TOOL 2

Examples of Initiatives which Foster Public-Private Partnerships in Rare Disease and Complementary Areas

Together4RD
RARE DISEASES

Together4RD Toolkit to foster ERN and Industry Collaborations

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Tool 1 outlines some of the reasons why public private collaborations (whether through long terms partnerships, or delivered as shorter-term, specific projects) are so important in rare disease.

A number of initiatives fostering these kinds of interactions, at the cross-disease level, are highlighted below (for examples of more specific case studies, see **Tool 7**). This Tool summarises each at a high level, provides links to relevant resources to explore those PPP opportunities further, and highlights resources, for instance the three sets of recommendations produced by the Rare Diseases Moonshot, which should have a particular applicability to supporting public-private partnerships.

The Innovative Medicines Initiative and Innovative Healthcare Initiative

The Innovative Medicines Initiative was set up in 2008 as a public-private partnership between the European Commission (public funding) and the European pharmaceutical industry (private funding, represented by EFPIA, the European Federation of Pharmaceutical Industries and Associations). The goal was to improve the medicines' development process and make it more efficient, and to ensure that patients will have faster access to better and safer medicines. The first phase of IMI covered the period 2008-14, before evolving into IMI2 which spanned 2014-2020. The total budget of IMI 2 was EUR 3.276 billion. Of this, EUR 1.638 billion (half the budget) was pledged from Horizon 2020, whilst EFPIA companies committed EUR 1.425 billion to the programme (up to EUR 213 million could come from other organisations that wished to contribute to IMI initiatives as Associated Partners, for specific projects).

Across IMI 1 and 2, almost 200 projects were funded, dealing with a broad range of conditions. Some of these were particularly relevant for rare diseases, including:

- ☑ conect4children (see below)
- ☑ ARDAT project (looking at Advanced Therapy Medicinal products or ATMPs, which often target rare conditions)
- ☑ Screen4Care (exploring newborn screening for rare conditions, and how to foster earlier diagnosis from health record data)
- ☑ STOPFOP, which was seeking a cure for Fibrodysplasia ossificans progressiva (FOP)
- ☑ U-PEARL, dedicated to better trial design, specifically exploring platform trials, in four focal areas, one of which was the rare condition neurofibromatosis (including types NF1, NF2 and Schwannomatosis)

At the end of 2021, the IMI became the **Innovative Health Initiative (IHI)**. This programme will last until 2027. The core principles remained the same, but the change in name reflected the recognition that different sectors need to be engaged in addressing life-sciences challenges and that "future breakthroughs in medical science will involve cross-sectoral discoveries, such as medical device / drug combinations or diagnostics based on artificial intelligence."[1] To reflect this broader scope, the private partners now include EFPIA, COCIR, Vaccines Europe, EuropaBio, and MedTech Europe. The total budget for IHI, for the period 2021-2027 is €2.4 billion. €1.2 billion comes from Horizon Europe; €1 billion will come from the IHI industry partners; and €200 million will come from other life science industries or associations that decide to contribute to IHI as contributing partners.

IHI has already supported rare disease-related projects, specifically Realise-D and PaLaDIn, with further rare-disease-related calls expected to follow.

For general resources on IMI and IHI, see below:

- **Booklet providing an overview of the IMI**
- **Short video introducing IMI**
- **Blog on the IMI and its value**
- **IMI to IHI**
- **IMI post relating to rare disease public-private collaborations**

A deeper exploration of IMI and IHI projects fostering public-private partnerships in rare disease

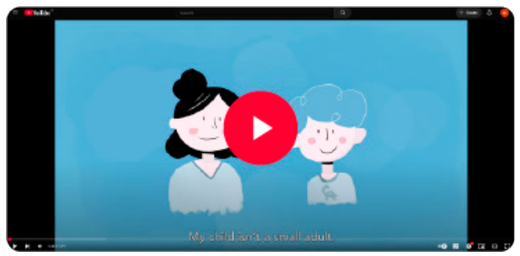
conect4children (c4c)

c4c is an IMI2 project (2018-2024, with extension to 2025) establishing a European network and streamlined ecosystem for clinical trials in paediatric diseases. It involves 36 academic partners, 10 industry partners from EFPIA, and an additional 500 affiliated partners.

