ANNEX 1

EU4Health Programme (EU4H)

Description of the action (DoA)

Part A
Part B

Version 1.0
15 April 2021

IMPORTANT NOTICE

What is the Description of the Action (DoA)?

The Description of the Action (DoA) is the Annex of the Grant Agreement which contains the details of how the project will be carried out. For EU framework partnerships for grants (FPAs) this Annex is called Action Plan.

It consists of 2 parts, which must be generated from the submitted proposal:

- Part A contains structured tables with project information
- Part B is a narrative description on the work to be carried out.

Part A is generated by the IT system. It is based on the information which you enter into the Portal Grant Preparation screens.

Part B (+ annexes) must be uploaded on the Grant Preparation Documents screen.

⚠ Make sure that Part B is synchronised with the information entered into the screens. Make sure that any changes are agreed with us.

DESCRIPTION OF THE ACTION (PART A)

COVER PAGE

Part A of the Description of the Action (DoA) must be completed directly on the Portal Grant Preparation screens.

PROJECT *Grant Preparation (General Information screen) — Enter the info.*	
Project number:	101129863
Project name:	Joint Action on integration of ERNs into national healthcare systems
Project acronym:	JARDIN
Call:	EU4H-2022-JA2-IBA
Topic:	EU4H-2022-JA2-IBA-05
Type of action:	EU4H-PJG
Service:	HADEA/A/01
Project starting date:	Fixed date: 01/01/2024
Project duration:	36

TABLE OF CONTENTS

PROJECT SUMMARY

Project summary

Grant Preparation (General Information screen) — Provide an overall description of your project (including context and overall objectives, planned activities and main achievements, and expected results and impacts (on target groups, change procedures, capacities, innovation etc)). This summary should give readers a clear idea of what your project is about.

Use the project summary from your proposal.

European Reference Networks (ERN) are multinational networks of highly specialized healthcare providers (HCP) across Europe in thematically coherent medical domains, addressing rare or low-prevalence complex diseases requiring exceptional concentration of expertise and resources. There are currently 24 ERN providing virtual expert consultations for patients from all member states (MS) and Norway, as well as support knowledge generation, professional training and education, and research. In this project, we aim to enhance the impact of the ERN even further by addressing all aspects of their better integration into healthcare systems in MS. This will include improving national governance of ERN-HCP, quality assurance models, patient pathways and ERN referral systems, supporting the formation of national reference networks and undiagnosed diseases programs or equivalent strategies interlinked with ERN, improving data management, aiming to finally achieve full interoperability of regional, national and European health data sources, and identifying national support options for ERN-HCP. Furthermore, we will develop strategies for systematic dissemination of information on the ERN, with a specific emphasis on patients as well as the medical community. A main focus will be to ensure the sustainability of the proposed actions and implementations, which we propose to integrate into updated national plans and strategies for rare diseases in MS.

LIST OF PARTICIPANTS

PARTICIPANTS

Grant Preparation (Beneficiaries screen) — Enter the info.

Partner No	Role	Short Name	Legal Name	Country	PIC
1	COO	MUW	MEDIZINISCHE UNIVERSITAET WIEN	AT	999989976
2	BEN	BHTC	SERVICE PUBLIC FEDERAL SANTE PUBLIQUE, SECURITE DE LA CHAINE ALIMENTAIRE ET ENVIRONNEMENT	BE	998853815
2.1	AE	Sciensano	SCIENSANO	BE	906160809
3	BEN	MUS	MEDICAL UNIVERSITY SOFIA	BG	999857571
4	BEN	MoH-CY	Ministry of Health of the Republic of Cyprus	CY	994267946
5	BEN	GUH	VSEOBECNA FAKULTNI NEMOCNICE V PRAZE	CZ	989280012
6	BEN	BMG	BUNDESMINISTERIUM FUER GESUNDHEIT	DE	998954889
6.1	AE	BfArM	BUNDESINSTITUT FUR ARZNEIMITTEL UND MEDIZINPRODUKTE	DE	998293931

6.2	AE	NAMSE-GSt	MUKOVISZIDOSE INSTITUT - GEMEINNÜTZIGE GESELLSCHAFT FUR FORSCHUNG UND THERAPIEENTWICKLUNG	DE	983390366
6.3	AE	UKT	Universitaetsklinikum Tuebingen	DE	986544224
6.4	AE	UKHD	UNIVERSITATSKLINIKUM HEIDELBERG	DE	999841081
6.5	AE	UKW	UNIVERSITAETSKLINIKUM WUERZBURG - KLINIKUM DER BAYERISCHEN JULIUS-MAXIMILIANS-UNIVERSITAT	DE	997297353
6.6	AE	UK FFM	KLINIKUM DER JOHANN WOLFGANG VON GOETHE UNIVERSITAET	DE	999883082
7	BEN	AUH	AARHUS UNIVERSITETSHOSPITAL	DK	999643880
8	BEN	TUH	SIHTASUTUS TARTU ULIKOOLI KLIINIKUM	EE	999518556
9	BEN	EODY	ETHNIKOS ORGANISMOS DIMOSIAS YGEIAS	EL	896563726
10	BEN	FIBHULP	FUNDACION PARA LA INVESTIGACION BIOMEDICA DEL HOSPITAL UNIVERSIATRIO LA PAZ	ES	996155760
11	BEN	THL	TERVEYDEN JA HYVINVOINNIN LAITOS	FI	996697893
12	BEN	DGOS	MINISTERE DE LA SANTE ET DE LA PREVENTION	FR	998887377
12.1	AE	HUS	HOPITAUX UNIVERSITAIRES DE STRASBOURG	FR	951206930
12.2	AE	HCL	HOSPICES CIVILS DE LYON	FR	999469765
12.3	AE	APHP	ASSISTANCE PUBLIQUE HOPITAUX DE PARIS	FR	999645432
12.4	AE	INSERM	INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE	FR	999997833
13	BEN	UHCZ	KLINICKI BOLNICKI CENTAR ZAGREB	HR	994956064
13.1	AE	MoH-HR	MINISTARSTVO ZDRAVSTVA REPUBLIKE HRVATSKE	HR	933852272

14	BEN	NNGYK	NEMZETI NEPEGESZSEGUGYI ES GYOGYSZERESZETI KOZPONT	HU	998706957
15	BEN	HSE	HEALTH SERVICE EXECUTIVE HSE	IE	993521919
16	BEN	IOR	ISTITUTO ORTOPEDICO RIZZOLI	IT	999445709
16.1	AE	AOUP	AZIENDA OSPEDALIERO UNIVERSITARIA PISANA	IT	988322040
16.2	AE	ISS	ISTITUTO SUPERIORE DI SANITA	IT	999978821
16.3	AE	HSM	IRCCS OSPEDALE POLICLINICO SAN MARTINO	IT	959034442
16.4	AE	RV	REGIONE DEL VENETO	IT	999465691
16.5	AE	ASUFC	AZIENDA SANITARIA UNIVERSITARIA FRIULI CENTRALE	IT	894464355
16.6	AE	FPG	FONDAZIONE POLICLINICO UNIVERSITARIO AGOSTINO GEMELLI IRCCS	IT	918081430
16.7	AE	IT-MoH	MINISTERO DELLA SALUTE	IT	999531942
17	BEN	VULSK	VIESOJI ISTAIGA VILNIAUS UNIVERSITETO LIGONINE SANTAROS KLINIKOS	LT	991636530
18	BEN	DISA	MINISTERE DE LA SANTE	LU	998888153
19	BEN	CCUH	BERNU KLINISKA UNIVERSITATES SLIMNICA VALSTS SIA	LV	904374748
20	BEN	MFH	Ministry for Health - Government of Malta	MT	974144423
21	BEN	VWS	MINISTERIE VAN VOLKSGEZONDHEID, WELZIJN EN SPORT	NL	998886601
21.1	AE	ZON	ZORGONDERZOEK NEDERLAND ZON	NL	999544164
21.2	AE	RUMC	STICHTING RADBOUD UNIVERSITAIR MEDISCH CENTRUM	NL	892057785
21.3	AE	LUMC	ACADEMISCH ZIEKENHUIS LEIDEN	NL	999990849

21.4	AE	ERASMUS MC	ERASMUS UNIVERSITAIR MEDISCH CENTRUM ROTTERDAM	NL	999988424
22	BEN	HSO	HELSE SOR-OST RHF	NO	905487726
22.1	AE	HDIR	HELSEDIREKTORATET	NO	974772304
22.2	AE	OUS	OSLO UNIVERSITETSSYKEHUS HF	NO	991104000
23	BEN	MoH Pl	THE MINISTRY OF HEALTH OF THE REPUBLIC OF POLAND	PL	973658453
24	BEN	DGS	MINISTERIO DA SAUDE - REPUBLICA PORTUGUESA	PT	986364095
24.1	AE	CHLN	CENTRO HOSPITALAR LISBOA NORTE EPE	PT	952617407
25	BEN	ECCHC	SPITALUL CLINIC JUDETEAN DE URGENTA CRAIOVA	RO	911286968
26	BEN	SoS	SOCIALSTYRELSEN	SE	998248147
27	BEN	UKCL	UNIVERZITETNI KLINICNI CENTER LJUBLJANA	SI	999882306
28	BEN	MOH SR	MINISTERSTVO ZDRAVOTNICTVA SLOVENSKEJ REPUBLIKY	SK	999825173
29	AP	MoH Ukraine	Ministry of Health of Ukraine	UA	883277442

LIST OF WORK PACKAGES

Work packages

Grant Preparation (Work Packages screen) — Enter the info.

Work Package No	Work Package Name	Lead Beneficiary	Effort (Person-Months)	Start Month	End Month	Deliverable No(s)
1	Coordination	1 - MUW	222.1	1	36	1-11
2	Dissemination	10 - FIBHULP	124.2	1	36	12-24
3	Evaluation	13 - UHCZ	87.5	1	36	25-29
4	Sustainability	17 - VULSK	81.5	1	36	30-31
5	National governance and quality assurance models	16 - IOR	345.25	1	36	32-35
6	National care pathways and ERN referral systems	17 - VULSK	307.5	1	36	36-39
7	National reference networks and undiagnosed disease programmes or equivalent strategies interlinked with ERN	6.3 - UKT	436.7	1	36	40-46
8	Data management	12 - DGOS	794.75	1	36	47, 48
9	National support options for ERN-HCP	5 - GUH	178.23	1	36	49-51

STAFF EFFORT

									Staff effort per participant *Grant Preparation (Work packages - Effort screen) — Enter the info.*
Participant	WP1	WP2	WP3	WP4	WP5	WP6	WP7	WP8	WP9	Total Person-Months
MUW	114.5	3.0	1.0	3.0	8.0	8.0	151.0	6.0	1.5	296.0
BHTC	4.5	2.0	0.5	0.5	1.5	0	0	0	0	9.0
Sciensano	0	0	0	0	0	0	0	42.0	0	42.0
MUS	5.0	4.0	1.5	2.0	5.0	6.0	18.0	6.0	3.0	50.5
MoH-CY	4.5	2.0	0.5	0.5	0	0	1.5	9.0	5.0	23.0
GUH	4.5	2.0	0.5	1.5	4.0	10.0	4.0	11.0	76.0	113.5
BMG	1.5	0	0	0	0	0	0	0	0	1.5
BfArM	0	0	0	0	0	0	0	12.0	0	12.0
NAMSE-GSt	0	2.0	0.5	0.5	31.0	26.5	3.0	0	6.0	69.5
UKT	0	0	0	0	8.0	0	44.0	0	0	52.0
UKHD	0	0	0	0	0	10.0	0	29.0	13.0	52.0
UKW	0	0	0	0	0	0	27.0	21.0	0	48.0

UK FFM	0	0	0	0	0	0	21.0	97.0	0	118.0
AUH	4.5	2.0	0.5	0.5	0	0	1.0	54.0	0.5	63.0
TUH	4.5	2.5	0.5	1.0	0	0	17.0	26.0	8.0	59.5
EODY	5.0	2.0	0.5	0.5	14.5	15.0	25	0	25	87.5
FIBHULP	4.5	58.0	0.5	0.5	3.0	3.0	0	24.0	0	93.5
THL	4.5	2.0	0.5	0.5	0	9.0	0	3.0	0	19.5
DGOS	1.5	2.0	0.5	0.5	0	4.0	2.0	44.0	2.0	56.5
HUS	0	0	0	0	0	10.8	0	0	0	10.8
HCL	0	0	0	0	0	0	0	5.5	0	5.5
APHP	0	0	0	0	0	0	0	52.0	0	52.0
INSERM	0	0	0	0	0	0	0	135.5	0	135.5
UHCZ	4.5	2.0	72.0	0.5	0	3.0	3.0	0	0	85.0
MoH-HR	0	0	0	0	3.0	0	0	0	3.0	6.0
NNGYK	6.0	2.0	1.0	1.0	0	23.5	5.5	0	0	39.0
HSE	3.6	6.7	0.5	0.5	0	19.2	10.0	15.0	4.7	60.2
IOR	1.5	4.0	0.5	0.5	90.5	1.7	1.1	1.35	1.45	102.6

AOUP	0	0	0	0	23.5	0	0	0	0	23.5
ISS	0	0	0	0	28.0	0	0	0	0	28.0
HSM	0	0	0	0	1.25	0	0	0	0	1.25
VR	0	0	0	0	23.0	0	0	0	0	23.0
ASUFC	0	0	0	0	21.2	0	0	0	9.38	30,58
FPG	0	0	0	0	7.0	0	0	0	0.4	7.4
IT-MoH	0	0	0	0.5	1.0	0	0	0	0	1.5
VULSK	4.5	2.0	0.5	60.0	2.0	60.0	2.0	9.0	4.0	144.0
DISA	4.5	2.0	0.5	0.5	0	0	0	0	0	7.5
CCUH	4.5	2.0	0.5	0.5	0	6.0	14.0	0	0	27.5
MFH	4.5	2.0	0.5	0.5	0	3.0	0	2.0	1.5	14.0
VWS	1,5	0	0	0.5	0	0	0	0	0	2.0
ZonMw	0	2.0	0.5	0	4.0	4.0	4.0	0	4.0	18.5
RUMC	0	0	0	0	0	0	21.0	39.6	0	60.6
LUMC	0	0	0	0	0	0	0	68.6	0	68.6
Erasmus MC	0	0	0	0	7.5	0	0	0	0	7.5

HSO	1.0	0	0	0	2.0	0	0	9.6	0	12.6
HDIR	1.0	0	0.5	0	0	0	0	0	0	1.5
OUS	1.5	2.0	0	2.0	2.0	0	9.6	9.6	0	26.7
MoH PI	4.5	4.0	0.5	0.5	14.0	4.0	1.0	4.0	1.0	33.5
DGS	4.5	2.0	0.5	0.5	13.5	43.0	0	20.0	0	84.0
CHLN	0	0	0	0	6.0	6.0	0	0	0	12.0
ECCHC	3.5	2.0	0.5	0.5	12.0	16.0	12.0	0	0	46.5
SoS	3.0	2.0	0.5	0.5	0	7.0	7.0	7.0	0	27.0
UMCL	4.5	2.0	0.5	0.5	0	0	32.0	32.0	0	71.5
MOH SR	4.5	2.0	0.5	0.5	8.8	8.8	0	0	8.8	33.9
Total Person-Months	222.1	124.2	87.5	81.5	345.25	307.5	436.7	794.75	178.23	2577.73

LIST OF DELIVERABLES

Deliverables

Grant Preparation (Deliverables screen) — Enter the info.

The labels used mean:

Public — fully open (automatically posted online)

Sensitive — limited under the conditions of the Grant Agreement

EU classified — RESTREINT-UE/EU-RESTRICTED, CONFIDENTIEL-UE/EU-CONFIDENTIAL, SECRET-UE/EU-SECRET under Decision 2015/444. For items classified under other rules (e.g. national or international organisation), please select the equivalent EU classification level.

Deliverable No	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month)
D1	JARDIN interim report	1	1 - MUW	R	PU	18
D2	JARDIN final report	1	1 - MUW	R	PU	36
D3	KOM report	1	1 - MUW	R	PU	4
D4	Annual meeting 1 report	1	1 - MUW	R	PU	23
D5	Annual meeting 2 report	1	1 - MUW	R	PU	33
D6	Internal newsletter 1	1	1 - MUW	R	PU	6
D7	Internal newsletter 2	1	1 - MUW	R	PU	12
D8	Internal newsletter 3	1	1 - MUW	R	PU	18
D9	Internal newsletter 4	1	1 - MUW	R	PU	24
D10	Internal newsletter 5	1	1 - MUW	R	PU	30
D11	Internal newsletter 6	1	1 - MUW	R	PU	36

D12	Communication and dissemination plan	2	10 - FIBHULP	R	PU	6
D13	Website and social media channels	2	10 - FIBHULP	R	PU	9
D14	Internal newsletter 1 disseminated	2	10 - FIBHULP	R	PU	6
D15	Internal newsletter 2 disseminated	2	10 - FIBHULP	R	PU	12
D16	Internal newsletter 3 disseminated	2	10 - FIBHULP	R	PU	18
D17	Internal newsletter 4 disseminated	2	10 - FIBHULP	R	PU	24
D18	Internal newsletter 5 disseminated	2	10 - FIBHULP	R	PU	30
D19	Internal newsletter 6 disseminated	2	10 - FIBHULP	R	PU	36
D20	National stakeholder analysis in three countries	2	10 - FIBHULP	R	PU	12
D21	Final blueprint	2	10 - FIBHULP	R	PU	35
D22	Pilot evaluation report	2	10 - FIBHULP	R	PU	32
D23	Interim Dissemination Report	2	10 - FIBHULP	R	PU	18
D24	Final dissemination report	2	10 - FIBHULP	R	PU	36
D25	Evaluation plan	3	13 - UHCZ	R	PU	4
D26	Survey 1	3	13 - UHCZ	R	PU	12

D27	Survey 2	3	13 - UHCZ	R	PU	24
D28	Survey 3	3	13 - UHCZ	R	PU	34
D29	Evaluation final report	3	13 - UHCZ	R	PU	36
D30	Compendium of sustainability models and solutions	4	17 - VULSK	R	PU	33
D31	Recommendations for better integration of JA actions and sustainable elements into the National RD Plans / Strategies	4	17 - VULSK	R	PU	34
D32	Interim progress report (WP5)	5	16 - IOR	R	PU	24
D33	Report including recommendations for national governance models adapted to the different types of national health systems in Europe	5	16 - IOR	R	PU	35
D34	Report including recommendations for quality assurance models adapted to the different types of national health systems in Europe	5	16.1 - AOUP	R	PU	35
D35	Final implementation report WP5	5	16 - IOR	R	PU	36
D36	Tool for signposting of national expertise and linkage to ERN pathways	6	15 - HSE	DEM	PU	25
D37	Compendium (blueprint) of model care pathways for RD or groups of RD	6	15 - HSE	R	PU	34

D38	Recommendations for the organisation of national care pathways, referral systems to ERNs and incorporation of CPMS advice for rare and complex diseases	6	17 - VULSK	R	PU	34
D39	Toolkit of best practices in the implementation of care pathways	6	17 - VULSK	R	PU	35
D40	State-of-play report NRN / UDP	7	6.3 - UKT	R	PU	10
D41	Recommendations and models for NRN or equivalent strategies	7	6.3 - UKT	R	PU	21
D42	Recommendations for UDP in Europe	7	1 - MUW	R	PU	21
D43	Recommendations for a registry of undiagnosed patients in Europe	7	6.6 - UK FFM	R	PU	21
D44	European SOP for assigning ORPHAcode 616874 (undiagnosed RD) in CoE and ERN-HCP	7	6.3 - UKT	R	PU	21
D45	Document on recommendations for national patient organizations (PO) for undiagnosed patients	7	1 - MUW	R	PU	21
D46	Implementation report WP7	7	6.3 - UKT	R	PU	35
D47	Report on barriers to RD data sharing and existing solutions	8	6.4 - UKHD	R	SEN	15
D48	Recommendations for integration of data management between National Health Systems and ERN	8	21.3 - LUMC	R	PU	35

D49	Report on existing mechanisms to support ERNrelated activities of ERN-HCPs in MS	8	6.4 - UKHD	R	PU	18
D50	Guidance document on the requirements for national support of ERN-HCPs	9	5 - GUH	R	PU	36
D51	Recommendations for CPMS reimbursement models	9	5 - GUH	R	PU	35

LIST OF MILESTONES

Milestones

Grant Preparation (Milestones screen) — Enter the info.

