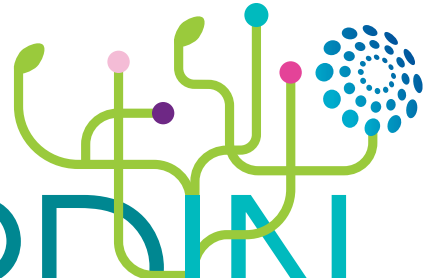




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# JARDIN

INTERNAL NEWSLETTER #2  
JANUARY 2025

## WELCOME

By Till Voigtländer

*Dear partners, colleagues, and friends,*

We are happy to welcome you back to the JARDIN internal newsletter! Not long ago, in the first issue, you had the opportunity to get to know JARDIN from the drafting of the Proposal to the signing of the Grant Agreement, and to journey through the **JARDIN work packages**, its **teams**, and its first **achievements**.

Now that you are more familiar with JARDIN, this second edition of the Newsletter will update you on **the tasks of JARDIN's nine work packages**, highlight the latest **achievements**, and inform you about **upcoming events** and **planned activities**.

Since the commencement of the JARDIN project, Work package teams have been hard at work, and there is much more to come. We are excited about

the ongoing activities in the work packages, grateful for the commitment of all our partners, and thrilled about this journey towards a completely new ERN ecosystem.

Please stay connected with us and keep an eye on JARDIN social media channels. If you have missed the first issue of the newsletter or have any questions, please contact us at [jardin@meduniwien.ac.at](mailto:jardin@meduniwien.ac.at).

The **JARDIN internal newsletter** is written by the **Work Package 1** Coordination Team, designed and disseminated by the **Work Package 2** team, and enriched with inputs and contributions from **all other Work Packages**. It is issued every six months as an official JARDIN deliverable of Work Packages 1 and 2.





## European Reference Networks

### A GLIMPSE OF JARDIN'S LATEST ACCOMPLISHMENTS

*These are the main achievements of JARDIN over the past six months:*

#### Second Amendment of the Grant Agreement

JARDIN's second amendment covers **the following topics:**

- **Work package (WP) 3 co-leadership between the University Hospital Center Zagreb (UHCZ) and the Medical University of Vienna (MUW)**

Due to unforeseen circumstances, the UHCZ team could not carry out its tasks to the extent foreseen in the original Grant Agreement. This issue was presented and discussed at Steering Committee and Management Board meetings and was finally put to an electronic vote by the JARDIN Competent Authorities.

A co-leadership of WP3 between UHCZ and MUW was the proposed and agreed solution to address the problem. In this co-leadership arrangement, UHCZ and MUW will share decision-making responsibilities, as well as the workload and the budget for WP3.

- **Changes in the budget of Cyprus**

Due to national legislation on purchasing and procurement, the Ministry of Health of Cyprus had to reallocate part of its budget for personnel costs in WP8 and WP 9 from category A.2/A.3 to the corresponding category B (subcontracting costs).

- **Changes in the budget of the Netherlands**

After reassessment of the budgetary needs of all Dutch partners and taking into account legal restrictions, the Competent Authority of the Netherlands and its Affiliated Entities agreed to budgetary adjustments including changes to the subcontracting category.

- **Splitting Deliverable D7.1 into two separate reports and adjusting the due date to M12**

The WP co-leads for WP7, Holm Graessner and Ursula Unterberger, decided to split deliverable D7.1 (State-of-play report national reference networks (NRN) / undiagnosed disease programmes (UDP) into two separate reports. The "State-of-play report NRN" will be D7.1, the "State-of-play report UDP" will be added as additional deliverable D7.8. The due date for both reports will be month (M) 12 instead of M11 in the original Grant Agreement.

- **Harmonisation of the order of participants in WP5 tasks (T) in the Description of the Action (DoA) Part B**

Upon request of AOUP, the WP5 overview table now consistently names IOR as first beneficiary in T5.1 to T5.8, where they are task lead. Accordingly, AOUP is at the first position for T5.9 and T5.10.

## Consortium Agreement

The signature of a Consortium Agreement between partners is an obligatory requirement for the majority of EU projects. The European Commission provides general guidance on the drafting of Consortium Agreements but does not endorse a specific model. The JARDIN Consortium Agreement has been prepared by WP1 with the support of the MUW legal department. It is based on the DESCA (Development of a Simplified Consortium Agreement; <https://www.desca-agreement.eu/desca-model-consortium-agreement/>) model and tailored to the requirements of the Joint Action and the provisions of the Grant Agreement.

The initial draft version of the JARDIN Consortium Agreement was sent to the competent authorities in early July 2024. After three rounds of review, the final version was ready for signature in mid-September. The signing process of the JARDIN Consortium Agreement was launched on October 7, 2024, and is still ongoing. Since the original deadline was at the end of October, we ask the remaining two Competent Authorities to sign the document as soon as possible.

## Preparatory Meeting for the First Meeting on JARDIN Sustainability with National Policy Contact Point Group (NPCP) and Hospital Managers Advisory Group (HMAG) Representatives in Brussels on March 19, 2025

A first meeting on strategies and national processes to safeguard results and activities on the member states' level after completion of the project is planned to take place in Brussels on March 19, 2025. A preparatory meeting took place on November 28, 2024, on the premises of the Európa Pont/Europe Point in Budapest, Hungary. The main goal of the preparatory meeting was to discuss JARDIN minimum sustainability requirements, and brainstorm on the Agenda for the upcoming Brussels meeting.

The meeting was attended by 23 participants, including members of the JARDIN Steering Committee, two additional ERN coordinators, and officers from HaDEA and DG SANTE B3, who participated in a fruitful debate.

# 2025 MEETINGS

## First Meeting on JARDIN Sustainability with NPCP and HMAG Representatives

The "First Meeting on JARDIN Sustainability with NPCP and HMAG Representatives" will be held on March 19, 2025, at the HaDEA headquarters in Brussels. Invitations including the provisional Agenda for the meeting were sent out in December. This meeting will consist of two sessions. The first session will be organised by JARDIN and will focus on the sustainability of JARDIN's activities and results after its life cycle. A second session will be organised by DG SANTE B3 to discuss future policy cooperations and will offer a session on Orphanet led by Ana Rath.

## Second JARDIN Annual Meeting

The second JARDIN annual meeting for all consortium members is planned for October 7-10, 2025, in Vilnius, Lithuania. Preparations are underway, in cooperation with a professional event management company. Further details about this meeting will be provided in upcoming JARDIN Management Board meetings, Bullet Point Newsletters, and Internal Newsletter.

# TO DOs COMPETENT AUTHORITIES



## SIGN CONSORTIUM AGREEMENT

As the original deadline for signing the JARDIN Consortium Agreement by the end of October 2024 has long passed, we ask the remaining two competent authorities to sign the Agreement as soon as possible.



## PROVIDE INFORMATION FOR FIRST PERIOD OF INTERNAL FINANCIAL MONITORING

The deadline for completion and submission of the first round of interim financial monitoring was October 24, 2024. We ask partners that did not yet get back to us regarding the reporting period from January to September 2024 to do so as soon as possible.

Please note that a short e-mail stating that you will not declare personnel or subcontracting costs for this period would be sufficient, if applicable to your institution.



## GET IN TOUCH WITH YOUR NATIONAL HEALTH AUTHORITY REGARDING NOMINEES FOR THE MULTI-STAKEHOLDER ADVISORY GROUP

Following a joint invitation sent by JARDIN and DG SANTE B3 in July 2024 and a follow-up e-mail by the JARDIN Coordination in November 2024, WP1 is still receiving nominations from member states for the NPCP, the Data Management Advisory Group (DMAG), and the HMAG.