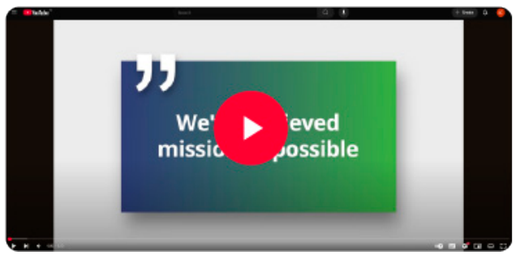
As so many paediatric diseases are also *rare* diseases, c4c's processes and tools to support better, more efficient and more successful clinical trials in children and young people also address broader rare disease needs. Besides developing tools to accelerate study start-up and address the pain points in initiating multinational paediatric trials, c4c included strands of work focusing on key topics such as education, training, PPIE and data standardisation. The achievements and resources of c4c, the public-private partnership, can be found on the **project website** in particular **Connect4Children Achievements**

1 https://www.ihi.europa.eu/about-ihi/imi-ihi

Useful videos : see **What is conect4children?** and **We are proud to conect4children! - The Movie**



What is conect4children?



We are proud to conect4children! - The Movie

I**n 2023, c4c launched a dedicated legal entity, c4c-Stichting**..For details of the c4c-S legal entity, see **Connect4Children** **For a 1-page overview on what the c4c-Stichting can offer to ERNs, specifically, see here**.

Screen4Care

Screen4Care is a €25 million IHI initiative which launched in late 2021. It has two broad and interconnected pillars (see the Screen4Care visual, below):

- genetic newborn screening (exploring the use of genetic testing and related advanced genomic technologies); and
- AI-based tools to bring accurate diagnoses to patients, earlier, via predictive algorithms leveraging the Screen4Care federated data machine learning environment, and algorithms embedded in Electronic Health Record (EHR) systems that will flag patients at risk for rare diseases based on the data *in* their EHR.)

It is a five year project involving 37 partners from 14 countries. For more details, see the **Screen4Care website** and publications **here** and **here**.

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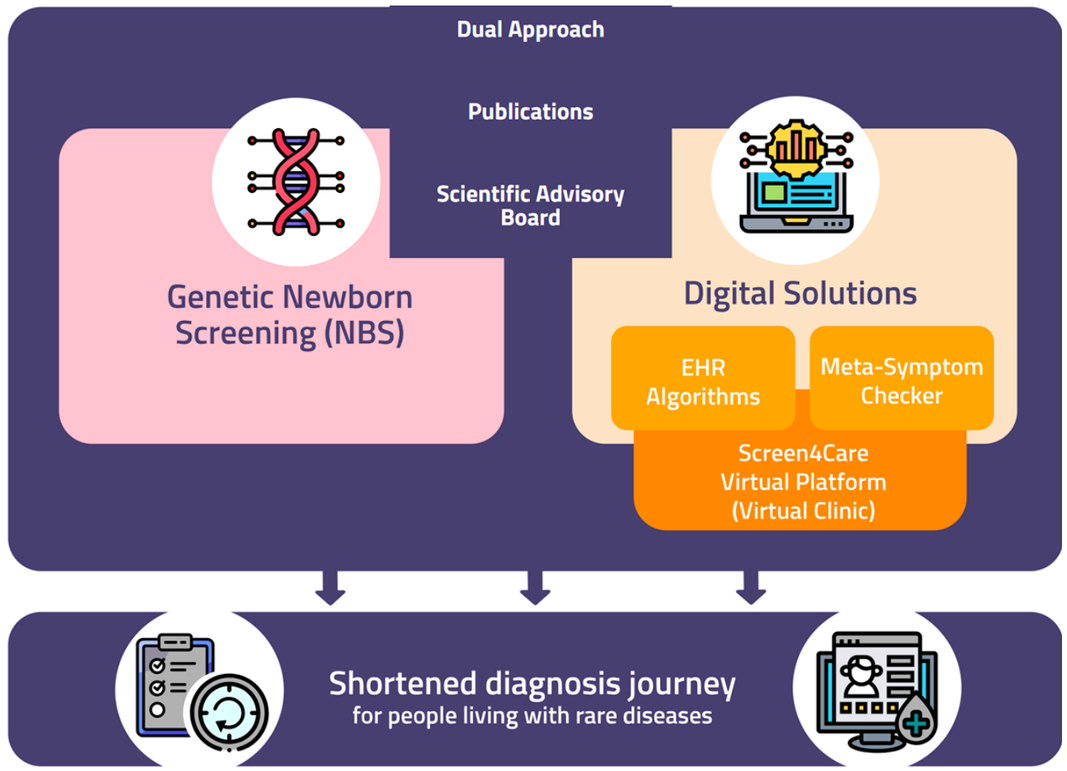