Milestone No	Milestone Name	Work Package No	Lead Beneficiary	Means of Verification	Due Date (month)
M1	Stakeholder analysis	2	10 - FIBHULP	Presentation of results to Steering Committee	12
M2	Dissemination progress	2	10 - FIBHULP	Presentation of results to Steering Committee	24
M3	Indicators	3	13 - UHCZ	SMART indicators accepted by the Steering Committee	2
M4	WP Leaders' Workshop	3	13 - UHCZ	Workshop report	17
M5	National Policy Contact Point Group	4	17 - VULSK	(Confidential) renewable list of national policy contact points in the internal database of the JA	4
M6	Actual national governance models	5	16 - IOR	Report document	14
M7	First compendium of complementary indicators	5	16.1 - AOUP	Proposal document	19

M8	Report including recommendations from T6.6 and 6.7	6	1 - MUW	Report available	35
M9	Implementable solutions 1 prepared	8	21.3 - LUMC	Review by designated WP8 reviewers	12
M10	Implementable solutions 2 prepared	8	21.3 - LUMC	Review by designated WP8 reviewers	21
M11	Implementable solutions 3 prepared	8	21.3 - LUMC	Review by designated WP8 reviewers	30
M12	First version of adapted SE-Atlas released	8	6.6 - UK FFM	SE-Atlas accessible through Orphanet for at least one country	24
M13	Analysis completed	9	5 - GUH	Report available	24

LIST OF CRITICAL RISKS

Critical risks and risk management strategy

Grant Preparation (Critical Risks screen) — Enter the info.

Risk No	Description	Work Package No(s)	Proposed Mitigation Measures
1	Partner withdrawal from the JA	1-9	Prevention: tight collaboration between coordination and WP leads to recognize /avoid problems; close monitoring of all progress (laid down in consortium agreement) in order to be able to move tasks to another partner Remedy: find another partner to step in in case of actual withdrawal
2	Conflicts between partners	1-9	Prevention: open communication, clarifying tasks and objectives and the role of each partner Remedy: mediation by coordinator
3	Conflict of interest of participants serving potentially opposing	1-9	Prevention: clear role definition for survey completion and recommendation formulation Remedy: consultation with responsible authority, abstain from voting

	institutions		
4	Low quality of written deliverables	1-9	Prevention: continuous follow-up and feed-back by responsible WP leads Remedy: revision of documents by the steering committee
5	Delays in submitting deliverables	1-9	Prevention: continuous follow-up by WP leads, coordinator, and WP3 on the progress of WPs Remedy: support by the steering committee, withdraw funding from partner in case of failure to deliver
6	A meeting cannot be delivered face-to-face due to unforeseen external factors	1-9	Remedy: workshop/meeting will be converted to on-line format
7	Insufficient information, interest, participation, support from member states	1-9	Prevention: close monitoring of partners by WP leads and reporting to steering committee and coordination Remedy: consultation with National Policy Contact Points; continuous discussions with policy makers, ministries, other stakeholders on JA objectives
8	Low participation in planned surveys	4-9	Prevention: tight collaboration with coordination, competent authorities, National Policy Contact Points; careful choice of methodology (user-friendly, clearly formulated, motivatory introduction) Remedy: repeatedly approach stakeholders, contact stakeholders personally
9	Consensus among member states on recommendations and models difficult to achieve / not achieved	4-9	Prevention: keep partners informed through regular communication of progress; personal communication with partners; adaptation of recommendations considering factors stratifying MS Remedy: consensus finding repeated until common agreement is achieved
10	Implementation hindered by insufficient commitment or objection of MS	2, 4-9	Prevention: tight collaboration with coordination, competent authorities; contact the national policy contact point of the MS Remedy: selection of another MS for the pilot
11	Board of National Policy Contact Points incomplete (non-participation of some member states)	4	Prevention: encouragement for participation sent through ERN BoMS; participation of the relevant ERN BoMS member will be asked for Remedy: identification and recruitment of board member(s) repeated until a solution is found
12	Implementation steps not completed within the proposed timeframe	2, 4-9	Prevention: tight monitoring of implementation steps by WP leads, coordination, and steering committee Remedy: apply for no-cost extension of the JA

DESCRIPTION OF THE ACTION (PART B)

Part B of the Description of the Action (DoA) must be uploaded on the Portal Grant Preparation Documents screen.

PROJECT	
Project name:	Joint Action on integration of ERNs into national healthcare systems
Project acronym:	JARDIN
Coordinator contact:	Till Voigtländer, Medical University of Vienna

TABLE OF CONTENTS

HISTORY OF CHANGES

Changes based on requirements from the HaDEA evaluation

Hungary: Based on Council Implementing Decision (EU) 2022/2506, as of 16th December 2022, the Hungarian partners University of Debrecen (CA) and Semmelweis University (AE) are currently not eligible to receive Union funding.

The previous partners "28 Hungary - The University of Debrecen (UD; CA)" and "30 Hungary - Semmelweis University (SU; AE)" have been deleted from chapter "2.2 Consortium set-up" and have been replaced by a newly nominated and eligible Hungarian Competent Authority, the new partner "28 Hungary - The National Center for Public Health and Pharmacy (NCPHP; CA)". All subsequent partner institutions have been renumbered in chapter 2.2 accordingly.

In a further revision, also the initially remaining (and in the beginning second) Hungarian affiliated entity, the now previous partner "29 Hungary - The Bethesda Children's Hospital (BCH; AE)" that did not fall under the above mentioned Council Implementing Decision and therefore was in principle eligible as AE, decided for administrative reasons to withdraw as AE from the Joint Action and to participate in the project via a subcontract with the now only remaining Hungarian institution in the project, the NCPHP (see chapter "Subcontracting", no. S6.3, page 106). Consequently, all subsequent partner institutions were renumbered in chapter 2.2 accordingly a second time.

Switzerland: Switzerland is not eligible to participate as Associated Partner in the Joint Action.

Partner 61 Switzerland - Les Hôpitaux Universitaires de Genève (HUG; AP) – has been deleted from chapter "2.2 Consortium set-up".

Answers to the evaluators comments:

The JARDIN consortium highly appreciates the work of the evaluators and warmly thanks for their comments and suggestions. Below, we provide the answers and modifications to their open questions and remarks.

Criterion 2.1 Quality — Project design and implementation

1) None of the specific objectives in the project are SMART (Specific, Measurable, Achievable, Relevant, Time-bound).

We apologize that we did not explain the SMART characteristics of our specific objectives in our indicator table in chapter 2.5 ("Project management, quality assurance and monitoring and evaluation strategy") in sufficient detail. In addition, and this was in part related to the 70-page limit of the original proposal, the full alignment of the different specific objectives across the various chapters of the proposal (chapters 1.2, 2.5 and 4.2) was lost in some instances.

We have now thoroughly revised the indicator table in chapter 2.5, clarifying and redefining existing process, output and outcome/impact indicators and exchanging or introducing new indicators to better highlight the SMART characteristics of the specific objectives. We have also now fully aligned all specific objectives across the proposal, adding missing objectives in the aforementioned indicator table, and linking the indicators to the various tasks and deliverables in the different work packages. Furthermore, we have added several deliverables in chapter 4.2 ("Work packages, activities, resources and timing") to better underline the various activities of our project and their most important expected outputs.

2) The risk assessment overlooks the possibility of participating countries not willing to implement the pilots or adopt the ERNs.

While the risks highlighted by the evaluators are in principle very relevant for the project, we tried to minimize these risks already in the frame of the proposal preparation in close collaboration and communication with all partners concerned, including the following steps and strategies:

a) Pilot implementations were coordinated with and agreed to by the competent authority of the respective implementing country, and the competent authorities of each participating country are either the ministries of health themselves, or institutions designated by them.
b) Additionally, implementing institutions were advised during the preparation of the grant proposal to inform other responsible authorities in their country (if applicable) about the planned pilot and to obtain their agreement, if necessary (not all pilots involve topics within the competence of national authorities).

c) Of note, implementing institutions volunteered for the pilots, and competent authorities contribute financially to the activities within the project, including the pilots, so it can be assumed that it is in their interest that the implementations are performed, and are successful.
d) Furthermore, all Member States and Norway have now endorsed national healthcare providers to participate as full members in an ERN, i. e. accepted and adopted the ERN system in general.

However, to demonstrate that the consortium is prepared for the unlikely event that a piloting institution or country is no longer willing or able to implement the pilot, this risk was mentioned in the table in section 2.7 in Form B of the grant proposal (risk #10). We have adapted it to make the situation and the consequences clearer.

3) Impact indicators do not truly measure impact; they function as output or process indicators. The evaluation methodology is to be developed at a later date

The establishment of true impact indicators for JARDIN is indeed a challenge and we agree with the evaluators that the impact indicators defined in the first version of our proposal lack the necessary characteristics in the vast majority of cases. The reason for this was that true measurement of broad impact of implemented actions will not be possible within the frame of the project because there is typically no baseline and no control group available. Furthermore, we cannot realistically expect any measurable impact directly at the end of the project, as typically the establishment of pilot implementations will be finalized at the end date of JARDIN and the structures will only then have reached their full functionality. In addition, all our pilots will be tested at a smaller scale (and thereby developed) during the project (e. g. undiagnosed diseases programs) and will unfold their real potential only after the project has ended. What will be measurable at the end of JARDIN are the numbers of successful pilot implementations, of successfully implemented structures according to the project plan, and of positive decisions of the responsible stakeholders to maintain these structures and activities, so that a medium- and long-term impact can be anticipated.

In line with this perspective, we have now completely revised the outcome/impact indicators proposed in the table in chapter 2.5 ("Project management, quality assurance and monitoring and evaluation strategy") in Form B (please refer also to our reply to evaluators' comment "1"). Concrete actions to obtain the best results for these indicators will be developed during the project in the frame of work package 3.

Measures and indicators to monitor long-term impact beyond JARDIN can only be developed as recommendations during the running time of the project, while actual decisions are beyond our capacity and in the hands of other institutions. Proposed measures would be in close relation to sustainability measures, which will be developed in work package 4. We plan to propose to follow up on JARDIN after it has ended via the joint BoMS and ERNs Working Group on Integration, which will be revived after the end of the project (and is, in the meantime, put on hold, but its members are either direct participants in JARDIN or will be kept in close contact to JARDIN via participation in an additional body, the "ERN BoMS' Working Group on Integration Extension Group", which will be linked to the JARDIN Management Board to ensure continuous and comprehensive flow of information (as shown in the graph of the consortium structure (Figure 1), now shifted from the Other Annexes section of the grant proposal to the revised version of chapter 2.4 "Consortium management and decision-making").

4) The subcontracting amount for Bojana Raičković seems excessive, adding to the fact that her CV is not provided, and her expertise is unclear.

The calculation of the subcontracting for BR is based on her monthly costs and the number of person months in the project. Concretely, the monthly cost of BR amount to EUR 4.188,06 and the PMs planned for the whole project correspond to 10,293 PM. Multiplication of these figures (EUR 4.188,06 x 10,293) results in a subcontracting amount (gross price) for service contracts of EUR 43.106,00 and in a net price for service contracts of EUR 29.639,69.

Regarding the expertise of BR, we can now add her detailed Europass CV in a revised version of the annex "CVs and team descriptions JARDIN" to underline her experience in this area.

5) The travel and subsistence budget for MUW is rather high at over 50K euros.

Please note that the MUW budget for travel and subsistence includes the costs for 35 external participants, to be identified at a later stage of the project, to allow their participation in the third consortium meeting in Dublin (travel costs: 10 145,10 Euro; accommodation costs: 14 595,00 Euro; subsistence costs: 15 120,00 Euro). For more details on the participants and the calculation of the costs please refer to the budget table (MUW budget, Section C. Purchase costs, work package 1: 3 Consortium meeting Dublin, table on the right-hand side). The travel, accommodation and subsistence costs for the Austrian team members for WP4 just amount to 10.972 Euro.

6) A significant percentage of the budget is allocated to the main partner (almost 70% of the total personnel cost for WP1 alone).

The evaluators are correct that a significant part of the budget is allocated to the main partner. Within the proposed budget, around 50% of the total personnel costs are allocated to WP1 and the total personnel costs for all work packages (WP1-9) resemble around 70% of the total budget of the partner.

For WP1, these costs mainly reflect the requirements for managing a project of this size and complexity, also repeatedly communicated by HaDEA, with a coordination team consisting of a project coordinator (half time for all WPs, core of the workload in WP1), a full time project manager (almost exclusively for WP1), an accompanying full time junior scientist (as flexible support in all WPs, assisting the coordinator in this regard, and a core workload in WP1), and a full time financial manager. A small part of the budget is reserved for local legal support of the main partner for the preparation of all the necessary subcontracting contracts with subcontracted persons and legal bodies within and outside Austria. This strong coordination team has been set up by purpose (and in close communication with HaDEA) to ensure as good as possible that JARDIN delivers all of its planned activities in time.
Finally, a bit north of 10% of the budget is related to the requirement hiring an external evaluator for the final evaluation of the project (strongly recommended by and calculated with the help of HaDEA).

For WP7, the personnel costs are related to the co-lead of WP7, covering the content of specific objective 7.2 "To develop structures and procedures for undiagnosed patients closely linked to ERN on a national and European level", and about 1/6th of the WP budget for direct contracts for experts to be reimbursed for their work in the proposed national and ERN-based undiagnosed disease program pilots.

Personal costs in all other WP reflect the partial participation of the coordinating team (mainly the full time junior scientist, but also the project coordinator) to support the work in all WPs and to ensure a strong management of the project and leadership of the consortium.

7) Details on surveys, checklists, data collection tools, and methods, as well as stakeholder analysis, clinical guidelines, and communication tools are lacking.

We are currently exploring commercial project management tools to be used as our main project management and communication tool. One of the main criteria is to respect the GDPR. The tool will be made available for all partners at the beginning of the JA. For surveys, we plan to use the EUSurvey tool which is suggested by the EC and free for users once registered on the EC Funding & Tenders portal, and which fulfils all data protection requirements. Other, locally used, software (e. g. for data collection, calculation, visualization etc.) is at the discretion of the partners performing the tasks and will be chosen in the frame of the project. Likewise, the concrete design of methods for surveys, creation of checklists, stakeholder analyses etc. will be developed in the frame of the respective tasks. Of note, no clinical guidelines will be developed in this project.

8) The project is very complex, and the Gantt chart lacks details, making it difficult to determine if the project will be completed within the proposed timeframe.

In principle, we fully agree with the comment of the evaluators. However, when preparing the proposal we used the mandatory chart provided in the proposal form which does not allow a more detailed presentation. For internal usage, as well as for information purposes, we developed a detailed Gantt chart containing information on milestones and deliverables. We provide this Gantt chart (a) ahead of the standard timeline provided in Annex B and (b) in the Other annexes section (annex 5 to Part B) of the grant proposal.

9) The proposal is missing a robust management structure for a complex project with 60 partners from 29 different countries.

Due to the 70-page limit of the original proposal, we have split the description of the management structure of our consortium between chapter 2.4 ("Consortium management and decision making") and a supplementary figure 1 in the uploaded folder "Other Annexes" We apologize if this approach has blurred the description of how we plan to implement a robust management of the project.

Key management structures of JARDIN include (a) the Coordination (WP1) with a dedicated coordination team consisting of project coordinator, a full time project manager, an additional full time junior scientist, and a full time financial manager, (b) a Steering Committee, composed of all work package (co-)leads, (c) a Management Board, comprising all 60 partners of the consortium, and (d) a Multi-stakeholder Advisory Group with four dedicated subgroups (the National Policy Contact Point Group, the Hospital Managers Advisory Group, the Data Management Advisory Group and the Patient Advisory Group) and further stakeholders like ERN coordinators and other external experts that are not part of the JARDIN consortium. Within each work

package, WP (co-)leads will establish further management structures and procedures for their internal WP management.

The Coordination will be in charge of the "day-to-day management" of the consortium, combining management, information, and support functions for all partners and activities. In addition, the Coordination will keep close connections to DG SANTE and HaDEA. For the "day-to-day strategic requirements", including discussions concerning all WP and the proceedings and planning of the JA in general, the discussion of risks and risk mitigation, any minor strategic decisions, and the regular monitoring of the progress of all activities of the project according to schedule, the Coordination will be supported by the Steering Committee that is situated at the core of this first higher-order management level. The Steering Committee will also be involved in the preparation of major strategic decisions for the Management Board. The Management Board constitutes the second higher-order management level and will be responsible for the more long-term and important strategic decisions, as well as potential amendments of the project content, budgets and timelines. Management Board meetings will also serve as tool to disseminate all relevant information regarding JARDIN on a regular basis to all consortium partners and further stakeholders.

The Multi-stakeholder Advisory Group, positioned outside the internal management structures, will provide advice to the Coordination, the Steering Committee and the Management Board on a regular basis, while the afore mentioned subgroups of the multi-stakeholder advisory group will support the work of the different work packages in a targeted manner, when their specific expertise and advice is required and requested from the respective WP (co-)leads.

Key to the management of the consortium and its activities are close communication links and loops between all structures described above (see also figure 1). The main, regular communication and interaction arrangements include:

(a) Coordination:
- Participation of the coordinator (supported by the project manager or the additional full time junior scientist) in all WP meetings in order to have a precise overview of the proceedings at any given time.
- Release of a regular internal newsletter (every 6 month) by the coordinating team in cooperation with work package 2 and other partners. This newsletter will contain not only information of recent activities and achievements within the project, as well as upcoming events, but also information on tasks to be performed by the partners, important deadlines, administrative announcements etc. This format has proven useful in other EU projects (we rely in particular on the experience with the Orphanet project). It will be mandatory for the partners to read this newsletter.

(b) Steering Committee:
- Bimonthly meetings with the Coordination. In these meetings, work package (co-)leads, which have (as one central obligation) the task to keep track of the activities and contributions of all partners in their work package, will report back on the progress, any challenges and proposed solutions, as well as any potentially up-coming risks within their work package to the coordination. Apart from the information, discussion and decision functions, these regular meetings of the work package (co-)leads will further support the establishment of close links between all WPs (in addition to regular bilateral meetings between certain WPs/tasks with relevant overlaps, as described in the respective WP descriptions).

(c) Management Board:
- Bimonthly meetings, alternating with the meetings of the Steering Committee, providing information of all the activities and the main decisions, which have to be taken at this management level. The meetings of the Management Board will also be open to other participants beside the actual partners, in particular the "ERN Working Group on Integration Extension Group", comprising those BoMS representatives that were part of the BoMS Working Group on Integration (which is paused for the duration of the JA), but are not partners in the JA.

(d) Multi-stakeholder Advisory Group:
- Regular interactions with the full-scale Multi-stakeholder Advisory Group and targeted interactions between the different subgroups of the Multi-stakeholder Advisory Group and the related work packages for specific questions and needs. For further details, please refer to our reply to evaluators' comment "14".

This set of regular structured interactions will be supplemented by further interaction and communication activities whenever this deems necessary (for instance setting-up targeted teleconferences with single participants to solve any urgent issue or delivery of ad hoc notifications and other urgent information to all partners).

Finally, there will be regular updates on the JA in BoMS meetings, as well as in the ERN coordinators group meetings, although they will not be directly linked to the JA. In analogy to that, the JA will continue to

establish links to other groups, official bodies etc, including ones that might be identified only during the project and in individual WP.

We have now revised section 2.4 ("Consortium management and decision making") thoroughly to explain the management structure and all management activities in better detail.

10) Many of the mitigation strategies focus on avoiding problems rather than addressing them if they arise. Some strategies are vague and unconvincing.

Our original risk mitigation strategy was mainly based on risk prevention (which we still consider the best way to avoid unfavorable outcomes). In addition, as suggested by the reviewers, we have now updated the table of potential risks accordingly and added concrete remedies for those instances where an adverse event actually comes true.

Lastly; as risk management and mitigation is a continuous process, we will add any new specific risk in any WP that arises during the course of our project, to our listing of critical risks and will provide strategic measures to mitigate its impact accordingly.

11) The budget distribution among partners and personnel costs for some partners seem high.

The reason that the budget distribution varies between the different partners is based on the complexity of the project (as highlighted several times by the evaluators), resulting in different types of activities the partners carry out (merely country oriented versus additional project oriented efforts), and the different level of personnel costs between Member States. In more detail:

a) To manage a project of this size and complexity, several partners (in particular work package (co-)leads, but also task leads) took over further responsibilities on a voluntary basis and perform – in addition to their different implementation activities on the national level – general, project oriented activities that benefit all partners. This includes the additional workload for leading and coordinating work packages or tasks, as well as the workload for the main elaboration of deliverables (documents, blueprints, etc.) and all other types of work package output. Therefore, the workload of these partners is higher compared to partners that are primarily focussed on their national implementation activities, which in turn results in higher budget needs.

b) In addition, the personnel costs differ significantly between the 27 Member States and to some further degree between different partner institutions within the same Member State. We have asked all our partners to provide us with official figures from their institutions and rely on this information.

The combination of these two factors is the primary cause for the proposed budget distribution.

12) The complexity of the project raises concerns about whether the implementation steps can be delivered within the proposed timeframe, particularly since most will occur in the third year.