We would kindly ask JARDIN Competent Authorities from member states that have not provided any or only incomplete nominations to date, to get in touch with their respective Health Authority to expedite the nomination process. If you are unsure whether JARDIN received all nominations from your country, please get in touch with us at:

[jardin@meduniwien.ac.at](mailto:jardin@meduniwien.ac.at)



# WP1

# TASKS

## T1.2: Establish the advisory bodies of the Joint Action

The establishment of the three subgroups of the Multi-stakeholder Advisory Group of JARDIN, the **NPCP**, the **HMAG**, and the **DMAG**, is underway.

Positioned outside the internal management structure of the Joint Action, the Multi-stakeholder Advisory Group, which will be complemented by the Patient Advisory Group in the future, will provide regular advice to the JARDIN Coordination, the Steering Committee, and the Management Board. It will also support the different WPs when their specific expertise and advice is requested by the respective WP leads.

As mentioned above, all member states that have not yet nominated their representatives should submit their nominations as soon as possible.

## T1.3: Monitor the progress of the Joint Action, ensure the quality of the implementation together with the Steering Committee and WP3

WP1 has been continuously monitoring the progress of JARDIN, including reviewing the deliverables to ensure their quality, supported by members of the Steering Committee. In the frame of the risk management strategies foreseen in the Grant Agreement, the unforeseen circumstances that affected the work of WP3 were reported to the Steering Committee, the Management Board, and HaDEA, and appropriate measures were taken.

## T1.4: Ensure communication and information exchange among project participants

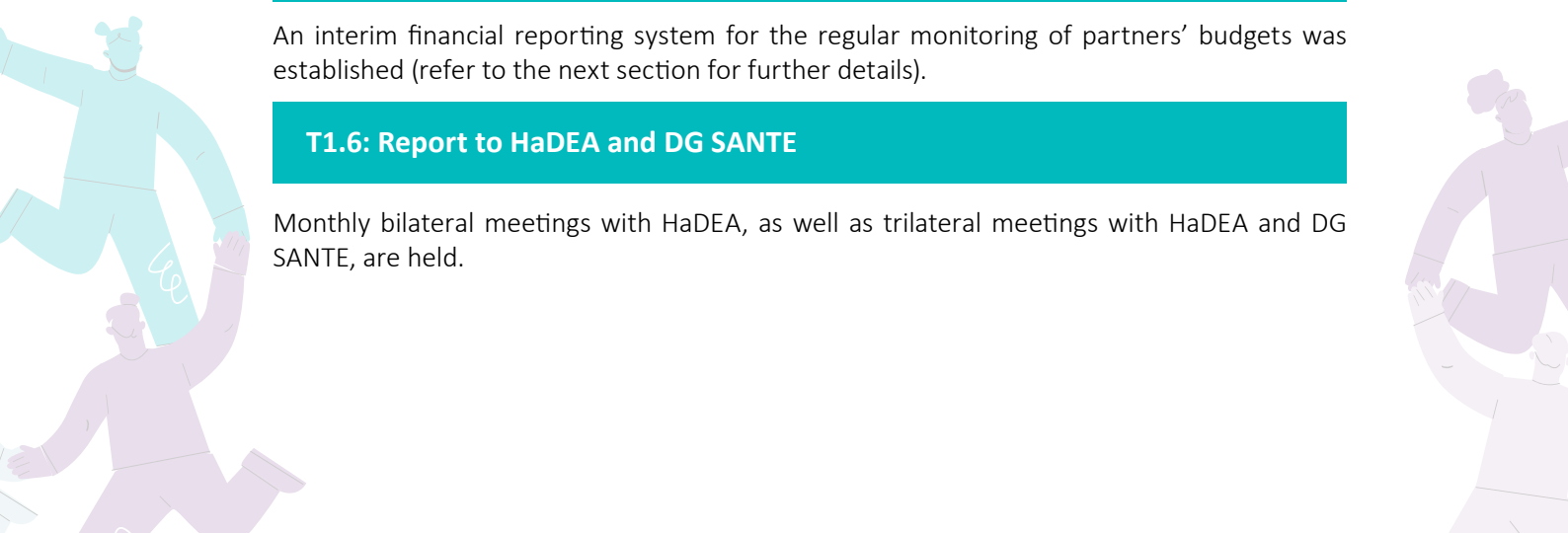
Regular meetings of the Management Board and Steering Committee have been carried out, and teleconferences with project partners have been organised. The Basecamp software is continuously used as a project management tool. The JARDIN Bullet Point Newsletter has been set up as an additional means of communication in between internal newsletters.

## T1.5: Provide day-to-day administrative support, ensure appropriate financial management

An interim financial reporting system for the regular monitoring of partners' budgets was established (refer to the next section for further details).

## T1.6: Report to HaDEA and DG SANTE

Monthly bilateral meetings with HaDEA, as well as trilateral meetings with HaDEA and DG SANTE, are held.



# ACHIEVEMENTS

## Interim Financial Monitoring

JARDIN is a large and complex Joint Action with a budget of €18.75 million. In order to promote transparency and accountability, and to ensure that financial and operational information is well documented and aligned with WP tasks/deliverables, a process for regular monitoring and reporting of the project partners' budgets was implemented.

An easy-to-use Excel form was designed for Competent Authorities and Affiliated Entities to submit their interim financial information, and teleconferences were held to explain the form in detail and assist partners with specific questions.

## Multi-Stakeholder Advisory Group

The establishment of the aforementioned JARDIN Multi-stakeholder Advisory Group is underway. 23 of 28 NPCP members have been nominated, as well as 19 HMAG and 17 DMAG members.

## Bullet Point Newsletter

WP1 launched the "Bullet Point Newsletter" to provide JARDIN participants and stakeholders (including BoMS representatives, patient organisations, healthcare providers, etc) with short but timely updates on ongoing tasks and upcoming activities. This new JARDIN communication channel is a "work in progress", maturing over time with suggestions and constructive feedback from readers.

# WHAT IS NEXT?

## Inaugural (virtual) meetings for NPCP and HMAG representatives

One-hour introductory (virtual) meetings will be held at the end of January and beginning of February 2025 to introduce JARDIN and explain the role and responsibilities of the Multi-stakeholder Advisory Group to all representatives.

## JARDIN Interim Report

The first reporting period of the project will end at M19 (end of July 2025), after which we have 60 days to complete the interim report for HaDEA. The report will comprise information on the work that has been done, as well as financial reporting. Details and instructions will be communicated to JARDIN participants in due course.

# WP2

# TASKS

## T2.2: Define the audience needs, especially patients and clinicians

Thanks to the stakeholder analysis country reports and the surveys completed by communication managers of all the ERNs, the WP2 team was able to define the needs of JARDIN's audience and adapt its communication and dissemination strategies, both within and outside the project, to promote the European Reference Networks (ERNs).

## D2.9: National stakeholder analysis

In addition to the specific dissemination and communication tasks, WP2 has been working on the stakeholder analysis country reports.

The response from countries to contribute to our stakeholder analysis has been excellent, with 20 out of 28 countries submitting their analyses. As the 20 analyses have been carried out according to the needs of the respective countries, the task of WP2 was to bring all these analyses together, to codify the data and to report on the overall results.

Based on these data, WP2 will identify the needs, issues and strategies that should be part of JARDIN communication and dissemination strategies. The tangible results obtained from the performance of all these tasks are presented in the following section as the achievements of WP2.