Screen4Care's dual approach (click to view interactive version on our website)

Realise-D

Realise-D stands for 'CompRehensive mEthodological Approach to cLinical trIalS in (ultra-) rarE Diseases'. This 5-year Realise-D public-private partnership began in January 2025, with an overall budget of €17 million. The goal is to optimise and accelerate the development of treatments for rare and ultra-rare conditions, by bringing together 40 partners representing stakeholders from many different groups (clinicians, methodologists, pharmaceutical industry researchers, representatives of patient organisations, regulatory agencies and HTA bodies) to develop cutting-edge operational and methodological tools and resources to dramatically advance treatment evaluation. The Realise-D project has a particular focus on ultra rare conditions and plans to create easy-to-use playbooks and digital tools for planning and running clinical trials.

Read more here.

PaLaDIn

This 4 year project began in 2024, with an overall budget of over €19 million. PaLaDIn is developing a state-of-the-art platform dubbed the 'Interactium' to drive innovative, real-world data collection from patients with rare diseases. The project focuses on rare

neuromuscular diseases (NMDs), specifically Duchenne Muscular Dystrophy (DMD) and facioscapulohumeral muscular dystrophy (FSHD). The Interactium is expected to be able to integrate data from diverse sources, including neuromuscular registries, patient-reported outcome/experience measures (PROMs and PREMs), as well as digital outcome measures from wearable devices, all of which will be co-created with patients. The project is coordinated by a patient organisation and the partners include experts in NMDs, patient advocacy and data science. They hope that their results will not only improve the lives of people with NMDs, but will prove useful to other rare disease communities around the world facing similar challenges.

Read more here.

The Rare Disease Moonshot

The Rare Disease Moonshot was launched in 2022, to bring together a coalition of partners able to accelerate scientific discovery and drug development in rare and paediatric diseases for which currently there is no therapeutic option. This is important, as the majority of rare conditions (approximately 95%) have no dedicated treatment, despite years of investment and research, and there has long been a question of how to shed much needed light and attention on these so-called neglected conditions (which not abandoning research in disease areas which have perhaps seen significant R&D but still lack satisfactory therapies and medicines).

The Moonshot coalition is informal, involving the CriticalPath Institute (C-Path), the European Infrastructure for Translational Medicine(EATRIS), the European Clinical Research Infrastructure Network (ECRIN), the Biobanking and Biomolecular Resources Research Infrastructure – European Research Infrastructure Consortium(BBMRI-ERIC), the European Federation of Pharmaceutical Industries and Associations (EFPIA), the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE), EuropaBio, EURORDIS-Rare Diseases Europe, and the European Joint Programme for RareDiseases (EJP-RD/ERDERA).

These partners set out to both accelerate innovative research for rare conditions in Europe (for instance by developing novel trial designs, enhancing data infrastructure and trial networks, and defining specific approaches for ultra rare diseases), and ensure patients with rare and paediatric disease can *access* the latest innovations. The 'USP' of this approach is its multistakeholder ethos; in particular, the Moonshot partners extol the advantages of leveraging public-private partnerships to help pool resources and solve problems more quickly.

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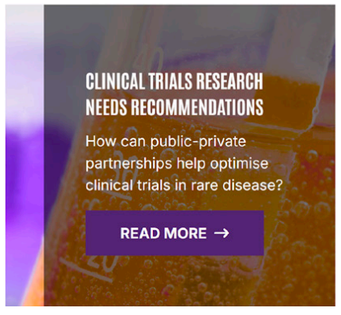
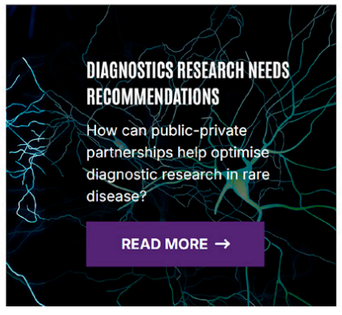
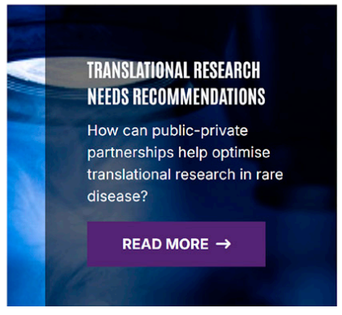
Key Resources from the Rare Disease Moonshot