We agree with the evaluators that the majority of the implementation steps will occur in the third year and that specific measures have to be taken to ensure as good as possible that these implementation steps will be delivered in time. On the other side, the late start of implementation steps in the third year reflects the nature of our project. Our pilot implementations can only begin after completion of the development of the respective recommendations and toolboxes, which is typically towards Q4 of the second year of the project. This development, on the other hand, can typically only start after a thorough evaluation of existing best practices.

However, we will make sure that the preparation of implementation steps starts as early as possible and local contributors and stakeholders will be involved in the project already at an earlier stage. Furthermore, we are aiming to establish and maintain close contacts with national authorities and/or other relevant institutions. This includes those representatives from the health ministries that either function as competent authorities themselves or nominated the competent authority in their countries, as well as a further high level target person in each health ministry (the "National Policy Contact Point"; see also our reply to evaluators' comment "18") with close contacts to all the departments and divisions in their ministry that are responsible for the different topics addressed in our project to facilitate targeted interaction with these departments/divisions regarding the preparation of future national implementation steps. It further includes hospital managers as key actors at the hospital level.
To institutionalise these latter two contact strategies, national policy contact points, as well as the hospital managers, will constitute two specific subgroups of the advisory board that we will establish (see also figure 1 in the revised version of chapter 2.4 ("Consortium management and decision-making") and our reply to evaluators' comment "18"). Supported by this contact and communication strategy, a high commitment to implement the pilots can be assumed.

In addition, the tight monitoring of all project steps and developments by the coordination team, as well as the option to specifically support task and work package leaders by the coordination team (coordinator and second project scientist) when an acute need for help and further manpower arises will help as good as possible to keep track on all project activities and to deliver implementation steps in time (please also refer to the description of the robust management and communication strategy developed for our project, as outlined in our reply to evaluators' comment "9").

Lastly, we now have added the risk of not completing one or more implementation steps in the revised risk management table in Form B (chapter 2.7; risk #.12).

13) The rationale for choosing Italy, Romania, and Ireland for the testing of the pre-blueprint and pilot implementation of the indicators in WP5 is not well justified.

Ireland was chosen first as the only English-speaking member state, where the pre-blueprint can be tested without translation (the pre-blueprint will primarily be produced in English). Romania and Italy volunteered in addition as piloting countries, serving as examples for countries with different prerequisites (Romania with a mostly centralised healthcare system and first steps in the direction of more decentralisation, Italy with a highly federal healthcare system, dominated by the Italian regions; Romania with an average information level about rare diseases in the public and political area, Italy with a high information level and already specific legislation at baseline). Of note, countries for pilots were chosen on a voluntary basis throughout all work packages. For an explanation on the pilot implementations in work package 5.2, please refer to the column "Description" of task T5.10.

14) The interactions between the multi-stakeholder group and the steering committee or the management board are insufficiently described

We have omitted a more detailed description of these interactions in the original proposal due to the 70-page limit. What we provided was a brief overview of the consortium structure and the links between the different bodies, groups, and work packages, including a detailed graphical representation of these structures and links (for reference, please refer to figure 1, now integrated into the revised version of chapter 2.4 ("Consortium management and decision-making") and previously provided in page 1 of the Other Annexes section of the first submission of the grant proposal).

The interactions between the multi-stakeholder group and the different structures of the JARDIN consortium will take place at two different levels:

a) Basic regular interaction level:
The coordination team will contact the whole multi-stakeholder advisory group (the members of the four subgroups [i.e. the National Policy Contact Point Group, the Hospital Managers Advisory Group, the Data Management Advisory Group and the Patient Advisory Group] and the other involved stakeholders) approximately every 6 month starting from month 3, when the consortium and the project is fully operational (i.e. around M3, 9, 15, 20, 27 and 34) in order to provide regular information on the progress of the project and its activities and to ask for feedback on any open issue. A more comprehensive feedback will be requested in M 3, M 21 and M 34, each time after the main consortium meeting, to check the operational setup of the consortium and its functionality (M3) and to seek guidance regarding the mid-term results, as well as challenges and strategies ahead (in M20) and regarding the almost final results presented at the final meeting (in M34). All feedback from the advisory group will be reported to the steering committee and the management board. If useful and possible, members of the multi-stakeholder advisory group will also be invited after consultation to participate in subsequent steering committee and management board meetings. In case the steering committee or the management board identifies further items to be discussed with the multi-stakeholder advisory group, the coordination team will consult the advisory group accordingly in addition to the regular consultation intervals.

b) Targeted specific interaction level:
To support the work on well-defined elements in the different work packages, work package (co-)leads will consult the respective subgroup within the multi-stakeholder advisory group in a targeted fashion seeking advice regarding a particular aspect and/or inviting them to participate in specific meetings for this topic. This includes, for instance, targeted interactions between WP8 (Data management) and the Data management advisory group (red connection line in figure 1) or between the Hospital manager advisory group and WPs 6,7,8 and 9 (orange connection lines in figure 1).

Criterion 2.2 Quality — Project team and cooperation arrangements

15) The proposal lacks clarity when it comes to outlining the management and decision-making structure of the consortium, leaving appropriate procedures and strategies for cooperation largely undefined. While a voting system will be utilized by both the steering committee and management board to make decisions, the

actual decision-making process remains unclear. Additionally, it appears that the potential link between the steering committee and the scientific committee has been overlooked, further highlighting the need for a more comprehensive approach to consortium management.

As outlined in our reply to evaluators' comment "9", we apologize that our procedure splitting the description of the management structure of our consortium between chapter 2.4 ("Consortium management and decision making") and a supplementary figure 1 in the uploaded folder "Other Annexes" has blurred our approach to implementing a robust management of the project.

Regarding the management structures of JARDIN, please refer to the detailed description in our reply on evaluators' comment "9". Regarding the voting system, the following rules will apply for the steering committee, as well as for the management board:

a) Voting rights:

In the steering committee, the coordinator and each work package (co-)lead have one vote. Based on 12 members of the steering committee, the maximum total number of votes will be 12. In the management board, each member state with a competent authority will have one vote. This voting right will primarily be exercised by the competent authority, but any country with additional affiliated entities in the consortium can delegate the voting power to one of these other national representatives in the board. Based on 27 European Member States and Norway, the maximum total number of votes will be 28.

b) Minimum number of participants and minimum number of votes for one voting option for successful voting:

Generally, we strive to reach unanimous agreements for every topic a voting on the level of the steering committee or the management board will be necessary. If this is not possible for a certain topic, the following rules will apply:
- For any voting decision, the minimum number of participants with voting power present in the respective meeting should be ≥ 70% (i.e. steering committee ≥ 9 members and management board ≥ 20 participants). If this quorum cannot be reached in a given meeting, the voting has either to be postponed to the next meeting or it has to be executed in written format;
- For any voting to be valid on the level of the steering committee or the management board, the minimum number of votes for one specific voting option should be ≥ 75 %. If this quorum cannot be reached, the respective voting option is rejected and other solutions have to be developed.

Criterion 3 Impact

16) Although the proposed timeframe for the project is considered feasible, there is a lack of compelling evidence to support the notion that the project will be completed within the allotted three-year period. This suggests that the project may be overly ambitious, which raises concerns about its timely completion.

To better demonstrate the activities and their individual timeline within the project within the given timeframe, we are providing in Annex B and in the the Other annexes section (annex 5 to Part B) an extended version of the standard Gantt chart in Form B of the proposal (see also our reply to evaluators' comment "8"). Strict adherence to this timeline will be achieved by tight monitoring of the progress within all work packages by the coordination together with the work package (co-)leads. Checklists will be employed by the coordination to have an overview of the status of all deliverables and milestones at any given time during the project, and the coordinator will be present in all work package meetings (please refer also to our reply to evaluators' comment "9").

To highlight the fact that we are aware of the challenges associated with completing our activities in time, we have now also included the risk of not finishing in time in the revised and extended risk management table.

17) While the dissemination strategy is based on thorough stakeholder analysis and is reasonably well-documented, it does not include quantifiable indicators for measuring its success. Additionally, the proposed dissemination/exploitation plan lacks specific targets for social media reach or other tangible metrics. Further, the plan does not provide sufficient details on standard measures to communicate the project's activities to different target audiences.

Again, we thank for the evaluators' comments and suggestions and we apologize for the lack of quantifiable targets in our proposal that is in part again related to the page limits of the proposal form that we wanted to keep by all means.

We have reworked chapter 2.5 "Project management, quality assurance and monitoring and evaluation strategy" for our work package (WP2) extensively, adding the two missing specific objectives mentioned in the introduction and the WP description and refining the indicators including some quantifiable indicators

(either described as "direct indicators" or as part of surveys that will provide quantifiable indicators/results in the form of an evaluation report). The latter is in particular relevant for the evaluation of the pilot implementation of the blueprint (T2.7), we have therefore adapted the wording in the task description as follows: "The evaluation phase would be on M25 - M30, in collaboration with WP3 (including surveys and a pilot evaluation report)".

As indicated in the work plan for WP2 (T2.6: "Measurement and evaluation of communications activity"), we will closely collaborate with WP3 to develop further indicators for the evaluation of the dissemination activities. This task will be led by WP2 and WP3 coordinators from the start of the Joint Action. We have modified the description in T2.6 to better highlight this cooperation (the new version reads as follows: "Develop further qualitative and quantifiable communications evaluation indicators (apart from those outlined in chapter 2.5) for all WP2 activities in close cooperation with WP3 (shared task lead) from the beginning of the joint action.").

Planed quantifiable indicators for the dissemination activities will focus on:
a) JARDIN website use indicators dedicated to website traffic dataset. The software or web analytics platform should comply with GDPR. We will use standard indicators such as: number of new visitors, number of returning visitors, source of traffic, sessions by countries, and most visited webpages (homepage, patient/families, ERNs, professionals, etc). The first two indicators have now been added to the indicators table in chapter 2.5 "Project management, quality assurance and monitoring and evaluation strategy", the other indicators will be defined early in the project as soon as the final build of the website is available. The last two indicators will be useful in measuring the impact by different target audiences.
b) JARDIN social media indicators, for instance: numbers of click and shares of the social media posts (at least by half of the ERNs accounts). As indicated in chapter 3.2 "Communication, dissemination and visibility" in our proposal, we will hire a team of C&D professionals (specialized journalists with management of social media and corporate communication) and we would like to benefit also from their experience when defining the best social media channels to be used for the different target audiences (current thoughts include LinkedIn towards professionals, TikTok towards patient organizations). Therefore, we deliberately did not provide any quantifiable indicators for social media reach in our proposal.
c) JARDIN webinar indicators, e.g. number of dissemination webinars dedicated to increase the awareness and number of attendees. Also these indicators will be developed in the beginning of the project.

In the context of the stakeholder analysis, we will conduct national stakeholder meetings that should cover at least clinicians and patients' groups. We expect meetings to be held in at least 90% of the EU-MS plus Norway. We also plan ERNs communications workshops/meetings in which we expect a participation of at least 90% of the ERNs.

As indicated above when describing the close cooperation between WP2 and WP3, further indicators will be provided during the execution of the Joint Action, and will be described defining the WP, the area/task, the indicator(s), the baseline (if applicable), and the targets.

18) Regarding sustainability, the proposal includes a dedicated work package (WP4) focused on ensuring the longevity of the project's results. However, the proposed actions do not directly ensure the sustainability of the project beyond EU funding.
The implementation and sustainability of the project will be ensured through ERN BoMS, but the proposed sustainability models and solutions are insufficiently demonstrated.

We agree with the evaluators that sustainability of our activities beyond the duration of JARDIN is of critical importance not only for the value and success of the project but also the sustainability of the ERN system across Europe as such for the forthcoming decade.

However, we have to clarify right from the start that the JARDIN consortium, even though composed of one competent authority per member state, each being nominated by its relevant national health authority, has no decision power at all in any MS at any level. Therefore, we do not have any means of directly ensuring the sustainability of our actions on the member states' national level after our project has finished, this is outside the legal competence of the consortium. What our project can provide are measures to indirectly promote the sustainability of the project activities after termination of JARDIN. These measures can be subdivided into structural elements on the European and national level and into activity/output elements from the different work packages.

The major structures to ensure sustainability at a European level are:

a) The Board of Member States for ERNs with its "Working Group on ERN Integration into National Systems" that was set-up by the Board in 2016 and later also joined by several ERN coordinators. In the past, this working group on integration prepared key documents our project will be based on. Its activities are paused during the preparation and duration of JARDIN, but it will be re-established after our project has finished and

is intended to continue some of the activities of JARDIN within the frame of the BoMS including continuation and up-dating of the JARDIN website with all its elaborated documents and monitoring of the ERN integration process based on the national integration indicators developed in work package 5. Many members of the working group are direct partners in JARDIN, and those not participating as CA or AE in the project will be linked to JARDIN at the management board level via an additional advisory group, the "ERN Working Group on Integration Extension Group" (for further information see our reply to evaluators' comment "9").
Of note, the Board of Member States, being a continuous body, legally based on the Commission Implementing Decision 2014/287/EU, consisting of national representatives from all EU/EEA Member States, delegated by relevant authorities (Ministry of Health or any other relevant), and from the EC, exerts its supportive sustainability function also on the national level (see below).

b) The ERNs which are instrumental for the continuation of certain pilots foreseen in this JA as a proof-of-concept (including the development of reference care pathways for further adoption/ adaptation in the national systems, the ERN framework for solving undiagnosed cases and measures for FAIRification and interaction of ERN datasets with national/regional datasets).

The main structures for the sustainability at the national level are:

a) The Board of Member States for ERNs with its linking function to the national health authorities (see above).

b) The "National Policy Contact Point Group", a subgroup of the multi-stakeholder advisory group (see reply to evaluators' comment "9") that will be set-up specifically for the actions of this JA. It will be composed of one higher level representative of the Health Ministry in each Member State with an overview of and close links to all the divisions within the ministries that are targeted by the different activities of our project. The profile of the members of this group ("higher level") should include a certain level of policy mandate and capacity. All sustainable elements of WPs 2 and 5-9 with relevance to the health authorities will be developed in close coordination with the National Policy Contact Point group, introducing all relevant topics early during the term of the JA into the health policy discussion level and planning in all member states, thereby ensuring as good as possible the national sustainability and further implementation of the projects' activities and results.

c) The "Hospital Manager Advisory Group", a further subgroup of the multi-stakeholder advisory group (see reply to evaluators' comment "9") that will also be set-up specifically for the actions of this JA. It comprises the hospital managers of all ERN coordination centres, as well as one hospital manager representative from every participating member state not covered by the hosting an ERN coordination centre. Similar to the previous subgroup, the Hospital Manager Advisory group will be involved in the development of all sustainable elements falling into the decision competence and implementation power of hospital managers in all partner countries, again aiming to ensure as good as possible the national sustainability and further implementation of the projects' activities and results.

On the work package level, the sustainability of the projects' activities and results will be promoted via the several channels including:

a) WP2:
- the sustainability of the blueprint for a national dissemination strategy on ERNs with the elaboration of specific materials, as leaflets, videos, etc., will be promoted through the placement of these materials onto the websites of ERNs, HCPs that belong to the ERNs or comprise national networks, national authorities/ relevant institutions and the EC and will be available beyond the project duration.

b) WP4:
- sustainable elements of the six relevant WPs (2, 5-9) will be developed together with the National Policy Contact Point group to set the grounds for further implementation beyond the duration of the project;
- two strategic concept papers (D4.1 "Compendium of sustainability models and solutions"; D4.2 "Recommendations for better integration of JA actions and sustainable elements into the National RD Plans / Strategies") will be widely disseminated through European and national conferences and meetings, including a workshop involving multinational multi-stakeholder communities under WP4, and placed onto the websites of ERNs, national authorities/ relevant institutions and EC, where relevant.

c) WP5:
- a framework for the collection of national monitoring data will be piloted in selected countries and included into the existing ERN monitoring system for the implementation across all EU and EEA Member States.

d) WP6:
-model (reference) care pathways for RD or groups of RD, developed by ERNs, will be placed onto ERN websites;
- besides, these model care pathways will serve as a proof-of-concept for the further development of model

care pathways beyond the duration of the JA;
- recommendations and guidelines for the implementation of care pathways will be published in the websites of ERNs, HCPs that belong to the ERNs or comprise national networks, national authorities / relevant institutions and EC and will be available beyond the project duration (including D6.2 "Compendium (blueprint) of model care pathways for RD or groups of RD", D6.3 "Recommendations for the organisation of national care pathways, referral systems to ERNs and incorporation of CPMS advice for rare and complex diseases" and D6.4 "Toolkit of best practices in the implementation of care pathways").

e) WP7:
- an expert panel for undiagnosed cases comprising all 24 ERN will be incorporated into day-to-day activities of ERNs, including supplementation of CPMS system;
- an SOP for assigning ORPHA code 616874 (undiagnosed RD) will be implemented in ERNs, HCPs that are members of ERNs and other Centres of Excellence in the national rare disease networks and will be used beyond the duration of the JA.

f) WP8:
- a visualisation of RD Expert Centres with ERNs and NRNs will be developed based on a harmonization of the Orphanet and SE-ATLAS data sets and data models, and subsequently implemented in interested pilot countries via individual national search start pages per country. Depending on its evaluation, success and national interest, this pilot visualisation could be available beyond the duration of the JA and could also be extended to other partner countries;
- a key recommendation (D8.2 "Recommendations for integration of data management between National Health Systems and ERN") will be placed onto the websites of ERNs, HCPs that belong to the ERNs or comprise national networks, national authorities / relevant institutions and EC and will be available beyond the project duration.

g) WP9:
- best practice and guidance documents (including D9.1 "Report on existing mechanisms to support ERN-related activities of ERN-HCPs in MS", D9.2 "Guidance document on the requirements for national support of ERN-HCPs" and D9.3 "Recommendations for CPMS reimbursement models") will be placed onto the websites of ERNs, HCPs that belong to the ERNs or comprise national networks, national authorities / relevant institutions and EC and will be available beyond the project duration.

The broad dissemination of the project activities and outputs via different channels should help to generate a public interest among various stakeholders in each member state in the project achievements and their continuation, creating a supportive framework to promote the further implementation and sustainability of the project results on a national level. One element in this context is the elaboration and implementation of new or revised national action plans or strategies for rare diseases, incorporating all the relevant documents, results and pilot implementations from JARDIN, a development that the JA should help to initiate in each member state towards the end of the project, further strengthening the sustainability of all achievements.

19) Lastly, the proposal lacks evidence and indicators to monitor the effectiveness of all interventions in each work package, as well as the short- and medium-term impacts of rare diseases.

The indicator table in chapter 2.5 ("Project management, quality assurance and monitoring and evaluation strategy") has been adjusted accordingly to assess expected impacts as far as possible within the frame of the project (please refer also to our reply to evaluators' comment "4" for detailed discussion).

20) Some of the proposed activities have already been carried out by different national/international organizations, and the long-term impacts and sustainability of the project may be overly ambitious for a single proposal.

We are not sure to which concrete national/international activities the evaluators are referring to. Regarding the European level, we made sure during the preparation of our proposal that no duplications with other EU-funded projects will arise at any point within JARDIN, while on the other hand achieving as much complementarity as possible. This complementarity is laid out in section 1.3 ("Complementarity with other actions and innovation — European added value") of the proposal.

Looking at the national level, we know and agree that some structures have already been built up in some member states, albeit often to a different level of maturity compared to that level we would like to reach within our project. We consider this fact a particular strength and clear advantage of JARDIN, since, wherever possible, we will use these structures as good practice examples for developing the recommendations and toolkits for all other countries. In some cases, participants from countries who already have implemented certain measures will be work package (co-)leads in JARDIN, thus providing their expertise from a prominent position in our project, ensuring that the other partners will be able to profit to a maximum extent from their experience (e. g. France is (co-)leading work package 8 on data management, the French Ministry of Health

also being responsible for the exemplary RD registry established in this country). Likewise, coordinators of other EU-funded projects will contribute as work package (co-)leads or task contributors (e. g. Orphanet, Solve-RD). In addition, experts at implemented structures will serve as senior partners in twinning activities (e. g. the Center for rare and undiagnosed cases in Würzburg, Germany). This strength is made possible by the broadness of our consortium, mirrored by the inclusion of a wide variety of experts and institutions all across Europe.

We agree that our proposal is very ambitious but this reflects the high expectations on the European level that were communicated to us during the proposal preparation period. As outlined in our replies to evaluators' comments "3", "12" and "18", we have developed and proposed many measures that will help us to ensure as good as possible the long term impact and the sustainability of the activities initiated during the project time.

#@APP-FORM-EU4H@#
#@PRJ-SUM-PS@# [This document is tagged. Do not delete the tags; they are needed for the processing.]