## T2.3: Key messages, and T2.4: Channels

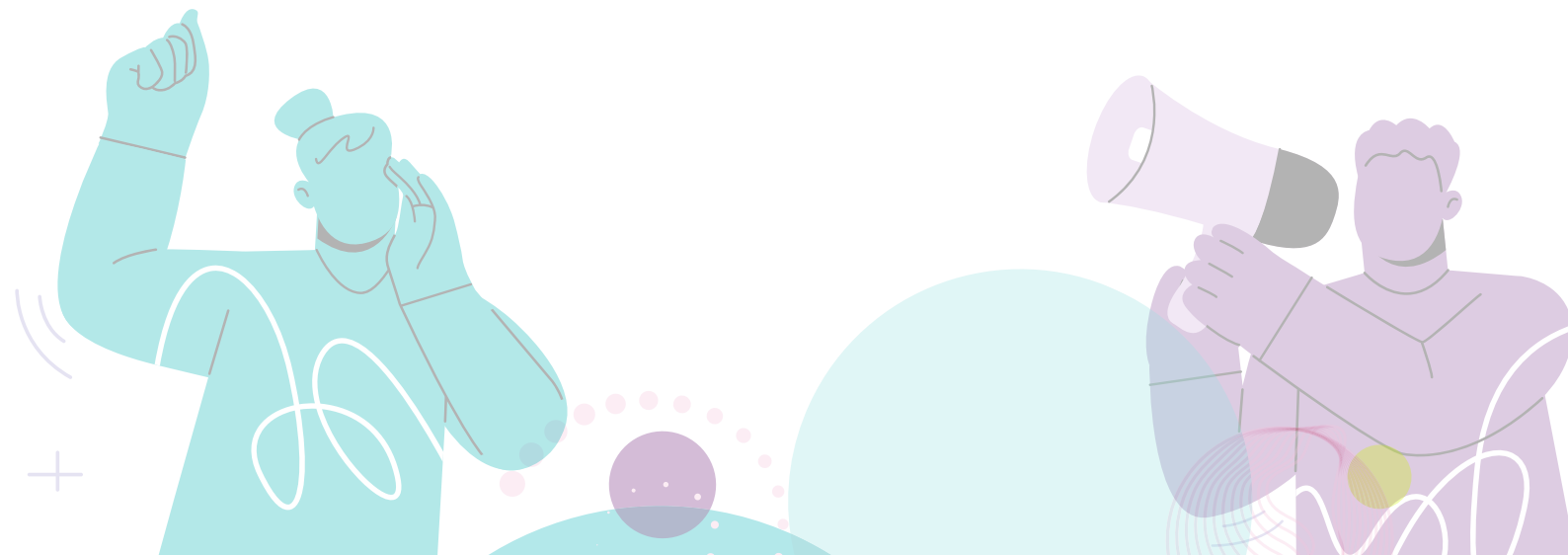
The WP2 team has defined the key messages and established all the communication channels foreseen in the Communication and Dissemination plan.

## T2.5: Dissemination planning and implementation

In terms of dissemination planning and implementation, WP2 translated the objectives proposed in the communication and dissemination plan into concrete actions.

These results can be seen in D2.2, on the JARDIN website and social media channels, and are detailed below in the achievements of WP2.

# WP2



# ACHIEVEMENTS

## WP 2

### The JARDIN website

WP2 has managed to develop and launch the JARDIN website ahead of schedule. The website was launched on July 29, 2024.

The design and development of the website was one of the major achievements of the dissemination work package. WP2 has achieved the objective of having [the core of the communication and dissemination plan](#) fully up and running: the corporate website provides relevant content describing the objectives of the Joint Action, the Work Packages, the partners, the participating countries and —above all— [the ERNs](#), to which a page has been dedicated with information and links to all 24 networks.

[www.jardin-ern.eu](http://www.jardin-ern.eu)

### Social media channels

The WP2 team has created profiles on all [the social networks foreseen in the communication and dissemination plan](#).

In addition to the first social networks created (X and LinkedIn), the team has integrated the other [channels](#) (Facebook, Instagram, and YouTube) to disseminate [key messages](#) and reach all target audiences in different formats: publications linking to our [website](#), dissemination of news and events, animated pills, short videos and reposting of publications from the 24 ERNs and other entities involved in JARDIN.

JARDIN is currently present on the main social networks.

[/jardin\\_EU\\_JA](#)

[/jardin-joint-action](#)

[/JARDINJointAction](#)

[/jardinjointaction/](#)

[/@JARDINJointAction](#)

### External newsletter

As planned, JARDIN's first external newsletter was [successfully distributed on October 30, 2024](#).

External newsletters will be published on the JARDIN website. The first one is already available under this link: <https://jardin-ern.eu/newsletter/>

The first [external newsletter](#) was sent out to 255 subscribers, and it is expected that the number of subscribers will increase, and the newsletter will reach more readers, in the next issue, which will be distributed in April 2025.

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## Communication and dissemination materials

WP2 has created a brand image and identity for JARDIN, as well as numerous visual materials, including [short animated pills](#) and a [video explaining the Spanish healthcare system](#) and its connection to the ERNs. All of these videos can be viewed on the [YouTube channel](#):

[Visit YouTube Channel](#)

JARDIN's visibility is growing, with more followers on social networks (we have reached more than 700 followers on LinkedIn in less than a year), more subscribers to the newsletter and more visits to the website, which continues to be updated with the aim of becoming a reference tool and an information bridge to learn about the operation of the 24 ERNs.

## National stakeholder analysis (D.2.9)

D2.9 is encompassed in T2.2 and it reports the national stakeholder's analysis in 18 countries participating in the Joint Action. The objective of this analysis is to understand their needs in order to adapt the communication strategy of JARDIN at EU and local level. This approach ensures that the resulting communication strategies and messages are well-aligned with the priorities of diverse stakeholders, enhancing engagement and impact.

The report focused on two main audiences: "clinicians" (or healthcare professionals from primary care and non-RD centres) and "patients".

The analysis will serve as a strong basis for [adapting the communication and dissemination strategy of JARDIN at EU and local level](#), and provide an excellent starting point for developing materials for the draft [blueprint pilot](#).

# WHAT IS NEXT?

Thanks to the stakeholder analysis, WP2 will have more data on the [level of knowledge about ERNs](#) among the main target groups healthcare professionals and patients and patient organisations. [These data will help the WP2 team to identify the needs, issues, and strategies](#) to be followed, and will allow WP2 to focus on its communication and dissemination strategy - using JARDIN social media channels and producing its materials in the most appropriate way.

WP2 "Dissemination" will continue to work with the future of JARDIN in mind and its need to gain visibility and raise awareness of the role of ERNs in improving the lives of people living with rare diseases or complex conditions.

WP2 has been working on [updating the website](#) to cover all communication needs in terms of content published on it. [The website](#) is dynamic and constantly updated.

To this end, the WP2 team is developing the local information page for each country, both in English and in the official languages of the country. These pages will be dedicated to health professionals and patients and patient organisations, and will be available online soon. Other sections, such as news and events, will be updated regularly.

To shape the future of JARDIN, we need a strong and involved community. All WPs are the cornerstones of this community, so it is important that the participants in the different WPs follow JARDIN on social media, subscribe to the newsletter, and visit the website to keep up to date with the latest news.

**Do you have something to communicate? Please, write to:**

[communication@jardin-ern.eu](mailto:communication@jardin-ern.eu)

# WP3 TASKS

*As described above, **MUW and UHCZ now co-lead WP3**, distributing some tasks between each other, and sharing the responsibilities for others*

## T3.1: Evaluation plan

The evaluation plan already submitted by MUW and UHCZ will be complemented by a new list of indicators which is developed under the primary responsibility of MUW on the basis of the information collected from WPs' roadmaps. These SMART indicators will be designed to measure alignment with JARDIN's objectives, as well as the progress of WPs and their effectiveness.