Three key topics were selected for multistakeholder collaboration, and new sets of recommendations have been created for each. **Although not specific to ERNs, these sets of recommendations are clearly very relevant to the goal of this Toolkit, namely advancing public-private collaboration between ERNs and Industry.**

THE RARE DISEASE MOONSHOT PARTNERS IDENTIFIED THREE AREAS OF ACTION WHERE PUBLIC PRIVATE COLLABORATIONS CAN ADD MOST VALUE:







These sets of recommendations are the fruits of months of broad consultations with stakeholders including

- patient representatives
- global pharma/biotech companies
- small innovative enterprises
- academic translational research experts
- biobanking, non-profit clinical and fundamental research communities
- non-profit PPPs.

Title of the Resource	What can I find here?	Link
Rare Disease Moonshot Recommendations: How can public-private partnerships help optimise clinical trials in rare disease?	Recommendations generated by a group of experts led by C-Path and ECRIN. 33 stakeholders contributed to their development, through workshops and consultations. The recommendations address topics such as disease prevalence, patient access, data standards, and regulatory support.	Clinical trial recommendations
Rare Disease Moonshot Recommendations: How can public-private partnerships help optimise diagnostic research in rare disease?	Work began in June 2023, with a workshop that brought together 30 industry participants. The final recommendations are the results of months of teamwork between the RD Moonshot team and stakeholders including industry, the research community and patient advocacy groups. Further input came from a series of additional workshops with industry partners and discussions in the EURORDIS Round Table of Companies, which gathered more than 100 participants.	Diagnosis recommendations
Rare Disease Moonshot Recommendations: How can public-private partnerships help optimise translational research in rare disease?	Development of this resource began with a workshop in February 2023, bringing together 33 participants from a broad spectrum of sectors. The initial discussions underscored the importance of interdisciplinary approaches, research coordination, and enhancing the skills of different stakeholders. The recommendations were further elaborated and refined via additional workshops, online consultations, and direct feedback sessions, involving over 20 stakeholders.	Translational research recommendations

For more on the mission of the Rare Disease Moonshot and why public-private partnerships are so important, **see here**.

See further on this publication **here**.

Click here to access the Rare Disease Moonshot website.

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The European Rare Disease Research Alliance (ERDERA)

ERDERA launched in September 2024, with an estimated budget of €380 million to support activities up to 2031. The overall goal of this large initiative is to improve the lives of people living with a rare disease in Europe and beyond. Over 170 organisations are involved, across 36 countries, giving ERDERA a global footprint to complement the European. This is a Horizon Europe Partnership, in which the EU is expected to contribute approximately €150 million, with the rest of the funding coming from EU Members States, countries associated to Horizon Europe, and in-cash and in-kind contributions **from public *and private*** partners. ERDERA is, in many ways, a successor to the EJP-RD, as it seeks to 'bring under one roof all knowledge, resources and services, boost clinical research and spur innovation' at the pan-rare-disease level. It also builds heavily on other key research initiatives like ERICA (the ERN-focused research action) and the Horizon 2020-funded diagnostics project Solve-RD.

There are many streams of interconnected activity in ERDERA, but the 3 main missions may be summarised as follows:[2]

- To bring together, in one place, a range of services, resources and cross-disciplinary expertise, in order to bring added-value to rare disease research
- To boost clinical research by ensuing every patient wishing to participate in research is somehow findable, and can be enrolled in a suitable clinical study
- To increase innovation and EU competitiveness, whilst evolving a global ecosystem for rare disease capable of linking the national, regional, European and global levels

ERDERA's activities are structured around four key pillars:

- **Funding** — including dedicated financial support for collaborative international research projects, clinical trials, and knowledge exchange and networking initiatives.
- **Clinical Research Network** — Encompassing all ERDERA's in-house research activities, this network will enhance diagnostics and clinical trial readiness
- **Support Services** — This includes a *Data Services Hub* to facilitate global data collection, integration, analysis, and sharing at a global scale; an *Expertise Services Hub* to offer guidance on specific aspects of translational and clinical research; and an *Acceleration Hub* that collaborates with industry partners to advance the most promising research projects and technologies. Additionally, ERDERA will maintain a robust *Education and Training program*.