PROJECT SUMMARY

Project summary
See Abstract (Application Form Part A).

#§PRJ-SUM-PS§# #@REL-EVA-RE@# #@PRJ-OBJ-PO@#

1. RELEVANCE

1.1 Background and general objectives

Background and general objectives
Describe the background and rationale of the project. *How is the project relevant to the scope of the call? How does the project address the general objectives of the call? What is the project's contribution to the priorities of the call?*
More than 6,000 rare diseases (RD) are known to date. While patients with each of these conditions are inherently not numerous (the definition being that a RD affects no more than 5 in 10,000 people in Europe), the collective disease burden is high, with an estimate of 30 million people living with a RD in Europe [1], making effective, concerted measures for diagnosis and treatment a necessity. However, RD patients are typically scattered across countries and regions, rendering the acquisition of knowledge and experience difficult, and expertise on RD is generally scarce and fragmented. Therefore, centralization is key for RD, with regard to expertise, as well as equipment and infrastructure. The EU recognized and acknowledged this fact several years ago and – in a process which eventually took more than a decade – developed the European Reference Networks (ERN) model as a voluntary, coordinated cooperation between all member states (MS) in the field of highly specialized health services, thereby generating immense added value for European citizens. The currently 24 ERN were established in March 2017 in accordance with Article 12 of Directive 2011/24/EU of the European Parliament and of the Council of March 9, 2011, on patients' rights in cross-border healthcare (Cross-border healthcare Directive, CBHD) [3]. They are multinational networks of highly specialized healthcare providers (HCP) across Europe in thematically coherent medical domains, addressing rare or low-prevalence complex diseases requiring exceptional concentration of expertise and resources, and designed to improve patients' access to high-quality healthcare and guarantee equity of care by providing virtual expert consultations for patients from all member states (MS) and Norway, as well as support knowledge generation, professional training and education, and research. The governance body of the ERN system is the Board of the Member States for ERN (BoMS), which is composed of national authorities nominated by each MS. After its constitution, the BoMS set up several working groups to address specific topics where improvements and solutions would be needed to guarantee the proper functioning and enhance the utility of the ERN, including the Working Group on Integration of ERN into national health systems. Its mandate was to steer the process of ERN integration and propose solutions to the BoMS and MS. Based on the input of this working group, the BoMS issued a statement in 2019, stressing that "to ensure a proper and sustainable functioning of the ERN and to reap all benefits for patients suffering from rare and low-prevalence complex diseases across the EU, the ERN need to be linked in a clear and stable way to the healthcare systems of the MS." [3] MS should be encouraged to facilitate the integration of ERN into their healthcare systems by a proposed set of measures (refer to section 1.2 for a detailed description) with the overall objective to facilitate access to timely diagnosis and high-quality and cost-effective healthcare for all patients with rare or low-prevalence complex diseases via the possibilities the ERN are offering, always in accordance with each MS' national legislation [3]. In addition, in 2018, the European Court of Auditors (ECA) conducted an audit on the effectiveness of cross-border healthcare in the EU and, in its 2019 report, concluded that the ERN face significant challenges to ensure their financial sustainability and ability to operate effectively within and across national healthcare systems [4]. Likewise, better integration of ERN into national healthcare systems was identified as one of the challenges for the ERN system in the frame of an open public consultation by the EC to evaluate the CBHD in 2021. 49% of the respondents indicated that insufficient integration of ERN in the national health system and lack of support for their activities by the national authorities were among the biggest barriers for HCP in accessing the expertise of the ERN. For patients, lack of awareness/information (62%) and absence of a clear pathway to refer patients to ERN (41%) were considered to be some of

the biggest issues [5]. EURORDIS responded to the consultation with a position paper mentioning that "the lack of an action plan by each MS to formally connect the Networks with their own health systems as well as the lack of a full EU service model definition for the ERNs virtual expert advice has substantially limited their impact." [6]
Therefore, in line with the general objective of the call within the 2022 work programme of the Programme for the Union's action in the field of health (EU4Health), area of action "Enhanced European Reference Networks" [7], this Joint action (JA) aims to improve the accessibility and support the long-term sustainability of the ERN system by contributing to the effective integration of ERN in the national health systems (while always respecting the autonomy of MS in this regard), thereby, on the other hand, also strengthening the resilience of the national health systems according to Regulation (EU) 2021/522 [8]. This will be achieved through the specific objectives defined in Article 4, points (f) (strengthening the use and re-use of health data, (g) (enhancing access to quality, patient-centred, outcome-based healthcare), and (i) (supporting integrated work among MS).
Activities will include identification and exchange of best practices, development of concrete recommendations, guidelines, and toolboxes suited for the needs of all MS (taking into account the different preconditions, such as size and population, economy, and structure of the respective healthcare system), as well as support of capacity building and performing pilot implementation steps on different levels in MS in the proposed fields of action. The latter will comprise national governance and quality assurance models, developing national care pathways and referral systems to the ERN, national reference networks (NRN) and undiagnosed disease programs (UDP) or equivalent strategies interlinked with ERN, data management, national support options for ERN-HCP, and dissemination of information on ERN, as well as structures and activities related and linked to them. A main focus will be to ensure the sustainability of the proposed actions and implementations, which we propose to integrate into the national plans and strategies for RD in the MS. All developments will be based on and/or take into account the preparatory and ongoing work by other relevant European and international projects and initiatives (refer also to section 1.3).
References: [1] Nguengang Wakap et al, European Journal of Human Genetics 28(2):165-173 (2020); [2] Directive of the European Parliament and of the Council of 2011/24/EU of 9 March 2011 on the application of patients' rights in cross-border healthcare (OJ L 88, 4.4.2011, p. 45–65); [3] Statement of the ERN Board of Member States on Integration of the European Reference Networks to the healthcare systems of Member States (adopted Statement on 25 June 2019); [4] European Court of Auditors, Special Report No 7/2019: EU actions for cross-border healthcare: significant ambitions but improved management required; [5] European Commission, 'Evaluation of patient rights in cross-border healthcare: Public Consultation Factual Report' Ref. Ares(2021)6103901 - 07/10/2021; [6] EURORDIS - Rare Diseases Europe responds to the evaluation of patients' rights in cross-border healthcare - and provides recommendations to improve the system: "An empty promise: accessing cross-border healthcare for people living with a rare disease" (July 2021); [7] Annex I to the Commission Implementing Decision of 14.1 .2022 on the financing of the Programme for the Union's action in the field of health ('EU4Health Programme') and the adoption of the work programme for 2022; [8] Regulation (EU) 2021/522 of the European Parliament and of the Council of 24 March 2021 establishing a Programme for the Union's action in the field of health ('EU4Health Programme') for the period 2021-2027, and repealing Regulation (EU) No 282/2014 (OJ L 107, 26.3.2021, p. 1-29).

1.2 Needs analysis and specific objectives

Needs analysis and specific objectives

Describe how the objectives of the project are based on a sound needs analysis in line with the specific objectives of the call. What issue/challenge/gap does the project aim to address?

The objectives should be clear, measureable, realistic and achievable within the duration of the project. For each objective, define appropriate indicators for measuring achievement (including a unit of measurement, baseline value and target value).

As mentioned above, inadequate integration into national healthcare systems has been identified as one of the major issues in the context of the challenges the ERN are facing with respect to their long-term sustainability and functionality. The following propositions were brought forward to amend this:
• The BoMS proposed in its Statement on Integration of the European Reference Networks to the healthcare systems of Member States of 2019 the following measures: a) assessing and, if necessary, adapting their national policies and/or legal framework (such as, for example, national action plans or strategies for RD); b) establishing well-defined patient pathways; c) developing transparent, seamless, and effective procedures for referral to ERN; d) developing a clear strategy for disseminating information on ERN to HCP in their country, as well as facilitating access for healthcare professionals to the knowledge generated by the ERN (such as clinical guidelines, training materials etc.); and e) reflecting

on the means to best support (administratively, financially etc.) HCP participating in ERN on a national level and strengthening the coordinating role of national authorities [1].
• The ECA, as a result of the 2018 audit on the effectiveness of cross-border healthcare in the EU, recommended the Commission to improve support to MS to facilitate patients' access to treatment for rare diseases (in particular by continuing the development of ERNs), in addition to better preparation for cross border exchanges of health data, which also involves the ERN [2]. In its response, the Council acknowledged this and encouraged the Commission and the MS "to continue to cooperate closely towards the full implementation of the CBHD by […] providing further support to the development of the ERNs", among other concrete measures [3]. The Commission accepted the respective recommendation of the ECA and stated that it was "fully committed to supporting the MS and the ERN" [2].
• In 2020, EURORDIS issued their "Recommendations to achieve a mature ERN system in 2030", stating that MS should focus on the national dimension of the ERN and integrate the ERN system into their national health systems; a mature ERN system should be "a network of expert centers, connected to local healthcare services, creating together an ecosystem". Concrete recommendations included updating of national policy and/or legal framework, defining clear patients' pathways and referral procedures to CoE and ERN, developing strategies to disseminate information about ERN, establishing national reference networks, facilitating access to the knowledge assets generated by the ERN, assessment and adoption of ERN guidelines and other clinical decision support tools, and supporting ERN members (administratively, financially etc.) [4]. They also mentioned the "need for sustainable programs dedicated specifically for undiagnosed diseases to enable rapid and equitable access to diagnosis and social support", which was based on the International Joint Recommendations to Address the Specific Needs of the Undiagnosed Rare Disease Patients, developed in 2016 by SWAN UK, the Wilhelm Foundation, EURORDIS, Rare Voices Australia (RVA), the Canadian Organization for Rare Disorders (CORD), the Advocacy Service for Rare and Intractable Diseases' stakeholders in Japan (ASrid), and the National Organization for Rare Disorders (NORD) [5].
• The 2021 recommendations from the Rare 2030 foresight study mentioned under recommendation 3 "Access to High-Quality Healthcare" that the EC should support MS and EEA countries to implement the actions outlined in the BoMS statement of 2019; specific actions listed in terms of greater support to – and integration of – ERN were renewing/updating national RD plans and strategies, updating national designation processes for CoE for RD, defining ERN referral pathways, and disseminating and utilizing the knowledge and evidence generated by the ERN (such as clinical practice guidelines), among others. Also, ERN should be supported in collecting and using findable, accessible, interoperable, and reusable data [6].
• In their standalone response to the 2021 public consultation to evaluate patient rights in cross-border healthcare, EURORDIS reinforced their recommendation to ensure the sustainability and consolidation of the ERNs by integrating them into national health systems and urged the MS to take action to implement the integration statement issued by the BoMS. They underlined the necessity of a defined reimbursement model for the ERNs' expert virtual advice in order to be able to scale up this service, in addition to clear, publicly available referral pathways in each MS, formal integration of ERNs' referral in the national referral protocols, agreement on clinical governance standards for quality assurance, integration of the service into the hospitals' clinical workflow and IT infrastructure, and training of the healthcare workforce as well as their National Contact Points for cross-border healthcare on the referral process [7].
Considering all the aforementioned inputs and aspects, and taking into account the objectives scope, and activities laid out in the call within the EU4Health 2022 work programme, we identified the following specific objectives as pivotal in order to achieve the overall goal of creating an integrated, seamless, and comprehensive system of ERN-HCP and national healthcare services (objectives continuously numbered linked to work package (WP); the obligatory, transversal WP 1 and 3 are not listed here):
2.1 To achieve efficient and effective visibility, awareness, and acceptance of the JA to internal and external stakeholders
2.2 To support national ERN-specific dissemination activities
2.3 To develop a blueprint for national dissemination strategies on ERNs
4.1 To develop the JA sustainability strategy including a) sustainability of JA actions at MS level and b) mechanism for sustainability/accountability at the EU level
4.2 To support capacity building in MS for the elaboration of new/updated National Plans / Strategies for RD (in terms of sustainability of JA actions)
5.1 To develop proposals for national governance models and practices for rare and complex disease HCPs and care pathways, fully interoperable with ERNs
5.2 To develop a proposal for national quality assurance models for rare and complex diseases
6.1 To develop recommendations for the organisation of national care pathways for rare and complex diseases interfacing with ERNs, including the recognition of and preferably full compliance with ERN-elaborated evidence-based resources (like Clinical Practice Guidelines)
6.2 To develop a proposal for referral systems to ERNs
6.3 To develop guidelines for the incorporation of CPMS advice into patients' care
7.1 To support capacity building in MS for the development of NRN or equivalent strategies for rare and complex diseases and their integration with ERN

7.2 To develop structures and procedures for undiagnosed patients closely linked to ERN on a national and European level
8.1 To develop recommendations ensuring the interoperability of data structures on MS level (local, regional, national) and ERN level
9.1 To collect and analyse good practices and mechanisms to provide support to ERN-hosting healthcare providers at national level as well as to individual ERN centres at the hospital (HCP) level
9.2 To develop specific recommendations for: 1. the national support to healthcare providers participating in ERNs, 2. the hospital support to individual ERN centres, 3. the CPMS service and reimbursement models
For each of the recommendations, strategies, and guidelines developed by this JA, as well as for the pilot implementation steps, a mechanism for monitoring the progress and implementation will be established (refer to section 2.5 and WP descriptions).
References: [1] Statement of the ERN Board of Member States on Integration of the European Reference Networks to the healthcare systems of Member States (adopted Statement on 25 June 2019); [2] European Court of Auditors, Special Report No 7/2019: EU actions for cross-border healthcare: significant ambitions but improved management required; [3] Council of the European Union, Draft Council conclusions in response to the European Court of Auditors' Special Report No 07/2019: "EU actions for cross-border healthcare: significant ambitions but improved management required"; [4] EURORDIS - Rare Diseases Europe, "Recommendations to achieve a mature ERN system in 2030" (December 2020); [5] SWAN UK, Wilhelm Foundation, EURORDIS - Rare Diseases Europe, Rare Voices Australia, CORD, and Advocacy Service for Rare and Intractable Diseases' stakeholders in Japan, "International joint recommendations to address specific needs of undiagnosed rare disease patients" (2016); [6] Recommendations from the Rare 2030 foresight study "The future of rare diseases starts today" (February 2021); [7] EURORDIS - Rare Diseases Europe responds to the evaluation of patients' rights in cross-border healthcare - and provides recommendations to improve the system: "An empty promise: accessing cross-border healthcare for people living with a rare disease" (July 2021).

#@COM-PLE-CP@#

1.3 Complementarity with other actions and innovation — European added value

Complementarity with other actions and innovation

Explain how the project builds on the results of past activities carried out in the field and describe its innovative aspects. Explain how the activities are complementary to other activities carried out by other organisations.

Illustrate the European dimension of the activities: trans-national dimension of the project; impact/interest for a number of EU countries; possibility to use the results in other countries, potential to develop mutual trust/cross-border cooperation among EU countries, etc.

Which countries will benefit from the project (directly and indirectly)? Where will the activities take place?

This JA will build on results and established structures of a number of past projects and actions and will use and create strong synergies with current ones wherever possible. Apart from the ERN project itself, these include in particular (in alphabetical order):
EHDS Pilot 2 (HealthData@EU Pilot) (https://www.ehds2pilot.eu) is the pilot implementation phase of the European Health Data Space (EHDS) aimed at providing a federated infrastructure of Data permit authorities and health and research infrastructures in order to enable linking and integrating data between data sources for secondary use of health data. Several partners in the JA also involved in HealthData@EU Pilot will ensure that data on RD produced at national level is ready for secondary use in EHDS.
European Joint Programme on Rare Diseases (EJP RD, 2019-2023; www.ejprarediseases.org) is an EU-wide RD-research oriented programme aiming to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care, and medical innovation. EJP RD builds on coordinated research funding, a federated data ecosystem infrastructure, capacity building and innovative methodologies for research and innovation. Many partners in this JA, not least ERNs, will ensure that EJP RD outputs, in particular data management solutions, are taken up.
The European Rare Disease Registry Infrastructure (ERDRI) provides an overview and interface of rare disease patient registration applications in Europe and offers services among which the generic pseudonymization tool (ERDRI.spider) and the metadata repository (ERDRI.mdr) will be utilized. Members of the JA consortium have been involved in developing the platform. The project will closely cooperate with the Directorate General Joint Research Centre to build on the ERDRI achievements and extend these in the direction of registration of undiagnosed patients.
European Rare Disease Research Coordination and Support Action (ERICA, 2021-2025; www.erica-rd.eu): all 24 ERN, as well as EURORDIS, Orphanet, EJP-RD, and EATRIS participate in this H2020-funded project, which aims to integrate clinical and translational research activities across the ERN. In

the WP for data integration and sharing, experts involved in the ERN registries work on ethicolegal aspects, data collection, interoperability, and data re-use for research. The JA (specifically WP8) will build on the operational documents, legal forms, data access policy, and data management strategy developed by ERICA.
IRDiRC (International Rare Diseases Research Consortium; https://irdirc.org/) is a global collaborative initiative launched in 2011 by the EC and the US National Institutes of Health to tackle RD through research and accomplish the vision to enable all people living with a RD to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention. This JA will contribute to the goals of IRDiRC and will be complementary to the actions of some Task Forces as TF on Primary Care and TF on Telehealth.
OD4RD and OD4RD2 (2022-2023, 2023-2025; www.orpha.net): Orphanet, which is currently co-funded via the OD4RD (from April 2023: OD4RD2) grant, produces and maintains the Orphanet nomenclature and classification of RD. Its main focus is now to collaborate with ERN to systematically revise and update the classification, and to facilitate its implementation in health information systems across Europe. The coordinating site at INSERM is part of this JA and will actively contribute to several tasks.
Solve-RD (2018-2023; www.solve-rd.eu) is a H2020-funded research program for solving the unsolved RD. Six ERN including more than 200 clinical centers are the main building block of Solve-RD. One main pillar of activities has focused on scientific cutting-edge re-analysis and re-interpretation of more than 20,000 existing exome and genome data sets (genomic, phenotypic and pedigree data). To implement this pillar a cross-ERN two-level-expert review board has been established with data scientists on the one hand and clinical scientists on the other hand. In this JA, we will build on and extend this structure to eventually all 24 ERN and non-genetic RD in WP7.2. The EURORDIS-led Community Engagement Task Force, which was initiated during the Solve-RD project, will continue to provide advice to ensure the inclusion of the undiagnosed patients' view and needs.
Towards the European Health Data Space (TEHDAS, 2021-2024; www.tehdas.eu) supports MS and the EC in building a European health data space by developing principles for cross-border secondary use of health data. The WPs address data quality, standards, interoperability, governance, and legal barriers. One of these examines 15 European data sharing initiatives of which the ERN is one. The results of TEHDAS will support the pan-European dialogue that follows the EHDS proposal.
X-eHealth (2020-2022; www.x-ehealth.eu) dealt with interoperable health documentation and data sharing, specifically in the patient summary and hospital discharge report. It is relevant to share information on the RD dataset (once it has been agreed by WP8 of this JA) and suggest incorporation of as many of its elements as possible into these interoperable generic documents.
In line with the continuous health policy commitment of the EU to RD, the JA will build on further past projects, such as the previous JA EUCERD and RD-Action, as well as the INNOVCare or RD-Code projects. We will align our activities with all relevant network activities in the respective field of action. In addition, we will closely follow, cooperate with, and integrate all ongoing EC efforts in the field, e.g. cross-border healthcare or the development of the new clinical patient management system (CPMS). This JA will involve all MS plus Norway and Ukraine, and it will, in a common, transparent, and integrated effort, create documents and resources concepted in such a way that each country can use them to adapt them to their own specific situation and needs. This is of particular importance as healthcare is by EU law exclusively in the autonomy of the MS. In addition, and in line with the unique initiative of the ERN, the JA will include a number of transnational/pan-european sub-projects (refer to descriptions of individual work packages for details). We propose to pilot the concepts developed in the frame of the project in the form of implementation steps on different levels in voluntary partner countries, always aiming at a coherent stratification (if many participating countries are involved) or diverse selection of participants, such as countries with centralized vs. decentralized healthcare systems, smaller vs. larger countries, etc. For some implementations, bilateral twinning will be offered as a means to facilitate the process in the less experienced country. Capacity building for the proposed actions will be supported in all participating countries and across all work packages. That way, we aim to achieve more overall equity between all European countries and patients from the different countries and regions with respect to access to healthcare of the highest quality. In addition, we will focus on the sustainability of the proposed measures in all countries, for example by encouraging and enabling the integration of JA contents into the national plans and strategies for RD.

#§COM-PLE-CP§# #§PRJ-OBJ-PO§# #§REL-EVA-RE§# #@QUA-LIT-QL@# #@CON-MET-CM@#

2. QUALITY

2.1 Concept and methodology

Concept and methodology

Outline the approach and methodology behind the project. Explain why they are the most suitable for achieving the project's objectives.