## T3.2: Workshop

UHCZ has started to prepare a workshop for WP leads which aims to reflect on the first half of the project, identifying strengths and weaknesses, collecting the overall satisfaction of WP leads with the state of the project after 19 months, and drawing conclusions to adjust the plans and strategies for the second half of JARDIN in order to ensure alignment across WPs and foster a collaborative approach.

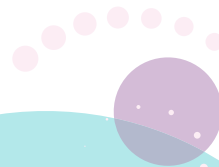
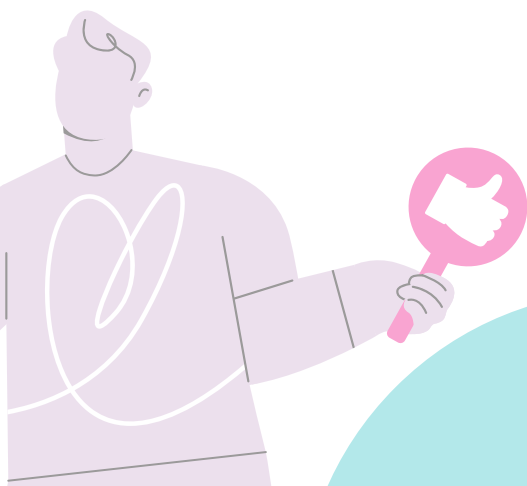
The workshop is planned to take place in June 2025, in Zagreb, Croatia.

## D3.2: Survey 1

UHCZ and MUW are currently finalizing the first annual survey to gather feedback from JARDIN participants. The survey will assess participants' satisfaction with project processes and their quality, with a focus on communication.

# WHAT IS NEXT?

**MS4 – WP Leads' Workshop in June 2025, in Zagreb, Croatia (see above)**



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WP4

# TASKS

## T4.1: Sustainable elements and sustainability mechanisms

Over the last few months, WP4 has been implementing T4.1 – “sustainable elements and sustainability mechanisms”. The task includes: (T4.1.1) the establishment of a National Policy Contact Point Group composed of national authorities with the mandate to support implementation and sustainability of JARDIN’s outcomes and outputs, and (T4.1.2) identification of sustainable elements for 6 WPs (2, 5-9).

**Two sets of sustainable elements** will be identified:

**i) minimum requirements across all member states** (methodology: interviews and workshops with JARDIN’s WP leads and Competent Authorities; consultation with members of the Multi-stakeholder Advisory Group and common agreement through Nominal Group Technique - consensus conference)

**ii) diverse set of solutions across member states and regions** that will enable the implementation and sustainability of the Joint Action actions across 6 WPs beyond the minimal requirements (methodology: interviews, workshops and case studies with the aim of reflecting five groups of member states according to their health system organisation, population/country size and other factors).

## T4.2: Support for better integration of sustainable elements into the National legislation, including National RD Plans/Strategies

For T4.2.1 – “Evaluation of ERN integration aspects in the existing National RD Plans/Strategies”, a rich resource of [Rare Disease National Plans and Strategies](#) (RD NP/NS) and related legal acts and documents have been collected through the digital links that were kindly provided by the member state representatives in the JARDIN survey and through further targeted searches and interviews.

WP4



# ACHIEVEMENTS

## WP4

### WP4 roadmap

During the first months of JARDIN, WP4 has developed a [roadmap](#) detailing each step towards achieving its final objectives. The roadmap is a living document that may change according to the results obtained, but it is essential to monitoring the activities of the WP within the limited timeframe of JARDIN.

### List of core sustainable elements

WP4 has prepared a preliminary list of [core sustainable elements](#), which was agreed with all WP leads (WP2, WP5, WP6, WP7, WP8 and WP9). It is [essential](#) that these core sustainable elements:

- arise from JARDIN activities and have both an impact on the sustainability of ERNs and benefits for member states;
- be feasible, agreeable and implementable across the full range of the EU member states and Norway;
- be supported by JARDIN's resources and activities to ensure a smooth implementation;
- are measurable or monitorable.

The preliminary list of [core sustainable elements](#) has already been discussed at the JARDIN Preparatory Meeting for the First Meeting on JARDIN Sustainability with NPCP and HMAG Representatives, which took place in parallel with the high-level conference "For an EU commitment to tackling rare diseases" organised by the European Economic and Social Committee (EESC) in Budapest in November 2024. Further wider consultations will include all JARDIN Competent Authorities and the Multi-stakeholder Advisory Group under WP4, other WPs and related external meetings to achieve a robust consensus before the final stage of bilateral negotiations with each member state and workshops to support and build capacity for implementation.

### Evaluation of ERN integration aspects in the existing National RD Plans/Strategies

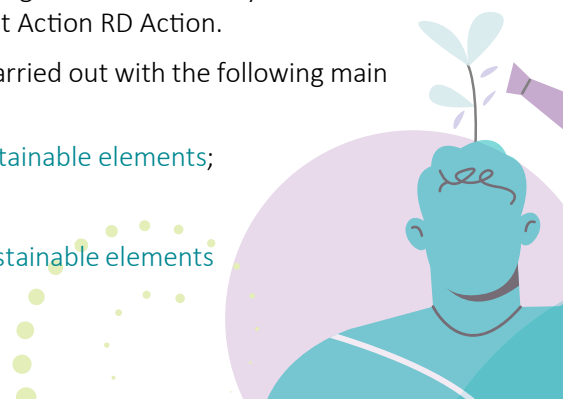
Based on the preliminary analysis of Rare Disease National Plans/National Strategies (RD NP/NS), three countries have never had a NP/NS, nine member states have expired RD NP/NS (for some of them new RD NP/NS are being developed or are in the adoption phase), eleven RD NP/NS were adopted in the pre-ERN era (i.e. until 2016). Many aspects of ERN integration are not or only minimally covered by the existing RD NP/NS (although in some cases they are supported by other legal acts or organisational measures).

A number of additional tasks and activities to collect as much information as possible include:

- translation of all available RD NP/NS and related legal acts through the Digital Europe eTranslation tool with further validation by the Competent Authorities;
- liaison with WP23 of the European Rare Disease Research Alliance (ERDERA), "National Mirror Group promotion and national alignment", with the aim to leverage on the established NMG channels and the activities foreseen in this WP, including collection of information through dedicated surveys based on the principles of the previous State-of-the-Art resource under the Joint Action RD Action.

After the compilation of these resources, an [in-depth analysis](#) will be carried out with the following main objectives:

- to examine the [current state of the coverage of the core sustainable elements](#);
- to [identify best practices of the ERN integration aspects](#);
- to [set the ground for the legal implementation](#) of the [core sustainable elements](#) and the exchange of best practices among member states.



## WP5

## TASKS

**T5.1: Mapping of existing national governance models for ERN-HCPs and care pathways in member states for a complete understanding of national governance systems and RD policies**

**T5.2: Identification of existing best practices, gaps and deficiencies**

Dialogue with regional stakeholders involved in RD programs. Assessment of the level of knowledge about ERNs among general practitioners. Dissemination of ERNs activities to territorial medicine services.

**T5.9: Development of monitoring indicators for ERN integration**

- Collection of data and information from different sources;
- Analysis of survey results to understand the current state of organizational integration in the NHS as well as integration into care processes at the national level;
- Scoping literature review to evaluate existing indicators for the implementation of healthcare plans and programs.