2 Much of this summary comes from the ERDERA website

- **International Alignment** — Through existing and newly established National Mirror Groups, the partnership will ensure alignment between national and international rare disease research strategies, particularly in nations that are behind in developing and implementing national plans. ERDERA will also host the Scientific Secretariat of the **International Rare Disease Research Consortium** (IRDiRC), a unique global consortium co-established by the European Commission and US National Institutes of Health back in 2011.

The ERDERA consortium includes private sector companies, including UCB Biopharma SRL, AstraZeneca AB, and Pfizer Inc.[3] ERDERA is expected to foster public-private projects and collaborations through, for instance, the funding activities and calls, and the Acceleration Hub, in particular.

The European Joint Programme for Rare Disease Research (EJP RD)

The EJP RD launched in January 2019, involving 93 beneficiaries and 48 linked third parties, with a total budget of approximately €110 million (€55 million directly from the EC, supplemented with substantial national and in-kind contributions). The EJP RD sought to create resources, services and expertise to advance rare disease research at the cross-disease level, through workstreams centred on funding opportunities, data, training, and accelerating innovation . **The main public-private focus here came from the 'Rare Disease Research (RDR) Challenges Call'**. Industry partners were invited to identify 4 challenges to form the topics of the call, and these were validated by EJP RD Partners. A total budget of € 1.5 million was anticipated, from the European Commission, to allow 4 projects to be funded. The idea was that an independent committee would review the proposals and the industry partners who identified each challenge would then join the successful consortium of applicants, bringing in-cash and in-kind support.

The topics/challenges were as follows:

1. **Development of a non-invasive tool for measuring rare disease patient mobility in daily living** (Industry sponsor - Chiesi Farmaceutici S.p.A. (Italy), CSL Behring (Australia))
2. **Delivery system for intranasal administration of biological drugs to neonates** (Industry sponsor -Chiesi Farmaceutici S.p.A. (Italy))
3. **Characterize Rare Bone Disorders (RBD) Mobility Challenges in Real World Setting** (Industry sponsor - Ipsen)
4. **Pre-clinical assay to detect instability of microsatellite repeat expansions** (Industry sponsor - LoQus23 Therapeutics)

An example of a funded project emerging from this scheme (related to the 1st challenge) is the **Digital Tools 4 Rare Disease (DT4RD) project**.

Find out more here on these challenges **here**.

IRDiRC

The International Rare Disease Research Consortium, IRDiRC, was launched in 2011, initiated by the European Commission and the NIH. It initially had two major goals: to create 200 new therapies for rare diseases and enable diagnostics for most rare disease, both by 2020. However, given the early success in meeting these goals the consortium revised its objectives in 2017. A new overarching vision was agreed, for the period 2017-2027: 'Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention'. To achieve its goals, IRDiRC has undertaken numerous dedicated actions to increase access to harmonized data and samples, enhance the molecular and clinical characterization of rare diseases, support translational, preclinical and clinical research, and streamline ethical and regulatory procedures.

IRDIRC is organised into 3 constituent committees (dedicated to funders, companies, and patient advocates respectively) and 3 scientific committees (Therapeutics, Diagnostics, and Interdisciplinary).

In terms of the opportunity for public private collaboration, companies can be members of IRDiRC, alongside public funders. Biotech, Pharma, MedTech and more can join, by investing more than 10 million USD over 5 years in rare disease research. Much of the work of IRDiRC is shaped by its three committees, which identify gaps and key issues in RD research, to be addressed via Task Forces and Working Groups who produce guidelines, recommendations & resources. These structures often involve both public and private sector experts.

Find out more **here**.

Critical Path Institute (C-Path)

Another public-private partnership of relevance to rare diseases is C-Path. This is a non-profit PPP launched in 2025 to accelerate the pace -and reduce the costs- of medical product development through creation of new data standards, measurement standards, and methods standards that support the scientific evaluation of safety & efficacy of new therapies. C-Path provides the legal, scientific, and regulatory infrastructure to generate

3 https://erdera.org/participants/

a unique neutral collaborative environment for stakeholders in the drug development ecosystem. The Institute fosters public-private collaboration across both rare and non-rare conditions; however, an important -and RD-specific- infrastructure under C-Path is the "**Rare Disease Cures Accelerator – Data and Analytics Platform**," where industry can share data into a centralised and standardised infrastructure to support & accelerate RD characterisation to accelerate dev. of therapies.

Read more about C-Path here.

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