The typical overall approach within most WP will be to begin with a state-of-play analysis regarding a specific aspect of integration of ERN into national healthcare systems, as well as a collection of best practices, followed by a structured analysis of the results and the development of recommendations and tools which are universally applicable and/or targeted at the diversity of MS, with key and optional elements, taking into account the specific situation in the different participating countries. In addition to developing recommendations, guidelines, and tools, we will – especially during the third year of the JA – carry out implementation steps in the form of voluntary pilots on different levels in a set of MS, or demonstrators to test the feasibility and set the grounds for continuous improvement. In some instances, twinning/training will be offered by a more advanced country in order to facilitate the acquisition of expertise and knowledge. Other capacity building strategies to empower implementation of the proposed measures in participating countries will include the organization of specific workshops targeted at national decision makers and payers.
Technical and procedural aspects of our proposed methodology include setting up a common pool of resources (tools and solutions, internal and external expertise) available to all partners throughout the project, which will be made visible via the internal project management tool, in order to achieve maximal synergies and sharing between WP. We will hold dedicated conference calls for WP leads and task leads to avoid duplication of efforts, offer advice, and to harmonize as much as possible certain activities, such as surveys, data analysis, consensus finding, extended consultations etc. We will uniformly use the EUSurvey tool to ensure GDPR compliance. Wherever possible, surveys will be prepared and launched jointly between WP/tasks. Likewise, we will build on existing knowledge and structures and use synergies with other projects and initiatives whenever we can. Stakeholder participation will be ensured during all steps by their inclusion in working groups, as well as, for certain stakeholder groups, regular exchanges via the management structure of the project, which will be established right at the beginning (see section 2.4). Inclusion of the patients' point of view and patients' needs will be guaranteed especially by the participation of EURORDIS as a leader or contributor in a number of tasks, in addition to the involvement of patients' representatives in the different working groups. Cross-linking between the WP will be achieved by establishing regular meeting series wherever necessary. Formal methods will be employed for consensus finding (Delphi process, nominal group technique). Face-to-face meetings will be organized back-to-back with the annual consortium meetings wherever possible.
Apart from these general aspects, each of the specific WP has its own approach and methodology to achieve its specific objectives:
WP2: Dissemination
The main communication language of the JA will be English. eTranslation (online machine translation service provided by the EC) can be offered for the 24 official EU languages and Norwegian. The definition of the audience needs will be carried out by the following approach: i. segment the audience into levels of interest and influence in readiness for development of channel strategy and key messages. Stakeholders will cover the target groups, especially primary care clinicians and patients, as well as other actors which will be identified at national level. In person or online national meetings will be conducted; ii. hold communications workshops/meetings with other WP, MS, and ERNs communications leads to understand their specific needs and challenges (e.g., system maturity, technology, etc.). A pre-blueprint for dedicated national campaigns will be produced by M18, taking into consideration the two main target groups: patients and clinicians. The pre-blueprint should answer how the ERNs would be more accessible and usable at national level, encouraging the empowerment of patients. It will be tested as a pilot in three countries (IE, IT, RO) from M18 - M24 and the results evaluated between M25 - M30 in collaboration with WP3. The final campaign blueprint will be produced from M31 to M36.
WP4: Sustainability
A strategy for the sustainability of JA actions will include measures at European and national level that are continuous, i.e., both precede and will be continued after the accomplishment of JA (ERN BoMS and its Working Group on ERN integration into national systems, and ERNs) and those that will be purposefully developed for this JA (National Policy Contact Point group). A strong focus is given on the achievement of common agreement on sustainability elements that goes beyond the JA through extended consultations, and capacity building to foster better integration of sustainable elements of the JA into the national legislation, such as national RD plans and strategies. Participation of all MS and Ukraine in this JA ensures full coverage of the EU in this JA.
WP5: National governance and quality assurance models
In this WP we will explore the current situation in the different MS through survey and mapping of existing national governance models for ERN-HCPs and care pathways; proceed to the identification of existing best practices, gaps, and deficiencies; evaluate all elements and means to align national and European ERN policies; and elaborate possible strategies and models for the integration of ERNs into the different types of national healthcare systems (including the proposal of governance structures for the integration of ERNs in NHS and inclusion in the data flow to guarantee the inclusion of national/ regional governments in the transmission of information/analysis regarding the current situation on ERNs inclusion/impact).
In order to develop national indicators aimed at mapping the level of integration of the ERNs into the

national healthcare systems in Europe, a co-designed approach will be adopted. Specifically, a maximum of 3 indicators will be co-designed together with the ERN Continuous Monitoring and Quality Improvement Working Group (that involves ERN BoMS representatives, ERN Coordinators and Managers and representatives from DG SANTE) and with the involvement of patient's representatives from the EURORDIS Amequis Task Force. The co-design will take into account the previous experience gathered in the ERN Continuous Monitoring data collections, in the current ERN evaluation as well as in the results of the Amequis project. An initial draft of the putative indicators will be designed with the support of the ERN Continuous Monitoring and Quality Improvement Working Group and with the patient's representatives in a dedicated discussion workshop. Afterwards, a Delphi process will be organised among the ERN Coordinators, the ERN BoMS, with the EURORDIS Amequis Task Force, as well as with DG SANTE to agree on the indicators. Subsequently, a pilot implementation of the national indicators developed will be organised in at least 3 different MS with different healthcare systems. After the data collection, a refinement of the indicators will be performed in order to take advantage of the experience gathered in the pilot implementation and to develop the final version of the set of indicators. The final set of validated indicators will be implemented by means of a data collection in at least 8 MS and based on the final implementation, a guidance document will be developed for the future data collections and also for the eventual integration of new indicators dedicated to the Integration of the ERNs into the national healthcare systems in the future ERN evaluation.

WP6: National care pathways and ERN referral systems

This will include a complete cycle from development of a sign-posting tool and model reference care pathways for identified RD or groups of RD to the comprehensive analysis of barriers and enablers for their implementation, to real-life pilots of care pathways in national systems and to the development of recommendations for care pathways, case management / care coordination, referrals to ERNs and inclusion of CPMS advice, based on the diversity of MS and set for continuous improvement. These actions will be complemented by the best practice sharing and capacity building activities. In addition, we will develop recommendations for an early and continued emergency response to sustain healthcare support and care pathways in nationwide disaster situations with severe impairment of the regular healthcare system (like pandemic outbreaks, earthquakes or military conflicts) with a particular focus on people living with rare and complex diseases, based on the analysis of the experiences of patients, advocacy organizations and aid agencies working in Ukraine and other countries. The development of this guidance document will be accompanied by capacity building activities for the new Ukrainian RD hub including training activities, development of procedures to sustain elementary care and treatment needs and building temporary gap-bridging pathways to neighbouring countries and the ERN system to restore disrupted national care pathways. To evaluate the success of these measures, appropriate indicators will be developed to allow the comparison of well-defined parameters at the start and the end of the project.

WP7: National reference networks and undiagnosed disease programmes or equivalent strategies interlinked with ERN

To address the objective of support capacity building in MS for the development of national reference networks, or equivalent strategies, for rare and complex diseases and their integration with ERNs, three tasks will be performed. Firstly, a state-of-the-art analysis of existing structures, models and initiatives on member state level will be performed by using a combined approach of quantitative and qualitative methodologies. A comprehensive survey will collect comprehensive overview information from the member states. This will be supplemented by a series of dedicated meetings - using the focus group methodology - with multi-stakeholder representatives of five informative member states. Based on the analysis of the state of the art, models and recommendations for national reference networks, or equivalent strategies, for rare and complex diseases and their integration with ERNs will be developed by means of a multi-stakeholder consensus process. Delphi process and the nominal group technique will be applied as methodology. In task three, capacity building workshops and pilot implementation for national reference networks, or equivalent strategies, will be done. While the capacity workshops will be based on a multi-aspect toolbox approach to adapt to the different capacities and situations in member states, implementation pilots will test the models and recommendations for national reference networks, or equivalent strategies. Implementation pilots will focus on different defined aspects of the national reference networks. Based on the selected aspect, the pilot implementations will tightly collaborate with WPs 5, 6, 8 and 9.

To develop ERN-linked structures and procedures for undiagnosed patients, covering every aspect of the specific needs of this large group of patients, we will first perform a state-of-play analysis and, based on the results, develop recommendations for national UDP or equivalent strategies, including their national and international integration and national governance. To make use of the exceptional expertise of the ERN, we aim to establish an ERN-overarching expert panel for undiagnosed cases unsolvable on a national level and requiring interdisciplinary input, and for which the target-ERN is primarily unclear, building on and extending the structure implemented in the Solve-RD project, which is designed to solve cases with a suspected genetic disease. To harmonize access to this panel, which will eventually comprise all 24 ERN and include also non-genetic cases, and to lay the foundation for a homogenous European undiagnosed patient cohort, we will develop a European SOP for assigning ORPHAcode 616874 (undiagnosed RD) based on the existing German SOP. In line with this, we will develop

recommendations for registration of undiagnosed patients in Europe. To enable multidisciplinary panel discussions for undiagnosed cases, we will identify requirements for teleconsultation systems – focusing on the new CPMS – and develop recommendations for its use in national UDP. Finally, to provide a further link for undiagnosed patients with ERN and all resources interlinked with them, we aim to facilitate the formation of specific patient organizations or equivalent strategies. Sustainability of the proposed actions will be promoted by pilot implementations and capacity building workshops. Specific cross-links exist with WP 5, 6, 7.1, and 8, for which regular joint meetings will be organized.

WP8: Data management

The methodology towards achieving WP8 is to promote improving the integration of data management between national health systems and ERNs on priority topics for RD, acknowledging the diversity between member states. Current barriers for integration per member state will be analysed and existing solutions to address them inventoried; existing solutions will be tested, and where needed reprocessed and documented to become implementable solutions for member states; improvement upon the baseline will be tested and demonstrated in selected member states and HCPs in close collaboration with ERNs in an Agile way. In addition, a tool that helps patients and non-specialised medical professionals find RD experts and expert centres will be deployed in some countries to demonstrate scalability to all member states. Integration stewards will play an important role in developing the guidance for national health systems to adopt the implementable integration solutions. Surveys, workshops, bilateral exchanges, and pilot developments will be used to achieve a set of recommended implementable solutions to be extended to all participant countries.

WP9: National support options for ERN-HCP

In order to be fully operational and constantly strive for improvement of their highly specialised healthcare services ERN centres require adequate support from their HCPs, which is often insufficient or absent. Examples include lack of recognition of extra time and effort requirements for RD specialist team consultation, insufficient manpower, clinic space etc. WP9 will map the situation and define the main barriers of various types (e.g. organisation, funding) at various levels (hospital - regional - national) across ERNs and member states. Series of online surveys will be directed at selected ERN clinical centres (lead physicians and hospital managers) and defined groups of stakeholders (e.g. national authorities, payers, charities, patient advocacy groups) to address multiple potential mechanisms of cooperation and support. Surveys will be coordinated with other WPs, namely WP 5, 6 and 8. Surveys will bring an inventory ERN-related activities requiring support at national/regional/local level (administrative, organisational and/or financial) and of potential solutions. Using online Delphi methodology recommendations will be formulated with the potential to be implemented across Europe. Recommendations for which consensus will not be reached online will be discussed in the series of face-to-face consensus conferences using Nominal Group Technique. Additionally, analysis of the CPMS reimbursement proposals by ERNs and of the results of their pilot implementation as well as analysis of existing cross-border reimbursement methods applicable to the CPMS will be performed.

#§CON-MET-CM§# #@CON-SOR-CS@#

2.2 Consortium set-up

Consortium cooperation and division of roles (if applicable)

Describe the participants (Beneficiaries, Affiliated Entities and Associated Partners, if any) and explain how they will work together to implement the project. How will they bring together the necessary expertise? How will they complement each other?

In what way does each of the participants contribute to the project? Show that each has a valid role and adequate resources to fulfil that role.

***Note:** When building your consortium you should think of organisations that can help you reach objectives and solve problems.*

The consortium of this project is composed of national competent authorities (CA) in each country, which have been endorsed by the respective Ministry of Health, and affiliated entities (AE) with a legal and/or financial link to the CA. The latter are equally important players in this JA, as they provide specific, indispensable expertise and resources that the CA itself does not have, and multi-stakeholder composition is a prerequisite for the project to achieve its goals. In this context, it is crucial to include field experts in the required areas (such as, for instance, medicine or informatics), as well as personnel experienced in policy preparation and with a strong link to the authorities in their country. Additional expertise and resources not already available in the consortium will be acquired by subcontracting. The Ukrainian Ministry of Health will be in the JA as an Associated Partner (AP). The CA, AE, and AP from each country in this JA are:

1 Austria - Medical University of Vienna (MUW; CA) is Austria's largest medical university with 8000 students, 6000 employees, 30 university clinics (including currently 16 full members or associated national centers in ERN), 2 clinical institutes, and 13 theoretical/pre-clinical centers. It has hosted

numerous European and international network projects, providing all the infrastructure and all technical and administrative support necessary to enable the coordination of a large consortium. It has also been host to Orphanet Austria since 2004, a section of the National Coordination Center for RD since 2011, and the Austrian National Office for RD since 2019.
2 Belgium - Federal Public Service Health, Food Chain Safety and Environment (FPS HFCSE; CA) has various responsibilities, one of which is the organization of care in Belgium. With the ambition to protect and improve the health of citizens, the FPS intends to be the Belgian promoter of the "One World, One Health" principle, by placing health and all its components at the heart of its concerns and missions, including human health, the health of the planet, animal and plant health and food.
3 Belgium - Sciensano (AE) has more than 850 staff members committed to human and animal health. As our name suggests, science and health are central to our mission. Sciensano's strength and uniqueness lie within the holistic and multidisciplinary approach to health. More particularly we focus on the close and indissoluble interconnection between human and animal health and their environment (the "One health" concept). By combining different research perspectives within this framework, Sciensano contributes in its unique way to everybody's health.
4 Bulgaria - Medical University - Sofia (MUS; CA) has a unique place in the Bulgarian education system. The high level of teaching, training and the achievements in research, health care, and public activities are integrated. The University strives for regional leadership while increasing the quality of the education on a national and international scale. MUS is the oldest institution for higher medical education in Bulgaria. It is number 1 in each of five areas of study: Medicine, Dental Medicine, Pharmacy, Health Care and Public Health. The university has more than 10,000 national and international students (about 4500 coming from abroad), and more than 900 professors and lecturers.
5 Cyprus - Ministry of Health of Cyprus (MoH CY; CA) has the mission to assure a people-oriented health system for the country, emphasizing prevention and aiming at the strengthening of social responsibility through the continuous improvement of the provided services, based on professionalism and respect, equally to all citizens. A National Strategy for RD has been in place since 2012. The MoH CY has participated in a number of projects, including the JA Health Equity Europe, JA supporting the eHealth Network, and Innovative Partnership for Action Against Cancer.
6 Czech Republic - General University Hospital in Prague (GUH; CA) is the second largest provider of highly specialised care for RD patients in CZ (7 full ERN members), leads national projects aimed at defining the role of ERNs and improving the organisation of care in RD, and was the first institution in CZ to have implemented OrphaCodes in its electronic hospital records (EHR). It initiated development of the Czech national RD registry and runs a project on the specification and implementation of RD dataset. It has significantly contributed to the Czech MoH project proposal on defining care pathways, referral patterns, and principles of shared care for RD patients.
7 Germany - Bundesministerium für Gesundheit (BMG; CA), the German Federal Ministry of Health, will serve as competent authority for a consortium with 6 affiliated entities, all institutions/organisations with profound expertise in the field of ERNs and health care concerning rare diseases.
8 Germany - The Federal Institute for Drugs and Medical Devices (BfArM; AE), an authority within the German Federal Ministry of Health, has been legally entitled recently to play a key role in the digital strategy for eHealth in Germany. BfArM is responsible for medicinal product authorisation and medical device pharmacovigilance. The department "code systems and registries" publishes official medical classifications, maintains medical terminologies, thesauri,nomenclatures, and catalogues, is the National Release Centre for SNOMED CT, and has established a National mirror group for LOINC. Since 2021 BfArM is the host of Orphanet Germany. BfArM contributed to a number of European eHealth projects and collaborates closely with the WHO.
9 Germany - The National Action League for People with Rare Diseases (NAMSE; AE) brings together all key bodies and organisations of the German health care system to enable concerted action. Since the beginning in 2010 the work of all 28 NAMSE partners is coordinated by the NAMSE main office in Bonn. It has supported the action league in the preparation and implementation of the National Action Plan for People with RD published in 2013 and provided substantial support in developing quality criteria for national CoE and preparing a quality assurance model for CoE.
10 Germany - The University Hospital Tübingen (UKT; AE) is one of the leading care and research hospitals in Germany. It hosts the oldest German RD centre (RDC) (founded in 2010, accredited in 2022) including an undiagnosed disease program to which >200 patients are admitted every year. All >8.000 patients annually seen at the 16 RDC centres are Orpha coded. The RDC comprises 10 ERN full members. ERN-RND is coordinated at UKT. The RDC has played a key role in the formation of the German Reference Networks (GRN) and coordinates 3 GRN, as well as numerous present and past research networks and projects including EUROSCA, NeurOmics, and Solve-RD.
11 Germany - Heidelberg University Hospital (UKHD; AE) is one of the largest providers of top-level care for RD patients and ranked as the leading biomedical research institution in Germany. 15 specialized units, including 5 ERN members, collaborate in UKHD's Center for RD. ERN ERKNet and ERKReg, the European Registry for Rare Kidney Diseases, are coordinated at UKHD. UKHD also plays a leading role in ERICA and in the development of a virtual ecosytem for RD research in EJP-RD.
12 Germany - The University Hospital Würzburg (UKW; AE) is a top-level care and research institution with all diagnostic and therapeutic facilities and medical expertise necessary for the most

complex cases. It is full member in 7 ERNs, coordinates one national reference network (NRN), participates as full member in several other NRNs, and is involved in many national and international projects on RD addressing best care pathways and IT solutions. It hosts the teleconsultation system KONSIL-SE and the Bavarian platform for RD patient data exchange BASE-Netz.
13 Germany - Universitätsklinikum Frankfurt (UK FFM; AE), closely linking the medical faculty of the Johann Wolfgang Goethe-University (GUF) with the services of a large university hospital and medical school, has extensive experience with the management of EU projects, such as the coordination of ERN-LUNG. >20 medical departments are actively involved in RD care and research. The FRZSE runs one of the oldest outpatient clinics in Germany for undiagnosed patients. The Institute of Medical Informatics (IMI) links medical/clinical issues and informatics, with a special focus on projects in the field of RD, such as SE-ATLAS and OSSE. It is part of EJP-RD and technical partner of ERN-LUNG.
14 Denmark - The University Hospital Aarhus (AUH; CA) is Denmark's largest hospital. Its highly digitized and mature electronic medical record system has an efficient BI-unit integrating all regional data. External watch dog "The Daily Medicine" has nominated AUH as Denmark's number one hospital for consecutive years. AUH ranked amongst the top 3 European hospitals in the 2021 global Newsweek ranking of "The World's Best Smart Hospitals". Research activities result in >2,600 peer-reviewed publications per year. AUH is an active partner in TEHDAS and EHDS pilot project (Healthdata@EU).
15 Estonia - Tartu University Hospital (TUH; CA) is the largest HCP in Estonia, offering high-quality tertiary medical care in nearly all specialties. It is the only academic hospital in Estonia, the leading teaching hospital, and the only medical institution to provide genetic testing and genetic counselling in a wide range of clinical indications and all age groups. Education and research are undertaken in close collaboration with the Faculty of Medicine. The tight cooperation between the University and the University Hospital creates mutual synergy between preclinical and clinical research.
16 Greece - Ethnikos Organismos Dimosias Ygeias (EODY; CA) The National Public Health mission is to provide services that contribute to the protection and improvement of health and increase the life expectancy of the population by enhancing the capacity of the National Healthcare System, with particular focus on public health services, to effectively respond to threats to human health by communicable diseases through the early detection, monitoring and evaluation of risks, reporting and submission of evidence-based proposals and intervention measures.
17 Spain - Fundación Para La Investigación Biomedica Del Hospital Universitarios La Paz (FIBHULP; CA) is the administrative and management structure receiving the command from the Hospital Universitario La Paz (HULP) to manage the European projects. HULP is the referral hospital for an area with a population of 750,000. FIBHULP concentrates the clinical, translational, and experimental research of the HULP. Since the start of FIBHULP in 2003, 86 international projects have been funded under the different European programmes. The institute harbours 55 research groups and coordinates ERN-TransplantChild. HULP also participates as a full member in 9 ERN.
18 Finland - Finnish Institute for Health and Welfare (THL; CA) is a national research institute that provides information for decision-making and activities in the field of health and welfare. THL is responsible for national coordination of rare diseases in Finland. THL is the competent authority of Finland in both Nordic and European RD networks including ERN BoMS.
19 France - Ministère de la santé (DGOS; CA) The RD mission inside the MoH and DGOS (General Directorate for Care Provision) is responsible for implementing the actions for RD policy and the third national Plan for RD. They are reporting back to the Strategic Committee and the cabinet. It ensures that actions are implemented in accordance with the scheduled calendar, assesses the results of the Plan using indicators and monitors spending compared to the scheduled budget. It prepares the annual report for the RD plan.
20 France - Hôpitaux Universitaires de Strasbourg (HUS; AE) is ranked among the top French university hospitals. The collaboration, which is internationally recognized in the Shanghai academic ranking, between the University Hospital of Strasbourg and the University of Strasbourg on various projects, stands as an example within the teaching hospital national landscape. From 2017, HUS coordinated ERN-EYE, and is member of ERNs ReCONNECT, RITA, EpiCare, CRANIO, Euro-NMD.
21 France - Hospices Civils de Lyon (HCL; AE) is France's second university hospital centre. It includes a staff of 24,000 of which 5000 are physicians and is involved in training 1850 students annually. More than 60,000 inclusions of patients in research programs and more than 2,000 clinical trials are conducted each year. HCL is the beneficiary of EC funds and Coordinator of ERN EpiCARE.
22 France - Assistance Publique-Hôpitaux de Paris (APHP; AE) is the biggest teaching hospital in Europe, federating 39 hospitals and groups of hospitals (22,474 beds, 8M patients per year, >19,000 medical staff incl. 1000 professors and >6000 residents and medical students). It is involved in / coordinates >100 EU research projects. It is also the first biomedical research center in Europe with >500 clinical trials and >8,000 publications per year. The National Rare Diseases Data Bank (BNDMR) is a priority project of the National Rare Diseases Plan 2, funded by the MoH. AP-HP has been commissioned with the project management of the BNDMR, in particular the BaMaRa application.
23 France - Institut National de la Santé et de la Recherche Médicale (INSERM; AE) is a public scientific and technological institute operating under the joint authority of the French Ministry of Health and Ministry of Research. It has coordinated the Orphanet consortium since 2001, the Rare Diseases Task Force (2004-2009), and the European Union Committee of Experts on Rare Diseases (EUCERD)