# ACHIEVEMENTS

**T5.1: Survey number 2**

All 28 countries contacted responded to the survey, and data was extracted and analysed. From the collected information on current rare disease healthcare policies and their governance, we can anticipate that 19 out of 28 countries have an active National Rare Diseases Plan or Strategy. The majority of countries, 20 out of 28, have a centralised healthcare system organisation. It is noteworthy that respondents from the same countries gave heterogeneous answers regarding their perception of the National Rare diseases Plan or Strategy (prevention, diagnosis, treatment, and research), which requires an in-depth analysis.

**Collaboration proposal between WP5 and WP8 to set up a National Data Hub**

During the online meetings held on October 8 and 30, 2024, with Orphanet France, the Italian Ministry of Health, and regional authorities, the feasibility of the proposal for the creation of a National Data Hub and the evaluation of ERNs was discussed. The National Data Hub project was introduced at the JARDIN Steering Committee meeting (October 31, 2024). The abstract of the proposal and the definition of the data flow were revised by the Italian Ministry of Health and the regional authorities, together with WP8 in December 2024.

**First pilot study on the Rare Disease Networks in Italy** was presented by RDCC Veneto (Paola Facchin and Monica Mazzucato) during the WP5 meeting on October 8 2024. At least four indicators were identified in Italy that could predict the selection of Rare Disease Reference Hospitals or HCP ERN hospitals. Several countries expressed an interest in replicating the pilot study using the same methodology to check whether the same or different indicators are identified.

**T5.9** – The literature review is ongoing and will be completed by January 2025.

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WP

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# WHAT IS NEXT?

## T5.1 - Survey number 2

Collect final answers from specialised referees for the heterogenous answers. Clarify the reimbursement systems by country with new close-ended questions.

## The third meeting of the Italian component

The third meeting of the Italian component of the JARDIN Joint Action is planned to be held in Rome on May 23, 2025, organised by the Italian Competent Authority, Istituto Ortopedico Rizzoli (Luca Sangiorgi). Italian healthcare providers that are members of the 24 ERNs will be present, including representatives of the planning departments of the Ministry of Health and regional authorities. The National Data Hub proposal will be presented at this meeting.

## First pilot study on the Rare Disease Networks

Replication of the study in other countries to explore which are the predictive variables to be selected as a RD Hospital or as a HCP ERN member.

## T5.2 – Planning of regional congresses aimed at disseminating the activities of ERNs to general practitioners and territorial medicine services

## T5.9 – Planning the first online meeting with key stakeholders

Including the WP5 working group, the ERN coordinators, the EURORDIS Amequist task force representative(s), and members of the BoMS in Q1-2025. The review results and surveys will provide the background for discussion at the meeting. The proposed date is February 19, 2025, at 4.30 pm.

WP5





WP6

# TASKS

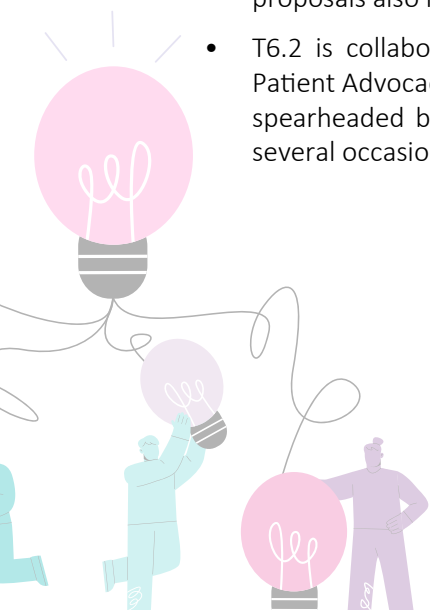
## T6.1: Signposting of expertise and multidisciplinary care pathways

- The HSE (NRDO) conducted an analysis of the revised Orphanet prevalence dataset consisting of 390+ conditions based on Wakap et al (EJHG, 2020) with an updated active list provided by Orphanet France.
- WP6 members by consensus agreed to exclude rare cancers, infectious diseases, and poisonings. WP members, Prof Madara Auzenbaha (Latvia), Prof Birute Tumiene, (Lithuania), Dr Ioana Streata (Romania), and Prof Eileen Treacy (Ireland) conducted an initial dataset validation with final validation completed by Orphanet France.
- Orphanet in OD4RD2 WP5 has assigned Orphacodes to the ERN thematic/sub-thematic groups and was validated by the ERN coordinators. This includes less common/ultra-rare conditions and this dataset will feed into T6.1.

## T6.2: Model (reference) care pathways for RDs or groups of RD

- Since its establishment in July, the T6.2 Working Group has developed a model process for care pathway development.
- Emerging from this work and in parallel, the Working Group developed a Step-by-Step Clinician Guide which is a summarised version of the model process detailing the main steps, actions, roles and responsibilities involved in the process, to support ERN discussions.
- Alongside this work, the Working Group has developed a glossary of terms to ensure consistency in the usage of common terms within the scope of the JARDIN project such as care pathway, patient journey etc. It is intended that the T6.2 Glossary will form part of an overall JARDIN Glossary.
- Earlier this year all ERN Coordinators were contacted to establish interest in the development of RD care pathways in the context of JARDIN's T6.2. 14 ERNs engaged with the T6.2 Leads and to date 12 ERNs have provided their proposed RD/group of RDs for care pathway development. Care pathway proposals also included the request to develop a generic care pathway.
- T6.2 is collaborating with EURORDIS, who are engaging with representatives of the European Patient Advocacy Groups (ePAGs) in parallel to the WG. An ePAG Guideline and Pathway Task Force, spearheaded by Matt Bolz-Johnson at EURORDIS, was established. This Task Force convened on several occasions to review and contribute to the methodology of care pathway development.

WP6



# ACHIEVEMENTS

## T6.1: Signposting of expertise and multidisciplinary care pathways

A refined dataset of 341 of the most prevalent rare diseases mapped to ERNs.

## T6.2 Model (reference) care pathways for RDs or groups of RDs

- The development of a model process for care pathway development including a Step-by-Step Clinician Guide.
- Development of a glossary of care pathway terms.
- A webinar themed 'Added Value of Patients Involvement in the Design of Care Pathways' was held that showcased best practice examples of patient involvement in the design of care pathways.
- The ePAG Guideline and Pathway Task Force developed a patient Toolkit for involving patients in the development of care pathways.

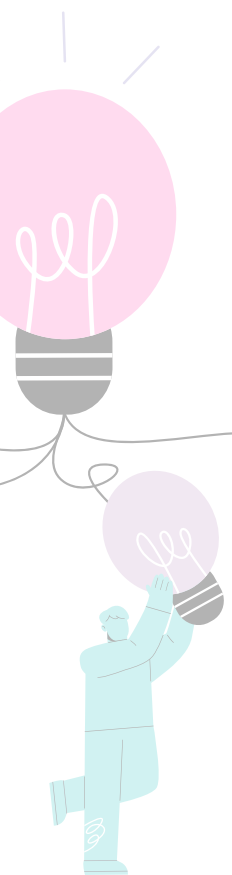
# WHAT IS NEXT?

## T6.1: Signposting of expertise and multidisciplinary care pathways

- The development of an SOP that will provide an agreed process for member states to name specialists as ERN entry points for the less common / ultra-rare diseases.
- T6.1 Leads will collaborate with WP 8 colleagues involved in T8.5 (Visualisation of RD Expert Centres with ERNs and NRNs) building on activities from T8.5 to adapt the SE-Atlas model, that will support the development of a sign-posting tool.

## T6.2 Model (reference) care pathways for RDs or groups of RD

- Integration of this work into one overall healthcare professional and patient representative Care Pathway Toolkit which will also include helpful resources and tools to support the care pathway development teams.
- T6.2 Leads will have briefing and training meetings with care pathway Clinical Leads, Pathway Project Managers and Patient Representative Leads across January and February 2025.







