	European Commission
EHDS	European Health Data Space
EIC	European Innovation Council
EIT	European Institute of Innovation & Technology
EJP RD	European Joint Programme on Rare Diseases
EOSC	European Open Science Cloud
EOSC	European Open Science Cloud
ePAG	European Patient Advocacy Group
ERDF	European Regional Development Fund
ERICA	European Rare Disease Research Coordination and Support Action
ERN	European Reference Network
ESF+	European Social Fund Plus
EU	European Union
FAIR	Findable Accessible Interoperable Reusable
GO	General Objective
HTA	Health Technology Assessment
IHI	Innovative Health Initiative
IMI	Innovative Medicines Initiatives
IRDiRC	International Rare Diseases Research Consortium
JA	Joint Action
JRC	Joint Research Centre
JTC	Joint Transnational Call
MSCA	Marie Skłodowska-Curie Action
NMG	National Mirror Group
OO	Operational Objective
PCOM	Patient Centred Outcome Measure
PENREP	Patient Engagement in biomedical Research Project
PLWRD	Patient Living With a Rare Disease
PROM	Patient Reported Outcome Measure
PSIP	Partnership Specific Impact Pathway
R&D	Research and Development
R&I	Research & Innovation
RDP	Rare Diseases Partnership
RD	Rare Diseases
RWE	Real-World Evidence
SDG	Sustainable Development Goal
SME	Small and Medium Enterprise
SO	Specific Objective
SRIA	Strategic Research and Innovation Agenda
THCS	Transforming Health and Care System

UN	United Nations
US	United States

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