(2010-2013) and led the scientific secretariat of the EUCERD JA (2012-2015), the Orphanet Europe JA (2011-2014), and the RD-ACTION JA (2015-2018). INSERM coordinated the Orphanet Network Direct Grant (2018-2021), the RD-CODE project, and the OD4RD pilot, co-leads the EJP-RD Pillar 2 on data and resources infrastructure for research, and coordinates OD4RD2.
24 Croatia - University Hospital Center Zagreb (UHCZ; CA) is the largest hospital in Zagreb, providing highly profiled procedures performed at top quality levels, comprising all medical specialties and diagnostic facilities necessary to diagnose and to treat the most difficult clinical cases. UHCZ is also the national seat for reference centers affiliated to 15 ERNs. In 5 of these the Croatian counterparts are full members. Extensive involvement in EU projects. In the H2020 project RECOVER-E, UHCZ coordinated the research activities involved in the implementation research (WP4).
25 Croatia - The Ministry of Health of the Republic of Croatia (MoH-HR; AE) is a state administration body that implements state policy related to the health care system. It prepares programs and projects and participates in the implementation of projects from the programs of the EU and international institutions. It created the National Program for RD 2015-2020 which was adopted in 2015 and included nine strategic areas of activity. As part of the National Health Development Plan for the period from 2021 to 2027, the MoH plans to improve health care models for people suffering from RD with the aim of ensuring early diagnosis and appropriate treatment.
26 Hungary - The National Center for Public Health and Pharmacy (NCPHP; CA) functions as a central budgetary authority being a central agency under the direction of the Minister responsible for public health. NCPHP has national competence within its scope of activities. In order to fulfil the public health goals set out in the legislation, NCPHP performs managing, coordinating and supervising activities related to public health (especially environmental and settlement health, food and nutritional health, child and young health, radiohygiene and chemical safety), epidemiology, health development (health protection, health education, health promotion), and public health administration, as well as supervision of healthcare provision; furthermore on the basis of delegated competence NPHC carries out tasks and duties relating to occupational health (workplace hygiene, occupational medicine), exercises and executes private law rights and duties in the field of occupational health.
27 Ireland - Health Service Executive (HSE; CA) is the national body responsible for the implementation of health and personal social services through state funded medical professionals, hospitals, and community services. It is a full member of 18 ERN led by 5 national academic hospitals. The National RD Office (NRDO) is the 'hub' for national RD activities and supports ERN integration into the Irish health system. The consortium for this application consists of NRDO core staff in a leadership and coordination role and medical senior personnel from the HSE ERN participating hospitals. The NRDO has coordinated Irish membership in RD-Code, ONW Orphanet Direct Grant, RD-Action, and EJP-RD Pillar II grants.
28 Italy - Istituto Ortopedico Rizzoli (IRCCS IOR; CA) is an international benchmark in orthopaedics development. It is clinically active throughout the entire orthopaedics and trauma field and has been recognized by the Italian MoH as a scientific research hospital since 1981. It is also a key component of the regional health care system of Emilia-Romagna and a teaching hospital for the University of Bologna. The Rizzoli Institute is ranked 11th among the World's Best Specialized Hospitals in 2021 and 1st in Italy. The Institute is the Coordination Center for ERN Bond and member of ERN EURACAN.
29 Italy - Azienda Ospedaliero Universitaria Pisana (AOUP; AE) is the coordinator of ERN ReCONNET and participates in this project through the Rheumatology Unit, which is part of the University Hospital of Pisa. Its clinical activity is focused on inflammatory rheumatic diseases, and it is the Tuscany referral centre for systemic autoimmune diseases. Its research activity is focused on epidemiological studies and prognostic factors of systemic autoimmune diseases, quality of care, imaging studies, outcomes measures. The research activity is documented in more than 500 published scientific articles over 40 years of activity, several participations to national and international congresses and a worldwide esteem of the research activity.
30 Italy - Istituto Superiore di Sanità (ISS; AE) will contribute to the project with the National Center for RD (NCRD). NCRD has a long experience of European and International projects, such as: NEPHIRD, EPIRARE (Coordinator), RD-Connect (WP-Leaders in Registries and Data Bases), Europlan (Coordinator), EJP-RD (WP Leaders), Undiagnosed RD: a joint Italy – USA project (Ministry of Foreign Affairs and International Cooperation, Coordinator). NCRD is also IRDiRC FCC Member and national Coordinator of the Italian Registry for Rare Diseases. As regards undiagnosed rare diseases, NCRD is the Italian representative of the Undiagnosed Diseases Network International.
31 Italy - IRCCS Ospedale Policlinico San Martino (HSM; AE) of Genoa pursues the aims of care, treatment, training and clinical and translational research and ensures the collaboration between the Regional Health Service and the University of Genoa. HSM is included in the "List of Accredited Centres and Reference Operating Units for the diagnosis and treatment of RD in the Liguria Region and operates as a reference centre for many RD. It participates to the Regional RD Desk for the Liguria Region. Experts of HSM are part of the Scientific Technical Committee "Biomedicine, Rare Diseases and Undiagnosed Diseases ". HSM is a full member of 5 ERN.
32 Italy - Veneto Region (AE): The Italian regions are in charge of the healthcare networks for RD. One of the common elements of these networks is the official identification of reference hospitals per

groups of RD. Some of these have undergone a pre-regional and national selection process to participate to the call for ERN full members. To harmonize health policies on RD, an interregional technical board has been set up, composed of representatives of each region, who usually are the coordinators of the RD coordinating center in charge of the whole RD healthcare network of each region or of a group of regions. Veneto Region coordinates the Italian RD Interregional Board and thus represents all the Italian regions in the JA.

33 Italy - Azienda Sanitaria Univeristaria Friuli Centrale (ASUFC; AE) of Udine is a third level health-care institution and the first hospital in Italy and one of the few hospitals in Europe to be accredited as Academic Medical Centre by the Joint Commission International. Every medical specialty is represented, equipped with the latest technological procedures. The hospital offers areas of excellence including the Regional Centre for RD and is also a higher education hospital recognized for high-level research and learning opportunities. Its Laboratory is the third laboratory in Italy for dimension of analysis and one of the best equipped in Italy. It is the Coordinator of MetabERN.

34 Italy - Fondazione Policlinico Universitario Agostino Gemelli IRCCS (FPG; AE) is the 37th best hospital in the World. Since 2021 it is also a Joint Commission International Accredited Healthcare organisation. It currently treats more than 13,000 RD patients and is member of 16 ERN. In a close collaboration between Obstetrics, Paediatrics, Adult Medicine and Geriatrics, the FPG has created a unique Transition Model, ensuring the health care for the whole life of a RD patient. Beside the clinical platform, FPG is also endowed with 21 research core facilities which are part of a Science and Technology Park (G-STeP). Access to all facilities is guaranteed through a dedicated web platform.

35 Italy - The Italian Ministry of Health (IT-MoH) is one of the main institutional partners in the project. It is already working to share its expertise and support the effort of Competent Authority to build the Joint Action. IT-MoH will make available the legal and administration's skills of the health attaché and the medical skills in RD of the doctors that work in the General Direction of health programmation. IT-MoH will support WP5 of the JA. The role of IT-MoH is also important in supporting the Competent Authority in sharing information with over 100 reference centers affiliated in ERNs.

36 Lithuania - Lithuania Vilnius University Hospital Santaros Klinikos (VULSK; CA) is funded by Vilnius University and Lithuanian MoH with three key assignments of highly specialized healthcare, research, and education/training. All rare disease activities in VUH SK are coordinated by Joint Center for Competences and Biomedical Research (JCCBR) that serves as a clinical and biomedical research hub providing IT infrastructures, managerial, administrative and coordination services for 36 Competence centres for rare and complex diseases. VULSK is designated national coordinator of Orphanet Lithuania, former partner of RD-Action, a hub of BBMRI-ERIC, full Member of 8 ERN.

37 Luxemburg - The Direction de la Santé (DISA; CA) has launched the first Luxembourg National RD Plan (PNMR) in 2018. One of the objectives of the PNMR is the development of international collaborative links in order to set up a sustainable anchor to the ERN. With the aim of integrating the strategy of the "ERN Hub" in the national health system, Luxembourg's participation in this JA is paramount to facilitate access to ERNs for patients and health professionals. DISA will coordinate the development and implementation of this JA in close collaboration with the Centre Hospitalier du Luxembourg and ALAN Maladies Rares Luxembourg association (subcontracting entities).

38 Latvia - Children`s Clinical University Hospital (CCUH; CA) is the only specialized children's multi-profile hospital in Latvia providing multidisciplinary children's care in more than 40 specialties. Specialists of CCUH provide a wide range of health care in the treatment of complicated and complex illness cases. The task of CCUH is to integrate everything created and achieved so far in the new, modern form of the hospital, using digital and new technologies and according to the requirements and needs of the public. CCUH is involved in 15 ERNs and has a specialized RD center.

39 Malta - Ministry for Health-Government of Malta (MFH; CA) is responsible for the provision of health services in Malta and for their regulation and standards. The major public health care provider in Malta is Mater Dei Hospital. The hospital is an entity that is fully under the responsibility of MFH. It is an acute general and teaching hospital and has close to 1000 beds. This hospital provides most of the secondary care and some tertiary care to the population residing in Malta. It is physically adjacent to the University of Malta and includes the national Oncology Center within its premises.

40 The Netherlands - The Ministry of Health, Welfare and Sports (VWS; CA) is responsible for governance, setting priorities, overall decision-making, connecting with other policies concerning RD.

41 The Netherlands - Zorgonderzoek Nederland (ZonMw; AE), The Netherlands Organisation for Health Research and Development, funds health research and stimulates use of the knowledge developed to improve health and health care. ZonMw is involved in several European programmes funded by the EC, amongst many others also for research on RD, like ERA-Nets E-Rare 1-3 and EJP-RD. Zonmw is also involved in several European Partnerships like ERA4Health (Fostering a European Research Area for Health Research) and THCS (Transforming health and care systems).

42 The Netherlands - The Radboud University Medical Center (RUMC; AE) in Nijmegen is a unique institution regarding the interaction of research, education, and care to establish an innovative approach to (genetic) diagnostics and therapeutic interventions in NL and abroad. It provides all medical specialties and the largest and most innovative genetics department in Europe. RUMC is affiliated to 18 ERN, coordinates 2 ERN, and has extensive involvement in EU projects, e.g. EJP-RD and Solve-RD.

43 The Netherlands - Leiden University Medical Centre (LUMC; AE) is a university medical center

for research, education and patient care with a high-quality profile and a strong scientific orientation. LUMC offers state-of-the-art research facilities to contribute to innovation of scientific research. LUMC participates in the League of European Research Universities (LERU), Eurolife, EIT Health, ELIXIR, EJP RD, and CODATA. It participates in 7 ERN and hosts two RD registries. LUMC is active in 140 Horizon2020 projects (45 as coordinator), 23 Innovative Medicines Initiative (IMI) projects, and 24 ERC grants. It actively participates in the national Health-Research Infrastructure (Health-RI) initiative of all Dutch University Medical Centres.

44 The Netherlands - Erasmus Medical Center Rotterdam (Erasmus MC; AE) provides top-clinical care for patients with complex care needs, RD, or acute needs for care and treatment. We work on distinctive, high-quality education that appeals to ambitious, inquisitive, and talented students and addresses the healthcare issues of tomorrow. We also work on cutting-edge, world class international medical research that helps to understand, predict, treat, and prevent diseases and health conditions. Erasmus MC is coordinating centre for two ERN (CRANIO, ERNICA) and participating in 23 ERN.

45 Norway - South Eastern Norway Regional Health Authority (HSO; CA) provides specialist healthcare for approximately 3 million inhabitants representing 57% of the population in Norway. Specialist healthcare within HSO is provided by eight local health hospitals (trusts), which are independent legal entities governed by independent boards with responsibility for the provided services. In addition, two private not-for-profit hospitals with defined catchment areas and a few private vendors provide healthcare services paid for by HSO. The hospital trusts accounts for 93% of the clinical activity in the region. The authority is subordinate to the Norwegian Ministry of Health and Care Services.

46 Norway - Norwegian Directorate of Health (HDIR; AE) has a mandate to improve the health of the citizens and the community through targeted activities across services, sectors, and administrative levels. The Directorate shall do so by virtue of its role as an executive agency, as a regulatory authority and as an implementing authority in areas of health policy. The directorate is an executive agency and professional authority under the Ministry of Health and Care Services.

47 Norway - Oslo University Hospital (OUS; AE) is a highly specialised hospital in charge of extensive regional and local hospital assignments and the provision of high-quality services for the citizens of Oslo. The hospital also has a nationwide responsibility for a number of national and multi-regional assignments and has several national centres of competence.

48 Poland - Ministry of Health Poland (MoH Pl; CA) is responsible for organisation and financing of health care for RD in Poland, currently focusing on developing and implementing National RD Plan, aiming at improving the coordination of health care for RDs, implementing RD expert centers, development of RD Registry. Furthermore, experts from Quality Section in the MoH can share knowledge and best practices in implementing accreditation standards, granting accreditation to medical entities. Now implementing legal framework aiming at improving quality in health care system in Poland and the coordination of oncological care, in particular implementing Breast Cancer Units model (and for other cancers) and hospital reference system.

49 Portugal - Ministério da Saúde – República Portuguesa (DGS; CA) is responsible for regulating and coordinating health promotion and disease prevention activities, defining the technical conditions for an adequate provision of health care, planning and programming national quality policies in the health system, and ensuring the elaboration and execution of the National Health Plan. The Department of Quality in Health has, among its attributions, the following co-dependent areas: National coordination of the Interministerial Strategy for RD, participation in the Orphanet consortium, management of "Person with RD Card", participation in the national mirror group of EJP-RD, and participation in the BoMS.

50 Portugal - Centro Hospitalar de Lisboa Norte (CHLN; AE) is a Central University Hospital integrated in Portuguese National Health System with all medical pediatric and adult specialties and sub-specialties recognized by the National Medical Council. It has more than 6,000 employees and is part of Lisbon Academic Medical Center.

51 Romania - Emergency Clinical County Hospital Craiova (ECCHC; CA) is the largest hospital in South-West Region of Romania. It hosts one of the six Regional Centers for Medical Genetics (RCMG) - RCMG Dolj, part of the Romanian Medical Genetics Network. RCMG Dolj is Expertise Center for RD, partner of Romanian Network of Multiple Congenital Anomalies with Intellectual Disabilities and member of ERN ITHACA. It has a strong expertise in prenatal genetic screening, diagnosis of chromosomal disorders, X-linked intellectual disabilities, microdeletion/ microduplication syndromes testing.

52 Sweden - The National Board of Health and Welfare (SoS; CA) is a government agency under the Ministry of Health and Social Affairs, with a wide range of activities and many different duties within the fields of social services, health and medical services, patient safety and epidemiology. The agency is in charge of the approval and termination of ERNs in Sweden.

53 Slovenia - University Medical Center Ljubljana (UMCL; CA) With over 8300 employees, UMCL is one of the largest institutions in Slovenia. It is the national seat for reference centres affiliated to multiple ERN, in multiple areas also as full members. Within UMCL our application is a joint application of two leading centres for RD: Clinical Institute of Genomic Medicine (CISLD) and University Children's Hospital (UCH), which are each hosting several ERN member centres and are reference centres for undiagnosed RD (CISLD) and hosting a national registry of non-malignant RD (UCH).

54 Slovakia - Ministry of Health of the Slovak Republic (MoH SR; CA) will be represented by Department of Health representative and General Secretary Office. On top of that, there is a

Multistakeholder Commission for RD established under the MoH with 16 experts including physicians, patient organizations, payers, screening centre, geneticists, and MoH experts.
55 Ukraine - Ministry of Health of Ukraine (AP) will coordinate the activities of Ukraine within the JA.

2.3 Project teams, staff and experts

Project teams and staff

Describe the project teams and how they will work together to implement the project.

List the staff included in the project budget (budget category A) by function/profile (e.g. project manager, senior expert/, junior expert, trainers/teachers, technical personnel, administrative personnel etc. — use the same profiles as in the detailed budget table, if any) and describe briefly their tasks. Provide CVs of all key actors (if required).