WP  
7-1

# TASKS

## T7.1: To perform a state-of-the-art analysis of existing structures, models, and initiatives on member state level

The first step in achieving the objective of WP7-1 is the analysis of existing structures, models and initiatives at member state level in relation to NRNs. For this purpose, a quantitative (Surveys: Current State NRN and Target State NRN) and a qualitative (focus group workshops) approach have been taken.

- **Survey Current State NRN:**

The Survey Current State NRN was carried out to obtain information on the existence of NRNs or similar structures in member states. We have received at least one response from each country on the current situation in their country. The results of this analysis formed the basis for D7.1 (State-of-play NRN) due in December.

- **Survey Target State NRN:**

The Survey Target State NRN aimed to find out how respondents think NRNs should be structured and implemented within healthcare systems. The results from a total of 401 respondents are currently being analysed. The analysis is planned by member state and by stakeholder group and is being carried out in collaboration with the WP7-1 core group, which was formed at the very beginning of the JARDIN project and consists of experts in the RD field from 10 different countries. Based on the results of the Target State NRN Survey, the core group members will develop national recommendations for the implementation and integration of NRNs for their respective countries.

- **Focus Group Workshops:**

To perform a qualitative analysis, five online workshops in the form of focus groups are organised in 5 member states – Germany, France, Estonia, Denmark, and Hungary. The objective of these meetings is to gain a more in-depth understanding of the challenges associated with establishing NRNs in those member states. These focus groups bring together experts from various fields of healthcare in rare diseases.

# ACHIEVEMENTS

## Survey Current State NRN

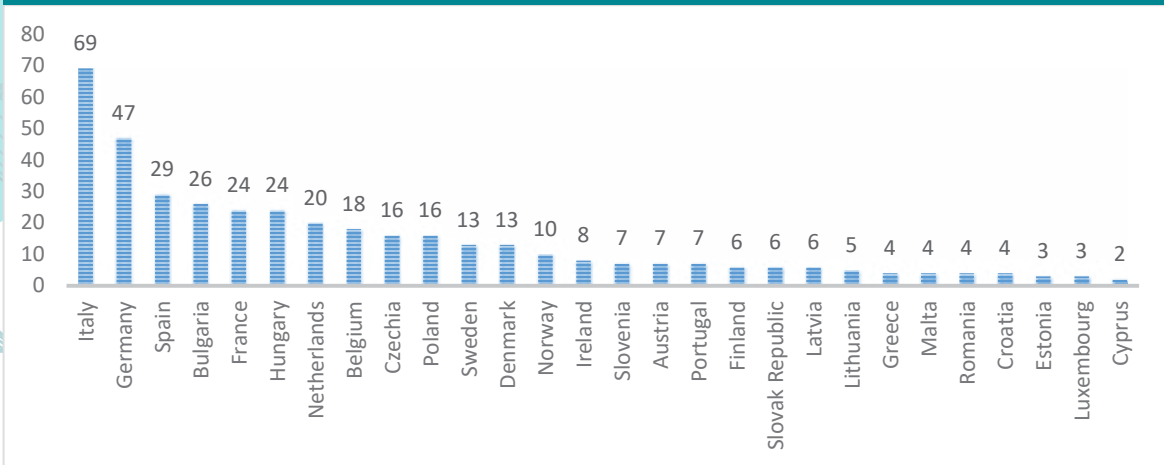
In December the analysis of the Current State Survey was completed.

## Survey Target State NRN

The Survey Target State NRN has been closed and a total of 401 respondents are included in the analysis. The responses from each country are shown in Figure 1. In mid-December, a meeting was held with the WP7-1 core group to discuss the scope for the development of the national recommendations. Therefore, the members of the WP7-1 core group received a descriptive evaluation of the survey with regard to all 401 responses. Additionally, they received a country-specific result report based on their respective country. The objective is to develop national recommendations based on these results and to specify these in a country-specific stakeholder group as part of a Delphi process.



**Figure 1: Target State NRN - responses per country**



### Focus Group Workshops

The focus group workshops have already been successfully conducted, and the qualitative analysis of those workshops is ongoing.

# WHAT IS NEXT?

### Survey Target State NRN

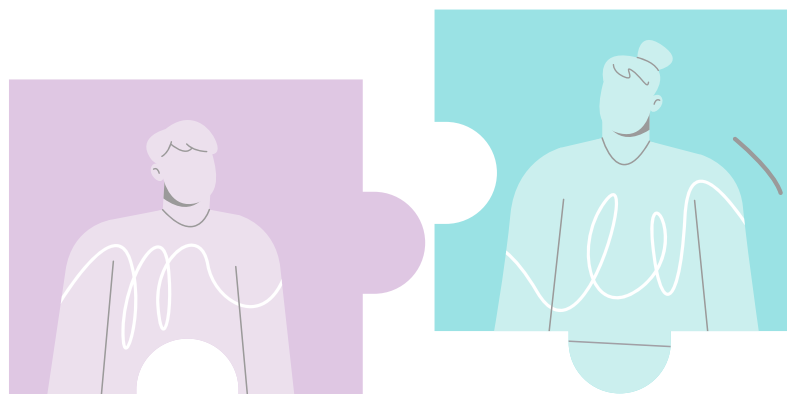
The national recommendations are to be developed by mid-April using an adapted Delphi process. During this period, several appointments will be offered to clarify questions and support the process of developing the national recommendations.

### Focus Group Workshops

The analysis of the five focus group workshops is currently ongoing, and it is anticipated that this will be completed by the middle of April.

### Survey Current State NRN

The analysis of the current state of NRNs and similar national structures raised issues that will be addressed by re-contacting the survey participants.



WP 7-1

WP  
7-2

# TASKS

In WP7-2 on undiagnosed diseases, the task of the first year of JARDIN was to [evaluate the status quo of measures and initiatives for undiagnosed patients in member states](#). This is needed in order to collect good practice examples that might serve as blueprints for measures within the different European healthcare systems, as well as to support capacity building for the implementation of the proposed measures in member states.

In a first step, after [identifying experts for undiagnosed diseases in each country](#), we launched a survey directed at health officials, as well as health professionals and researchers. In addition to questions on undiagnosed disease programs (UDPs) and initiatives, it also included questions regarding other tasks in WP7-2, namely the work on recommendations for undiagnosed registries, national teleconsultation systems for multidisciplinary panel discussions, and an SOP for assigning of ORPHA:616874 for undiagnosed rare diseases in Centres of Expertise. The survey was concluded in September 2024.

Subsequently, [personal interviews with experts from each member state were conducted](#) between October and December in order to refine and extend the survey results regarding undiagnosed disease programs and initiatives. In parallel to the survey on UDPs, a survey directed at patient organisations was conducted, forming the basis for the task on recommendations for undiagnosed patient organisations.

# ACHIEVEMENTS

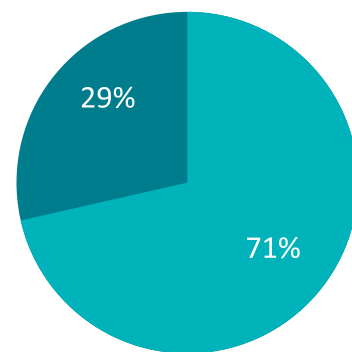
## D7.8 - State-of-play Report Undiagnosed Disease Programs

The survey on the status of UDPs in member states was analysed and a report on the results prepared. It was submitted as D7.8 in December and is currently under review by HaDEA.