Name and function	Organisation	Role/tasks/professional profile and expertise
Due to the strict page limit in Form B, the following chapter was kept as short as possible. Further information on selected experts and their teams is also found in the related budget tables, as well as in annex "CVs".		
Till Voigtländer, JA coordinator	MUW	Associate professor, MD, board-certified consultant in neurobiology with >20 years of professional experience in neurobiological laboratory medicine and neuropathology. Orphanet country coordinator since 2004, head of National Coordination Center/ National Office for RD in Austria since 2011, leading the drafting of National Action Plan for RD published in 2015. Past and present member of several boards and expert groups related to RD healthcare policies (EUCERD 2010-2013, CEGRD 2014-2016, Cross-border Healthcare Directive Expert Group 2012-2013, ERN BoMS since 2015, advisory board on RD and advisory board on newborn screening to the Austrian MoH). Presently co-chair of the ERN BoMS.
Ursula Unterberger, co-lead WP7	MUW	MD, board-certified consultant in neurobiology. Member of Orphanet since 2004, National Coordination Center/ National Office for RD since 2011, RD advisory board Austrian MoH since 2016. Alternate BoMS representative for Austria.
Saskia Van den Bogaert, project leader	FPS HFCSE	MD by training. Experienced senior advisor and team leader in the Federal Public Service (FPS) Health, Food Chain Safety and Environment. Involved in the field of RD policy since 2010. Represents Belgium in the ERN BoMS.
Katrien Van Der Kelen, project leader	Sciensano	Master in Biotechnology, PhD in Plant Genetics (Ghent University). Experience in next-generation sequencing for the identification of genomic mutations. Certificate in Project Management. Joined the RD team in 2018, currently managing the Central Registry RD. Collaborates with the Orphanet team in OD4RD.
Atanaska Petrova Elenkova, project leader	MUS	Assoc. Prof., MD, PhD, Head of Clinic for Hypothalamic, Pituitary, Adrenal and Gonadal Diseases; Department of Endocrinology, MUS, Expert Center for Rare Endocrine Diseases (Full Member of ENDO-ERN).
Olga Kalakouta, project leader	MoH CY	Chief Health Officer, MoH, MD, MPH, FRIPH, Coordinator of EU Affairs with several participations in EU Projects including Health Programme, Structural Funds, Technical Support Instrument. Coordinator for issuing national ERN endorsement and for recognition of National Clinical Centres of Excellence and Expertise.
Pavla Doležalová, lead	GUH	M.D., Ph.D., Professor of Paediatrics, Director of Center for Paediatric Rheumatology and Autoinflammatory Diseases, member

WP9		ERN RITA, Chair of the IT/eHealth working group of ERN RITA, Director of National Coordinating Centre of PRINTO, Czech BoMS representative, co-author of multiple clinical recommendations for ERN RITA diseases and of the Czech national RD strategy.
Cornelia Blaschke, project leader	BMG	Coordinates as Scientific Expert and representative of the Competent Authority the technical and scientific input at the interface between the Ministry of Health and the JA.
Stefanie Weber, project leader	BfArM	Lead of department "Code systems and registers", Head of German WHO Collaboration Centre for the Family of International Class., SNOMED Int. General Assembly representative, Chair Subgroup on Semantics of eHealth Network. Oversees/ coordinates e.g. electronic death certification, semantic standardisation for electronic health record. Involved in RD-Code, X-eHealth, EHDS Pilot.
Miriam Schlangen, project leader	NAMSE	PhD Biology, Managing director German Cystic Fibrosis Institute (gGmbH). Head of NAMSE main office; expertise in developing, monitoring and implementation of national plans; registry for RD, quality assurance models, clinical guidelines; coordinating the German national action league.
Holm Graessner, co-lead WP7	UKT	Ph.D., MBA, Managing director of RD Centre, University Hospital Tübingen, coordinator of H2020 project Solve-RD and coordinator of ERN-RND.
Franz Schaefer, task co-lead T8.1, T8.3	UKHD	MD, Ph.D., Professor of Paediatrics, Head of Paediatric Nephrology Division: ERKNet and ERKReg Coordinator, ERICA WP lead, EJP-RD Pillar co-lead. Participant in Together4RD, RD Partnership task force. Coordinator of PodoNet, EURenOmics, ESCAPE, IPDN. SAB member of RD research consortia Screen4Care and ItineraRare. Current President of International Pediatric Nephrology Association.
Helge Hebestreit, task lead T7.9, lead demonstrator 8.4.2	UKW	MD, PhD, Professor of Paediatrics, Director of the Center for RD at, UKW. Speaker of the German Working Group of the Centers for RD, coordinator of several international and national research projects for RD such as ACTIVATE-CF and ZSE-DUO, consortium member in >20 national and international projects over the past 10 years.
Thomas O.F. Wagner, task lead T7.7	UK FFM	Prof. Dr. med., Head of Dpt. for RD UK FFM (Frankfurter Referenzzentrum für Seltene Erkrankungen), Coordinator ERN-LUNG; numerous RD projects (EJP-RD, ECORN-CF, ENCE-CF-LAM-LTX, ERN-LUNG RD Registry data warehouse, ERN-LUNG Helpdesk, ERN-LUNG eSupport, ERN-LUNG eIntegrate).
Holger Storf, task lead T8.5	UK FFM	Prof. Dr., Head of Institute of Medical Informatics UK FFM, many years of experience in various projects in the context of RD (EJP RD, ERN-LUNG RD registry data warehouse, OSSE, SE-ATLAS, and various national projects in research data management.
Diana Riknagel, project leader	AUH	Ph.D. Part of AUH management team leading the international affairs unit and director of BETA health unit at the hospital.
Katrin Õunap, project leader	TUH	MD, PhD, Head of RD Centre at TUH, consulting clinician (paediatrics, medical genetics). Professor of Clinical Genetics, Institute of Clinical Medicine, University of Tartu. Area of expertise is discovery of RD (multi omics approach). Tight collaboration with Broad Institute of MIT Harvard. Strong knowledge of epidemiological studies of RD.
Lamprini Pappa, project leader	EODY	RN, MSc, Head/ Department of Alert Planning and IHR, Project Manager SHARP JA (2019-2021), NFP for IHR/ EWRS/Preparedness and Response.

Javier de Castro, lead WP2	FIBHULP	MD, PhD, Specialist in Medical Oncology. Head of Section of Med. Oncology, Head of Lung Cancer Unit HULP, Assoc. Lecturer Oncology and Palliative Care Universidad Autónoma de Madrid, Director Thoracic Oncology Programme Centro Integral Oncológico Clara Campal, Coordinator IdiPAZ's Innovation Support Unit.
Satu Wedenoja, project leader	THL	MD, PhD, Specialist in obstetrics and gynecology; Senior Medical Officer at the national coordination center for RD in Finland; Member of ERN BoMS; Expertise in RD research and national strategy and planning in the field of RD.
Anne-Sophie Lapointe, co-lead WP8	DGOS	Senior expert, involved in RD field since 1998. Member of board of directors of national and European RD associations for 15 yrs. Member of INSERM ethics committee for nearly 6 years. Thesis and a master's degree in health ethics. Project manager at MoH for the RD mission, in charge of coordinating the actions resulting from PNMR3 launched in 2018. French representative ERN BoMS.
Hélène Dollfus, project leader	HUS	MD, PhD, specialized in med. genetics, ophthalmology. Head of HUS Med. Genetics Dpt., Coordinator of RD in Ophthalmic Genetics Reference Center and national RD network SENSGENE, Director of Med. Genetics Laboratory Inserm U1112, Instigator of future Inst. of Med. Genetics of Alsace, President scientific council of Retina FR, coordinator ERN-EYE, chair ERN coordinators 2021/22.
Alexis Arzimanoglou, project leader	HCL	Head of Dpt. of Paediatric Clinical Epileptology, Sleep Disorders and Functional Neurology, HCL. Director Rare Epilepsies Research Group, Barcelona, Children's Hospital San Juan de Dios. Editor-in-Chief of the peer reviewed educational journal of the ILAE. Current Chair of ERN Coordinators' group, elected member of European Council of the ILAE. Coordinator ERN EpiCARE.
Arnaud Sandrin, project leader	APHP	BNDMR operational director, will be in charge of operational project management of WP8.4
Teresinha Evangelista, project leader	APHP	ERN EURO-NMD Coordinator, will provide medical and scientific expertise on the activities of WP8.
Alain Verloes, project leader	APHP	ERN-ITHACA Coordinator, will provide medical and scientific expertise on the activities of WP8.
Ana Rath, task lead T8.2	INSERM	MD, master's degree in philosophy. Orphanet Director since 2014. Managing Editor of the WHO's ICD11 Topic Advisory Group for RD since 2009. Editor Ontologies section of the Orphanet Journal of RD. Chairs the Orphanet RD ontology (ORDO). Pillar 2 co-chair in EJP-RD, coordinated RD-ACTION JA, previous Orphanet Direct Grant, RD-CODE, OD4RD, OD4RD2.
Fran Borovečki, lead WP3	UHCZ	MD, Ph.D., Head of Division for neurodegenerative disorders at Dpt. of Neurology, UHZC, coordinator of FP7 project INTEGERS, consortium member in Horizon 2020 project EATRIS PLUS and coordinator of over 20 national and international projects.
Vesna Rems-Dobrin, project leader	MoH-HR	Master's degree in social work, senior expert advisor, Service for public health, health projects and programs in MoH, monitors implementation of projects and programs of health associations, involved in creation/monitoring of the RD National Plan 2011-2016.
Péter Csizmadia,	NNGYK	Sociologist, specialist of the Department of Public Health. As a project manager, he participated in the implementation of many

project leader		international projects, among which there were many joint actions.
Eileen Treacy, co-lead WP6	HSE	Clin. Lead National RD Office. Clinical Prof. of Medicine of RD Trinity College Dublin, Full Clinical Prof. Inborn Errors of Metabolism University College Dublin. BoMS rep. since 2015, member of MetabERN, Orphanet Nat. Coordinator; site PI RD-Code, Orphanet ONW, OD4RD2. HRB rep. to EJP-RD General Assembly, member ERICA Advisory Board, Lead Irish RD Clin. Pathway project.
Luca Sangiorgi, co-lead WP5	IOR	MD, PhD, MS, head of rare skeletal disorders department, coordinator ERN BOND.
Marta Mosca, co-lead WP5	AOUP	Full Prof. in Rheumatology University of Pisa (UNIPI); Chief of Rheumatology Unit and coordinator of RD at AOUP. Vice-president inter-department school of medicine of UNIPI. ERN ReCONNET Coordinator. Member Italian national committee of RD; coordinator connective tissue and musculoskeletal diseases of Tuscany Regional Network of RD.
Marco Salvatore, project leader	ISS	Head of Undiagnosed RD Interdepartmental Unit; UDNI member; ISS Responsible for project "Sviluppo di un modello diagnostico efficace e sostenibile per l'inquadramento di pazienti "orfani" di diagnosi"; Scientific Responsible for Italian Registries cystic fibrosis, tuberous sclerosis, Sturge Weber, IPF.
Angelo Schenone, project leader	HSM	Prof., senior researcher, EURO-NMD referent.
Paola Facchin, project leader	RDCC-Veneto Region	MD, Ph.D., Coordinator Veneto region Coordinating Centre and Registry and national interregional Technical Board on RD; participated in elaboration of National RD Plan 2013-2016; Task lead RD-action, RD-CODE; Past member RDTF, CEGRD, Member Rare 2030 Panel of Experts.
Maurizio Scarpa, project leader	ASUFC	MD, PhD, Director Regional Coordinating Center for RD Udine University Hospital. Coordinator of MetabERN, involved as PI or partner in several EU projects.
Giuseppe Zampino, project leader	FPG	MD, Assoc. Prof. of Medicine and Surgery, Coordinator of RD Units at FPG IRCSS, Director Dpt. of Paediatrics. Created a network of specialists trained to care for various health problems in congenital RD. Developed management strategies and models used in regional and national plans for RD. Contributed to understanding the molecular basis of a number of genetic syndromes.
Giovanni Paolo Latella, project leader	IT-MoH	Health attaché in Italian Ministry of Health, DG Programmazione sanitaria, Italian representative in the ERN BoMS.
Birute Tumiene, lead WP4, co-lead WP6	VULSK	Clinical geneticist, Coordinator for Competence Centers at VULSK, lecturer and researcher at Vilnius University, Faculty of Medicine, with special interests in clinical genetics, RD, epilepsy genetics and inborn errors of metabolism; Orphanet Lithuania National Coordinator, ERN BoMS Lithuanian representative.
Silvana Masi, project leader	DISA	MD, Head of Division for School Medicine, representing DISA at the National Committee for RD, representing Luxembourg in the BoMS.
Madara Auzenbaha, project leader	CCUH	MD, PhD, Chief physician RD Clinic of Medical Genetics and Prenatal Diagnostics, Leading researcher Riga Stradiņš university, expert by the Latvian Council of Science. Project leader/researcher in projects financed by Latvian Science Council, ESF and ERDF.

Miriam Dalmas, project leader	MFH	Ph.D., MBA, MSc (Public Health), MD, Consultant Public Health Medicine within the Policy in Health Department/Office of the Chief Medical Officer of the MFH. Representing Malta in ERN BoMS.
Onno Spruijt	VWS	Dr., policy advisor of the Minister of Health on the subject of Curative Care, Dutch BoMS representative.
Sonja van Weely, project leader	ZonMw	PhD, scientific officer multidisciplinary Dutch Steering Committee on Orphan Drugs. Co-chair Network Steering Committee, leader WP Strategy European programmes on RD (ERA-Nets ERare 1-3). Co-Pillar leader, WP leader EJP-RD. Participating in writing of Strategic Research and Innovation Agenda and application of the European Partnership on RD. ZonMw rep. IRDiRC Consortium Assembly.
Wendy van Zelst-Stams, project leader	RUMC	MD, Ph.D., Professor Care for Rare, Head of Section Clinical Genetics, Dpt. of Human Genetics, RUMC, Dutch coordinator of Orphanet, consortium member of OD4RD (EU4H) and EJP-RD.
Marco Roos, co-lead WP8	LUMC	Leader Biosemantics group LUMC. Expertise on models for bridging from computer science to health care/life science, particularly in RD, implementation of FAIR principles, organising FAIR stewardship for RD registries, data integration. Co-leading ELIXIR RD community, enabling sustainable FAIRness and Federation at the record level for RD data, patients, and samples in the EJP-RD.
Irene Mathijssen, project leader	Erasmus MC	Prof. Dr., plastic surgeon, head of Dpt. Plastic and Reconstructive Surgery and Hand Surgery at Erasmus MC. Research focus on congenital craniofacial malformations. Coordinator ERN CRANIO.
Toril Orrestad, project leader	HSO	Chief Adviser Dpt. of Medical and Health Services HSO. Project manager for restructuring the national RD system as part of the Norwegian National strategy. Cand. Polit. in Comparative Politics.
Siren Sletten Borge, project leader	HDIR	Advisor in the global health department at the Norwegian Directorate of Health. Bachelor in political science and anthropology SOAS, London University. Master in global health policy, London school of economics.
Stein Are Aksnes, project leader	OUS	MHA, MSc, Genetic Counsellor (GC). Head of Norwegian National Advisory Unit on RD. National coordinator for Orphanet.
Anna Leśniewska, project leader	MoH Pl	Head of Quality Section MoH, experience in developing accreditation system for hospitals and other medical entities, medical registry, quality indicators for public purposes. Responsible for implementation of National Cancer Control Strategy's tasks. Involved in EC WG on patient safety and quality of care.
Carla Pereira, project leader	DGS	Head of Division of Planning and Quality Improvement Dpt. of Quality in Health. PhD in Public Health. Master in Health Services Management (University Institute of Lisbon). Post-graduation in neurodevelopment. Degree in Physiotherapy. Development national table of functionalities. Coordinator Interministerial strategy for RD. National Orphanet coordinator, BoMS rep. for Portugal.
Rui Tato Marinho, project leader	CHLN	Head Dpt. of Gastroenterology and Hepatology, University Centre of Northern Lisbon; Full Professor and Director of Centre of Palliative Medicine of Medical School of Lisbon. Director of National Program for Viral Hepatitis, MoH. President of National Commission of Reference Centres.

Ioana Streață, project leader	ECCHC	MD, PhD, Assoc. Prof. Dpt. of Cell and Molecular Biology, University of Medicine and Pharmacy of Craiova. Medical Geneticist at Regional Centre for Medical Genetics Dolj. Coordinator or team member in more than 15 national and international projects.
Kristina Wikner, project leader	SoS	Head of Unit for Highly Specialised Care, Dpt for Knowledge-based Policy of Health Care, The National Board of Health and Welfare.
Jadranka Buturović Ponikvar, project leader	UMCL	MD, specialist in internal medicine and nephrology, Prof. of Medicine. Medical Director of UMC. Member of the Council of Medicine Slovenian Research Agency. Chair of Eurotransplant Ethical Committee, member of Eurotransplant Board.
Borut Peterlin, project leader	UMCL	MD, specialist in neurology and clinical genetics. Head of Clinical institute of genomic medicine, UMC Ljubljana. Professor of Human Genetics Faculty of Medicine, University of Ljubljana. President of the Board of the European Society of Human Genetics.
Tatiana Foltanova, project leader	MoH SR	PharmDr, PhD., professor assistant at Comenius University seated in Bratislava, Head of the Commission for RD at the MoH. Has been involved in ERN call and national and international projects in RD.

Outside resources (subcontracting, seconded staff, etc)

If you do not have all skills/resources in-house, describe how you intend to get them (contributions of members, partner organisations, subcontracting, etc.).

If there is subcontracting, please also complete the table in section 4.

This JA comprises a broad spectrum of topics which cannot be fully covered by the partners (BEN/AE); moreover, the political aim is to involve further stakeholders outside the consortium according to their expertise. Therefore it will be necessary to include a number of institutions or companies in the project via subcontracting. These are listed in the table in section 4, as well as in the budget tables.

Experts (if applicable)

*Explain if **national** and/or **international experts** will be nominated by national authorities to support the project implementation. Describe the specific professional and technical expertise and experience of each proposed expert and their contribution to the project implementation. Provide CVs (if required).*

Minimum requirements:

- *Qualification: A level of education which corresponds to a Bachelor's degree.*
- *Professional experience: At least 4 years of proven experience in XXX*
- *Other skills: ability to work in English (minimum B2 level)*

N.A.

2.4 Consortium management and decision-making

Consortium management and decision-making (if applicable)

Explain the management structures and decision-making mechanisms within the consortium. Describe how decisions will be taken and how regular and effective communication will be ensured. Describe methods to ensure planning and control.

***Note:** The concept (including organisational structure and decision-making mechanisms) must be adapted to the complexity and scale of the project.*

The consortium and the integration of all JA structures will be managed by the Coordination. The respective key management structures include (a) the Coordination (WP1) with a dedicated coordination team consisting of project coordinator, a full time project manager, an additional full time junior scientist, and a full time financial manager, (b) a Steering Committee, composed of all work package (co-)leads, (c) a Management Board, comprising all 60 partners of the consortium, and (d) a Multi-stakeholder Advisory Group with four dedicated subgroups (the National Policy Contact Point Group, the Hospital Managers Advisory Group, the Data Management Advisory Group and the Patient Advisory Group) and further stakeholders like ERN coordinators and other external experts that are not part of the JARDIN consortium. Within each work package, WP (co-)leads will establish further management structures and procedures for their internal WP management. For a graphic representation, see figure 1 below.

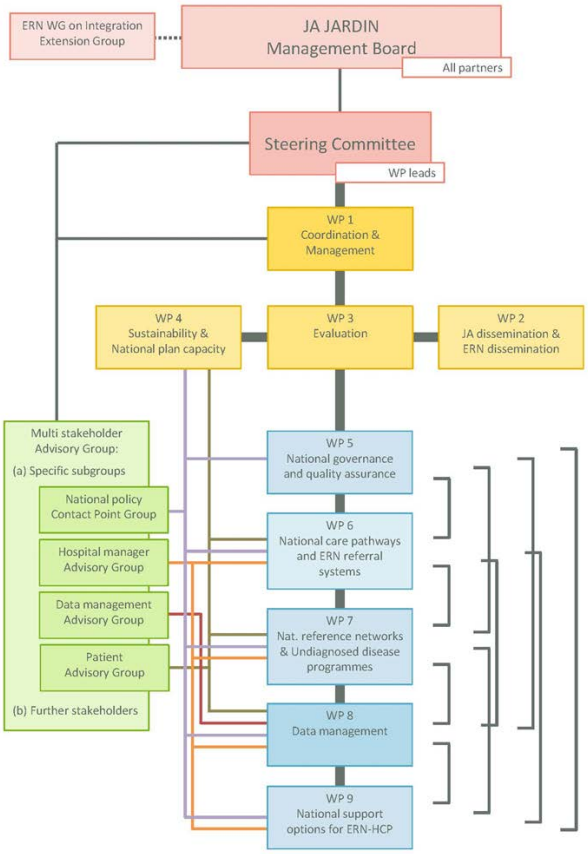


Figure 1: Consortium structure. Core of the Consortium are the four horizontal work packages (WP) (orange group) and the five vertical WPs (blue group) which are tightly connected by regular, as well as ad hoc virtual meetings on the lead level (thick grey cross), configurational represented in the Steering Committee (SC) (red group). While the SC serves the "day-to-day" strategic requirements, including regular monitoring of the progress of all activities according to schedule, it is the Management Board (MB) (red group), consisting of all consortium members, where the more long-term and important strategic decisions are taken. Additionally, the MB also serves to disseminate all relevant information on a regular basis to all consortium members, as well as those Board of Member States (BoMS) representatives that are not part of the consortium, but participated in the BoMS Working Group on Integration, a predecessor to this JA, which is now paused for the duration of the JA. These representatives are combined in the "ERN working group on integration Extension Group" (red group) and linked to the MB, receiving all information and participating in all discussions, albeit without voting rights. Advice is provided by the Advisory Group (green group) with dedicated subgroups for well-defined stakeholders which need to be in close contact with selected

WPs that are of special interest and/or importance for the respective subgroup. Four levels of short to long brackets on the right side visualize the close linkage and interaction between the five vertical WPs.

The Coordination will be in charge of the "day-to-day management" of the consortium, combining management, information, and support functions for all partners and activities. In addition, the Coordination will keep close connections to DG SANTE and HaDEA. For the "day-to-day strategic requirements", including discussions concerning all WP and the proceedings and planning of the JA in general, the discussion of risks and risk mitigation, any minor strategic decisions, and the regular monitoring of the progress of all activities of the project according to schedule, the Coordination will be supported by the Steering Committee that is situated at the core of this first higher-order management level. The Steering Committee will also be involved in the preparation of major strategic decisions for the Management Board. The Management Board constitutes the second higher-order management level and will be responsible for the more long-term and important strategic decisions, as well as potential amendments of the project content, budgets and timelines. Management Board meetings will also serve as tool to disseminate all relevant information regarding JARDIN on a regular basis to all consortium partners and further stakeholders.

The Multi-stakeholder Advisory Group, positioned outside the internal management structures, will provide advice to the Coordination, the Steering Committee and the Management Board on a regular basis, while the afore mentioned subgroups of the multi-stakeholder advisory group will support the work of the different work packages in a targeted manner, when their specific expertise and advice is required and requested from the respective WP (co-)leads.