Regarding [concrete results of the survey on undiagnosed disease programs and initiatives](#), 339 persons responded in total. There was at least one response from each member state. Overall, it is encouraging that the majority of member states have at least some initiatives for undiagnosed patients, as shown in the pie chart. Detailed insights are provided in the official report, which was sent out to all JARDIN partners as a draft version, as well as in an internal document on the results of the interviews, which is currently in preparation.

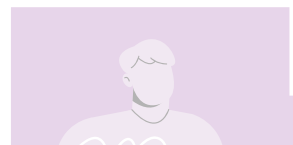
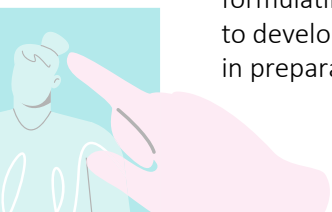
Availability of measures/initiatives for undiagnosed diseases in member states

■ Yes ■ No



# WHAT IS NEXT?

Work on the [recommendations for undiagnosed disease programs](#) got underway in January and will be continued over the following months, including a consensus process within the JARDIN consortium. In parallel, the working groups on the aforementioned other tasks in work package 7-2 will be formulating their recommendations and initiate the consensus process. Of note, a position paper "How to develop and implement Undiagnosed Patient Registries in Europe – Recommendations" is currently in preparation in the frame of T7.7.

WP  
7-2

WP8

# TASKS

## T8.1: Identification of current barriers to RD data sharing and inventory of existing solutions

In T8.1, we analysed the current situation of barriers and existing solutions for data sharing at the HCP, national, and ERN levels. To do this, we surveyed clinicians and IT experts from ERN healthcare providers (HCPs), as well as ERN Registry Coordinators and national authorities. Our aim is to understand how data on rare diseases (RDs) is organised within HCPs and ERN registries, and to identify implementable solutions to improve interoperability between different stakeholders. Currently, we are focusing on improving data quality from the RD data management surveys. Our goal is to generate actionable insights that support accurate research and enable practical improvements across ERNs, for the benefit of people living with rare diseases and complex conditions.

## T8.2: Implementable solutions to improve semantic accuracy and interoperability of RD health data

The overarching objective of T8.2 is to identify implementable solutions that will improve the semantic accuracy and the interoperability of Rare Disease (RD) Health Data. This is to be accomplished via two subtasks:

- 1. Defining minimum data sets of core data elements that will maximise the use of semantic standards across national systems and ERN registries.**
- 2. Raising awareness on ORPHAcodes and providing support for their implementation at the local, national, and European levels.**

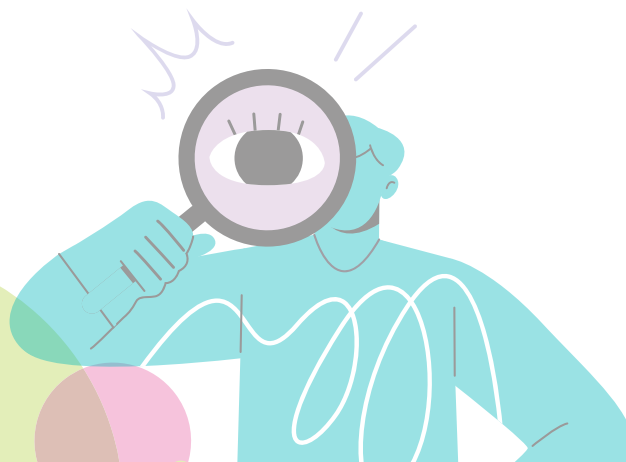
Ultimately, the enhanced capture of interoperable health data is expected to improve the visibility of RD patients, thereby leading to better health outcomes and care pathways for these patients.

In order **to achieve these objectives**, T8.2 is working closely with

- **Tasks 8.1 and 8.3 to identify and to develop solutions to the barriers that impede RD data capture and sharing**
- **T8.4 to demonstrate the added value of ORPHAcode implementation within a healthcare provider (HCP) and regional health network**
- **Work Package (WP) 5 to validate the use-case of a minimum data set (comprising ORPHAcodes in addition to other core data elements) in appraising HCPs against certain KPIs for ERN eligibility.**

These synergistic and collaborative efforts are taking place through surveys, data analyses, expert forums, and in the case of WP5, a pilot study.

8  
WP8



# 8 WP

## T8.3: Propose and develop implementable solutions to overcome organisational, technical, and legal barriers to integration of national health systems and ERN data management

Under the umbrella of T8.3, Subtask 8.3.1 is responsible for [identifying technical solutions between HCP and RD management systems](#) to support the creation of implementable solutions to overcome technical barriers to the integration of national health systems and ERNS.

The current mid-term objectives of T8.3.1 are to conduct a requirements analysis from internal expertise to identify initial use cases that can drive experimentation (e.g. hackathons) with current tools. These experiments will help to [identify the most prominent challenges and possible solutions to technical barriers](#).

## T8.4: Testing and implementing integration solutions in agile mode

As part of the initiative to establish an efficient, secure, and comprehensive data transfer between the Electronic Health Records (EHR) of Healthcare Providers (HCP), national registries, and the ERN registries, T8.4 aims to enhance and apply the recommendations developed through T8.2 and T8.3, following an agile way of implementation.

Under the purview of T8.4, Subtask 8.4.3 is charged with steering a demonstrator project designed to showcase the added value of using implemented rare disease (RD) datasets for monitoring HCP and ERN activities and key performance indicators (KPIs), and for data exchange with ERN registries.

[This project is to be conducted in three phases over three years](#), initially with Aarhus University Hospital, but also open to additional countries and healthcare providers wishing to partake. The **three phases** are as follows:

- 1. Technical implementation and onboarding of the ORPHAcodes.**
- 2. Coding and capacity building.**
- 3. Data extraction and analysis against defined KPIs.**

[At present, 8.4.3 is in transition between the first and second phase](#). Regular planning meetings for Phase 2 began in September 2024, and Phase 1 has concluded with the participation of the technical team of the AUH to the OD4RD2 technical workshop held on 18 December 2024, and the subsequent release of the ORPHAcodes in their electronic health records at the end of January 2025. This workshop, organised by the OD4RD2 project ([www.od4rd.eu](http://www.od4rd.eu)), has also included T8.3 as invitees, given the interlinkages and synergies between these tasks and projects.

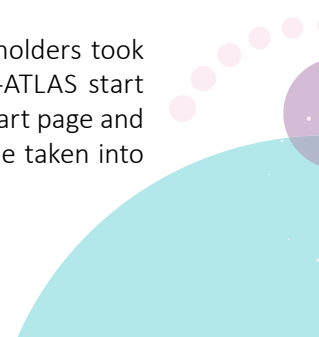
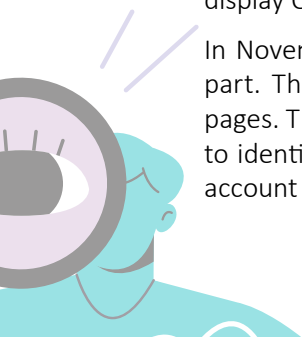
Furthermore, 8.4.3 continues to collaborate with T8.2, and will include the demonstrator participants from WP5 with the aims to validate the use-case of a minimum data set (comprising of ORPHAcodes in addition to other core data elements) in appraising HCPs against certain KPIs for ERN eligibility.

## T8.5: Visualisation of RD Expert Centres with ERNs and NRNs

T8.5 focuses on the further development of SE-ATLAS with regard to multilingualism and the integration and management of international Orphanet data.

To implement multilingualism, various translation tools were analysed and evaluated using relevant metrics. For the integration of Orphanet data, the SE-ATLAS team is in continuous contact with Orphanet partners in France and Germany in order to identify the best solution for integration and to be able to display Orphanet data.

In November, a usability workshop was held in which various international JARDIN stakeholders took part. The workshop focused on the usability and content design of country-specific SE-ATLAS start pages. The aim was to test the options for displaying country-specific information on the start page and to identify potential for design optimisation. The results are being analysed so they can be taken into account for further development.



# ACHIEVEMENTS

## WP8

### T8.1

We have finalised the data collection of the RD management survey and have gained first insights into the results.

### T8.2

Thus far, T8.2 has successfully:

- **Completed a first analysis of the WP8 survey data, gaining preliminary insights into the practices and obstacles surrounding the first capture and interoperability of RD patient data.**
- **Established the foundation for the pilot study, in conjunction with WP5, to be completed in Italy.**

### T8.3 and T8.4

- We contributed to the analysis of survey results from T8.3.1, with a specific focus on the FAIR data module.
- We have defined a use case centred on supporting clinicians conducting clinical trials during weekly meetings among those involved in 8.3.1 and 8.3.2. (these meetings will now be held every fourth Wednesday of the month):
  - The proposed use case involves providing a clinician with a list of patient IDs, diagnosis status, patient age, genotype, and hospital, enabling patient recruitment either directly or through the respective centres.
  - This use case is currently open for feedback to refine its scope and ensure alignment with project goals.
- T8.4.3: successfully transitioning between Phases 1 and 2 of the demonstrator project.

### T8.5

On November 19, 2024, the usability workshop for T8.5 was successfully conducted. 21 participants from partner countries and various work packages came together to evaluate the usability and content design of country-specific SE-ATLAS start pages. Valuable feedback was collected during the workshop which is now being analysed.



# WHAT IS NEXT?

## 8 WP

### T8.1

- Conduct follow-up interviews as needed to capture diverse ERN perspectives.
- Host a webinar with experts to discuss results and identify implementable solutions.

### T8.2

Going forward, T8.2 plans to:

- Conduct interviews in order to build upon the insights from the WP8 survey and to better refine the minimum data sets necessary to [satisfy the primary and secondary use-cases for RD data](#).
- Continue the [collaboration with WP5](#) to advance the pilot study.
- Present our [recommendations](#) regarding minimum RD data sets and ORPHAcodes implementation to [Ministry of Health level representatives and other key stakeholders](#) during a [workshop](#) scheduled for June 2025.

### T8.3 and T8.4

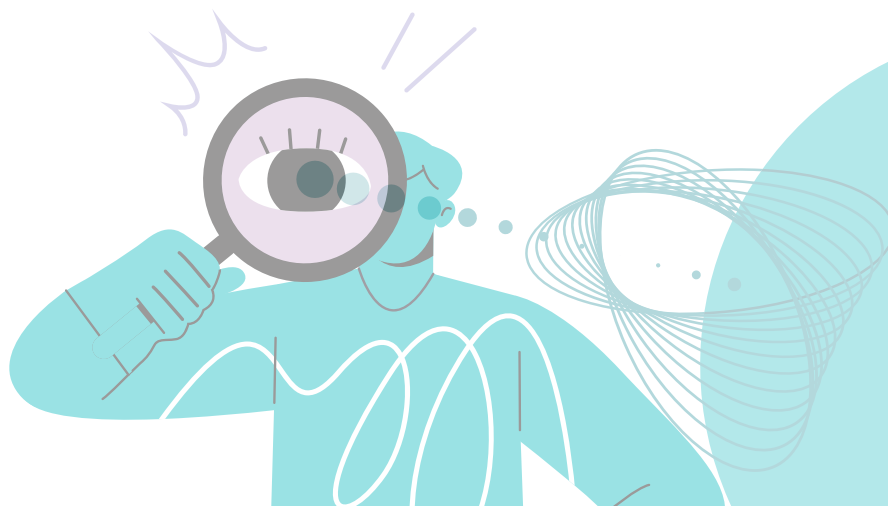
In the next phase, we will plan and prepare a hackathon to develop a proof of concept based on the defined use case. This hackathon will serve a dual purpose: implementing the use case and identifying practical challenges in [FAIRification](#). We aim to foster collaboration by inviting contributions from other tasks, such as 8.4 and 9.1, to enhance the alignment and impact of the hackathon outcomes.

For task 8.4.3, next steps are as follows:

- [Finalise the transition to Phase 2 of the project](#)
- [Continue the collaboration with WP5 and T8.2](#)

### T8.5

The results of the usability workshop on the individual country-specific SE-ATLAS start pages are analysed in T8.5. The objective is to evaluate the gathered feedback thoroughly and incorporate it into the further development of the SE-ATLAS.



# WP9

# TASKS

Currently, the WP9 team has been focusing on [key tasks related to the survey and mapping of existing mechanisms to support ERN-related activities at the national, regional, and local/hospital levels](#), which are coordinated with WP 5, 6, and 8.1.

As part of T9.1.2, the team has been [identifying ERN-related activities](#) (administrative, organisational, and/or financial) and [exploring potential mechanisms to strengthen the sustainability of ERNs](#).

Survey 4 focused on the current working conditions and potential support needs of hospitals/healthcare providers hosting ERN clinical centres. JARDIN WP9 is interested in learning how each ERN centre has been organising its clinical work, education, and research activities within the ERN context and what sources of funding are currently available for these purposes.

In addition, WP9 participants have started to work on an in-depth analysis of previous studies on the utility of the CPMS and its reimbursement models, particularly the analysis of recent ERN pilots initiated and supported by the European Commission, using different reimbursement strategies and concepts. This analysis also includes assessing the feasibility of the CPMS in real-world clinical practice. Furthermore, the team is analysing the legal aspects of CPMS reimbursement models and how they can be implemented based on existing national reimbursement mechanisms.

Lastly, the WP9 team has convened the first meeting of the Health Economy group, with the goal of conducting a general analysis of cost-effectiveness and economic impact within the healthcare system by integrating ERN HCPs, focusing on reality-based cost analysis and the development of a pilot health economic model.

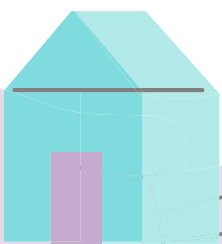
# ACHIEVEMENTS

After a detailed analysis of the first round of the survey data, the WP9 team has identified that, unfortunately, many countries as well as ERNs were underrepresented in the survey sample. For this reason, WP9 re-opened the survey to recruit more respondents and to ensure the representativeness and accuracy of the results. The survey has just been closed and the first part of the final analysis is running.

[The working group for the CPMS utility and reimbursement](#) has been formed to include the ERN Helpdesk Administrator and IT team representatives as well as a lawyer with expertise in the healthcare legislation. The CPMS survey has been finalised and distributed among all 24 ERN managers and coordinators for completion within 3 weeks.

[The collaboration with health economy experts](#) has been established and a working group on this topic has started to gather published resources.

# WHAT IS NEXT?



The next WP9 goal is to draw concrete results from the survey and formulate the identification of ERN-related activities requiring support at the national, regional, or local levels (administrative, organisational, and/or financial), along with potential mechanisms to strengthen the sustainability of ERNs: an exhaustive inventory of items. Additionally, the team will evaluate the results from the CPMS questionnaire and work on its reimbursement models within cross-border healthcare for patients with rare diseases. They will also prepare an analysis of the available literature on the cost-effectiveness and economic impact within the healthcare systems by appropriate organisation of the rare disease healthcare.





# JARDIN

INTERNAL NEWSLETTER

ISSUE No 2

JANUARY 2025

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*Let's keep in touch!*

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