Key to the management of the consortium and its activities are close communication links and loops between all structures described above (see figure 1). The main, regular communication and interaction arrangements include:

(a) Coordination:
- Participation of the coordinator (supported by the project manager or the additional full time junior scientist) in all WP meetings in order to have a precise overview of the proceedings at any given time.
- Release of a regular internal newsletter (every 6 month) by the coordinating team in cooperation with work package 2 and other partners. This newsletter will contain not only information of recent activities and achievements within the project, as well as upcoming events, but also information on tasks to be performed by the partners, important deadlines, administrative announcements etc. This format has proven useful in other EU projects (we rely in particular on the experience with the Orphanet project). It will be mandatory for the partners to read this newsletter.

(b) Steering Committee:
- Bimonthly meetings with the Coordination. In these meetings, work package (co-)leads, which have (as one central obligation) the task to keep track of the activities and contributions of all partners in their work package, will report back on the progress, any challenges and proposed solutions, as well as any potentially up-coming risks within their work package to the coordination. Apart from the information, discussion and decision functions, these regular meetings of the work package (co-)leads will further support the establishment of close links between all WPs (in addition to regular bilateral meetings between certain WPs/tasks with relevant overlaps, as described in the respective WP descriptions).

(c) Management Board:
- Bimonthly meetings, alternating with the meetings of the Steering Committee, providing information of all the activities and the main decisions, which have to be taken at this management level. The meetings of the Management Board will also be open to other participants beside the actual partners, in particular the "ERN Working Group on Integration Extension Group", comprising those BoMS representatives that were part of the BoMS Working Group on Integration (which is paused for the duration of the JA), but are not partners in the JA.

(d) Multi-stakeholder Advisory Group:
- The interactions between the multi-stakeholder group and the different structures of the JARDIN consortium will take place at two different levels:

a) Basic regular interaction level:
The coordination team will contact the whole multi-stakeholder advisory group (the members of the four subgroups detailed above and the other involved stakeholders approximately every 6 month starting from month 3, when the consortium and the project is fully operational (i.e. around M3, 9, 15, 20, 27 and 34) in order to provide regular information on the progress of the project and its activities and to ask for feedback on any open issue. A more comprehensive feedback will be requested in M 3, M 21 and M 34, each time after the main consortium meeting, to check the operational setup of the consortium and its functionality (M3) and to seek guidance regarding the mid-term results, as well as challenges and strategies ahead (in M20) and regarding the almost final results presented at the final meeting (in M34). All feedback from the advisory group will be reported to the steering committee and the management board. If useful and possible, members of the multi-stakeholder advisory group will also be invited after

consultation to participate in subsequent steering committee and management board meetings.

In case the steering committee or the management board identifies further items to be discussed with the multi-stakeholder advisory group, the coordination team will consult the advisory group accordingly in addition to the regular consultation intervals.

b) Targeted specific interaction level:
To support the work on well-defined elements in the different work packages, work package (co-)leads will consult the respective subgroup within the multi-stakeholder advisory group in a targeted fashion seeking advice regarding a particular aspect and/or inviting them to participate in specific meetings for this topic. This includes, for instance, targeted interactions between WP8 (Data management) and the Data management advisory group (red connection line in figure 1) or between the Hospital manager advisory group and WPs 6,7,8 and 9 (orange connection lines in figure 1).

This set of regular structured interactions will be supplemented by further interaction and communication activities whenever this deems necessary (for instance setting-up targeted teleconferences with single participants to solve any urgent issue or delivery of ad hoc notifications and other urgent information to all partners).

Finally, there will be regular updates on the JA in BoMS meetings, as well as in the ERN coordinators group meetings, although they will not be directly linked to the JA. In analogy to that, the JA will continue to establish links to other groups, official bodies etc., including ones that might be identified only during the project and in individual WP.

Regarding the voting system, the following rules will apply for the steering committee, as well as for the management board:

a) Voting rights:
In the steering committee, the coordinator and each work package (co-)lead have one vote. Based on 12 members of the steering committee, the maximum total number of votes will be 12. In the management board, each member state with a competent authority will have one vote. This voting right will primarily be exercised by the competent authority, but any country with additional affiliated entities in the consortium can delegate the voting power to one of these other national representatives in the board. Based on 27 European Member States and Norway, the maximum total number of votes will be 28.

b) Minimum number of participants and minimum number of votes for one voting option for successful voting:
Generally, we strive to reach unanimous agreements for every topic a voting on the level of the steering committee or the management board will be necessary. If this is not possible for a certain topic, the following rules will apply:
- For any voting decision, the minimum number of participants with voting power present in the respective meeting should be ≥ 70% (i.e. steering committee ≥ 9 members and management board ≥ 20 participants). If this quorum cannot be reached in a given meeting, the voting has either to be postponed to the next meeting or it has to be executed in written format;
- For any voting to be valid on the level of the steering committee or the management board, the minimum number of votes for one specific voting option should be ≥ 75 %. If this quorum cannot be reached, the respective voting option is rejected and other solutions have to be developed.

#§CON-SOR-CS§# #@PRJ-MGT-PM@#

2.5 Project management, quality assurance and monitoring and evaluation strategy

Project management, quality assurance and monitoring and evaluation strategy

Describe the measures planned to ensure that the project implementation is of high quality and completed in time.

Describe the methods to ensure good quality, monitoring, planning and control.

Describe the evaluation methods and indicators (quantitative and qualitative) to monitor and verify the outreach and coverage of the activities and results (including unit of measurement, baseline and target values). The indicators proposed to measure progress should be relevant, realistic and measurable.

All activities within this JA will be overseen by the coordination in WP1 in cooperation with the Steering Committee (consisting of all WP (co-)leads) and WP3 to ensure the quality and timeliness of project implementation, which will be continuously monitored by evaluation activities in WP3 (including tracking of and evaluating whether all process, output, and outcome indicators are fulfilled). Evaluation tools planned for use in internal evaluation are: (a) check-lists for activities, tasks and documents, (b) surveys and (c) a work package (co)-leads workshop in the design of a focus group. In detail:

(a) Check-lists:
- Check-lists will allow a continuous monitoring of the quality and timeliness of the activities and the project implementation in general by providing input into whether each indicator is reached, and, in case this is not the case, why some deadline was exceeded and/or why an output may differ from that originally planned.
- Regarding outputs and outcomes, we will update the checklist of indicators regularly so that any future inconsistencies can be prevented and deliverables will be timely submitted. We will develop a follow-up mechanism through constant reminders so that we can be notified in advance when changes in the content delivery of work packages do occur.

(b) Surveys:
- Surveys will be used after each important meeting including the three plenary meetings around M3, M18 and M34, the Steering Committee meetings, and during the evaluation workshop by focus group at M17. They will be sent out as soon as possible after the respective meetings, and reminders will be sent to those who have not answered in time. In addition, annual surveys are foreseen for the whole consortium at M12, M24 and M34.
- Surveys will assess participants' satisfaction with project processes and their quality. They will be short and simple in order to improve response rates and to reach satisfactory response level. In this regard, special attention will be given to the participants' motivation emphasizing the importance of their input. Furthermore, whenever possible, questions with multiple choice answers will be used, since this type of questions is more practical and more specific, allowing a standardized retrieval and analysis of participants' opinions.
- Technically, survey data will be collected online using EuSurvey (https://ec.europa.eu/eusurvey/) as a tool for construction of surveys.
- Results of surveys will be presented on the following Steering Committee with focus on interesting results in order to improve further progress of project

(c) Workshop with work package (co-)leads applying a focus group design:
- To gain a deeper insight into work-package (co-)leads' satisfaction with the project processes and the direction of development of the JA itself, a workshop with all leads is foreseen in M17.
- To ensure valuable and high-quality information, the workshop will have a participant-oriented design, containing engagement (to guide participants towards our topic), exploration (to generate ideas from participants), and exit (to wrap up a discussion on a particular topic and to give all participants a chance to be heard) questions. Participants will be invited in time and will receive all information they need to prepare for the discussion topics.

The final evaluation report will be based on the data collected from surveys, the workshop, and all documents developed within the work of the other work packages. It will include quantitative and qualitative analysis of surveys, insights into the satisfaction of work package (co-)leads from the workshop, and whether all tasks and activities defined in the Grant Agreement were completed. The main focus of the final evaluation report will be on the possibility of implementation of deliverables in participating countries, satisfaction with the deliverables, and the evaluation of the entire project outputs and outcomes. This includes:

a) Process evaluation:
- Process evaluation will measure the day-to-day strengths and weaknesses of the operational part of the project, like satisfaction with the organization of the meetings, and reaching the satisfactory level of cooperation of all of the partners in the project will be a part of process evaluation;

b) Output evaluation:

- Output evaluation will be focused on (1) determining whether the deliverables are produced on time and in agreement with what was discussed and arranged and (2) assessing the quality of all deliverables. If necessary, the quality control of deliverables might include requests for improvement and a subsequent reassessment of the output;

c) Outcomes evaluation:
- Outcomes evaluation will have the focus on the possibility of the project results, and its main deliverables to produce desired changes what needs to be evaluated at the end of the project. Besides this, the Outcomes evaluation will also include an evaluation of all the dissemination activities of the Joint A (like statistics of the website, social media statistics, etc.).

The evaluation team will collaborate closely with WP1, WP2, and WP4 to collect all the information on deliverables and milestones regarding the other WPs. Summary of the results will be written in a form of a SWOT analysis, i.e. the evaluation team will make a list of observed and expected strengths, weaknesses, opportunities and threats for the project and find out whether all tasks and activities were carried out, and documents were delivered, and for those with defined time of delivery, the team will check whether they were delivered within the timeframe.

Apart from the indicators listed in the table below, defined for each specific objective of our project, we will create further indicators during the course of the project, using again the SMART framework (specific, measurable, achievable, relevant, and time-bound). These indicators will also be monitored based on measurable goals: deliverables, milestones and their planned timeline, their assessment will be described in detail in the Evaluation Plan, along with all evaluation methods and activities that will be used for evaluation.

As initial set (designed at the stage of the final submission of the proposal document), the following process, output, and outcome/impact indicators for each specific objective will be monitored and their fulfilment evaluated:

Specific Objective	2.1 To achieve efficient and effective visibility, awareness, and acceptance of the JA to internal and external stakeholders
Process Indicator(s)	**Target**
1) Development of a communication and dissemination plan for JARDIN in general, identification of dissemination tools and channels (T2.5)	1) Communication and dissemination plan (M6)
Output Indicator(s)	**Target**
1) Implementation/start of the JARDIN website and of selected social media accounts (T2.4) 2) For internal stakeholders (all JA partners) publication of a biannual newsletter 6 times during the JA (T2.5; together with WP1, T1.2)	1) Website online and social media channels open by M1-9 2) Internal newsletter on M6, 12, 18, 24, 30, 36
Outcome/Impact Indicator(s)	**Target**
1) Measurement of website visits against baseline (BL = 0) (T2.6) 2) For internal stakeholders annual satisfaction surveys regarding the newsletter in M13 and M25 (T2.6)	1) Number of new visitors 100 (M18) and 200 (M36); number of returning visitors 64 (M18) and 128 (M36) 2) > 90 % (M13), > 95% M25
Specific Objective	2.2 To support national ERN-specific dissemination activities
Process Indicator(s)	**Target**

1) Identification of the internal and external stakeholders (T2.2) 2) Dissemination planning and implementation (T2.5), and communication activity, identification of dissemination tools and channels (T2.4) 3) Production of set of key messages for each stage of the program (T2.3)	1) Stakeholder identification (M12) 2) Communication plan (M6) 3) 1 set of key messages by M10
Output Indicator(s)	**Target**
1) Stakeholder analysis (MS1) 2) Interim Dissemination Report (D2.12) 3) Final Dissemination Report (D2.13)	1) Stakeholder analysis report (M12) 2) Interim Dissemination Report submitted (M18) 3) Final Dissemination Report (M36)
Outcome/Impact Indicator(s)	**Target**
1) Handover plan for decommissioning/closure, will include plans for a sustainable way forward following the end point of the JA (T2.5)	1) 1 strategic document by M32
Specific Objective	2.3 To develop a blueprint for national dissemination strategies on ERNs
Process Indicator(s)	**Target**
1) Identification of the internal and external stakeholders in pilot countries (T2.2) 2) Analysis and planning of campaign elements and strategies for extended information campaigns in 3 pilot countries (T2.7) taking into consideration the two categories of audiences to be reached namely (a) patients and (b) clinicians	1) National stakeholder identification in three countries (M12) 2) Report (M17)
Output Indicator(s)	**Target**
1) Draft blueprint as basis for the pilot implementation of extended information campaigns in 3 pilot countries (starting in M18) (T2.7) 2) Revised final blueprint integrating the results and conclusions from the pilot campaigns (T2.7) taking into consideration the two categories of audiences to be reached namely (a) patients and (b) clinicians	1) Draft blueprint M18 2) Final blueprint M35
Outcome/Impact Indicator(s)	**Target**
1) Analysis of the success and of weaknesses of the pilot campaigns with regard to better information about ERNs in the target groups (a) patients and (b) clinicians with a baseline survey (M18) and a final survey (M31)	1) Pilot evaluation report (M32) (includes numbers of audiences' members reached per types of audiences namely (a) patients and (b) clinicians)
Specific Objective	4.1 To develop the JA sustainability strategy including a) sustainability of JA actions at MS level and b) mechanism for sustainability/accountability at the EU level
Process Indicator(s)	**Target**

1) Identification of one National Policy Contact Point person per member state (T4.1.1) 2) Identification of sustainable elements and models per 6 WPs (2, 5-9) (T4.1.2)	1) Identification of the National Policy Contact Point Group participants (M3) 2) 6 interviews and workshops held in a virtual setting and 1 consensus conference held by M35
Output Indicator(s)	**Target**
1) A compendium of sustainability models and solutions (T4.1.2, D4.1) 2) Recommendations for better integration of JA actions and sustainable elements into the National RD Plans / Strategies (D4.2) 3) National Policy Contact Point Group established (T4.1.1)	1) 1 strategic concept paper by M33 2) 1 strategic concept paper by M34 3) List of national policy contact points (M4)
Outcome/Impact Indicator(s)	**Target**
1) National Policy Contact Point Group constituted (T4.1.1) 2) Set of agreed sustainable elements and identified sustainability models per 6 WPs 3) Diverse set of solutions across MS and regions that enable implementation and sustainability of JA actions	1) ≥ 2 group meetings by M34 2) ≥ 2 sustainable elements and 2 sustainability models per WP 2, 5-9 (M34) 3) 1 set of recommendations by M35
Specific Objective	4.2 To support capacity building in MS for the elaboration of new/updated National Plans / Strategies for RD (in terms of sustainability of JA actions)
Process Indicator(s)	**Target**
1) Evaluation of ERN integration aspects in the existing National RD Plans / Strategies (T4.2.1.) 2) Development of recommendations for better integration of sustainable elements into the national legislation, including National RD Plans / Strategies (T4.2.2) 3) Support for capacity building of MS in terms of ERN integration into National Plans / Strategies (T4.2.3)	1) Evaluation of ERN integration aspects in the existing National RD Plans / Strategies (M31) 2) A consensus conference held by M33 3) Support for capacity building of MS in terms of ERN integration into National Plans/ Strategies (M33)
Output Indicator(s)	**Target**
1) Recommendations for better integration of JA actions and sustainable elements into the National RD Plans / Strategies (D4.2) 2) Workshop to support capacity building in MS (M33)	1) Recommendations for better integration of JA actions and sustainable elements into the National RD Plans / Strategies (M34) 2) Workshop to support capacity building in MS (M33)
Outcome/Impact Indicator(s)	**Target**

1) Recommendations to support capacity building in MS for the elaboration of new/updated National Plans / Strategies for RD (in terms of sustainability of JA actions) are provided to MS (D4.2)	1) 1 strategic document (Recommendations for better integration of JA actions and sustainable elements into the National RD Plans / Strategies) to be supported/approved by European Commission-DG Sante (M34)
Specific Objective	5.1 To develop proposals for national governance models and practices for rare and complex disease HCPs and care pathways, fully interoperable with ERNs
Process Indicator(s)	**Target**
1) Presentation of work progress in the internal newsletter of the JA 2) Presentation of work progress in Steering Committee meetings 3) Regular work package meetings held	1) ≥ 4 contributions to internal newsletter at M36 2) Presentations in ≥ 90% of meetings (M36) 3) n ≥ 18 (M36)
Output Indicator(s)	**Target**
1) Report on results of (a) the state-of-play analysis mapping of existing national governance models (T5.1) and (b) the results of the analysis of existing best practices, gaps, and deficiencies (T5.2, MS6) 2) Report on the results of the case study analysis exploring possible systems that could be applied to different national contexts (T5.4, D5.1) 3) Report including recommendations for national governance models adapted to the different types of national health systems in Europe (T5.1 - T5.2, T5.4, D5.2) 4) Final WP implementation report including the analysis of elements and means to align national and European ERN policies (T5.1 - T5.10, D5.4)	1) n = 1 (M14) 2) n = 1 (M24) 3) n = 1 (M35) 4) n = 1 (M36)
Outcome/Impact Indicator(s)	**Target**
1) Number of MS with existing national governance models (to be assessed in a survey together with other WP) 2) Number of MS with existing best practices (to be assessed in a survey together with other WP) 3) Recommendations and implementation report (T5.3, T5.4, D5.2, D5.4) published and available as starting point for MS health care systems	1) n ≥ 1 (M35) 2) n ≥ 1 (M35) 3) All documents available at M36
Specific Objective	5.2 To develop a proposal for national quality assurance models for rare and complex diseases
Process Indicator(s)	**Target**
1) Co-design meeting organised with ERN Coordinators, BoMS and other stakeholders (T5.9)	1) Co-design meeting attended by ≥ 15 ERNs and 5 BoMS representatives
Output Indicator(s)	**Target**

1) First compendium of indicators to map the level of integration of the ERNs into the national healthcare systems in Europe (T5.9, MS7) 2) Report including recommendations for quality assurance models adapted to the different types of national health systems in Europe (T5.5-T5.8, T5.10, D5.3)	1) n = 1 (M19) 2) n = 1 (M35-36)
Outcome/Impact Indicator(s)	**Target**
1) Number of pilot implementations (T5.10) at the end of the project 2) Compendium of indicators (T5.9, MS7), report with recommendations (T5.8, T5.10, D5.3) and implementation report (D5.4) published and available as starting point for MS health care systems	1) Pilot implementation of indicators performed in ≥ 3 MS 2) All documents available at M36
Specific Objective	6.1 To develop recommendations for the organization of national care pathways for rare and complex diseases interfacing with ERN, including the recognition of and preferably full compliance with ERN-elaborated evidence-based resources (like clinical practice guidelines)
Process Indicator(s)	**Target**
1) Identification of RD or groups of RD for the development of model - a survey of patient representatives and ERNs (T6.2.1) 2) Mapping of patients' care trajectories, design, and consensus on optimised care pathways (T6.2.2.1)	1) 1 survey conducted (M18) 2) 1 consensus conference held by M18
Output Indicator(s)	**Target**
1) Sign-posting tool for national expertise and multidisciplinary team access, and linkages to ERNs including a tool to identify pathways for conditions not covered by the country's expertise and/or not included in ERNs (T6.1, D6.1) 2) Model (reference) care pathways for identified RD or groups of RD (T6.2.2, D6.2) 3) Development of recommendations and guidelines for implementation of care pathways that take into account diversity across MS (T6.4, D6.3) 4) Identification of BEAM (barriers/ enablers/ accelerators/ motivators) for the implementation of care pathways in the national systems by using Systems Engineering Initiative for Patient Safety framework (T6.3, D6.4)	1) 1 tool developed by M25 2) 1 compendium (blueprint) of model care pathways for RD or groups of RD by M34 3) 1 set of recommendations delivered by M34 4) A toolkit of best practices in the implementation of care pathways delivered by M35
Outcome/Impact Indicator(s)	**Target**
1) Capacity building for the implementation of care pathways in national systems (T6.5) 2) Implementation of pilots for care pathways in national systems (T6.5)	1) 1 workshop (M33) 2) 4 pilot projects implemented (M36)
Specific Objective	6.2 To develop a proposal for referral systems to ERNs
Process Indicator(s)	**Target**
1) Development of recommendations for the organisation of national care pathways, referral systems to ERNs and incorporation of CPMS advice for rare and complex diseases (T6.4, D6.3)	1) 1 set of recommendations (M34)
Output Indicator(s)	**Target**

1) Toolkit of best practices in the implementation of care pathways, including referral systems to ERNs (D6.4.)	1) 1 Toolkit (M35)
Outcome/Impact Indicator(s)	**Target**
1) Capacity building for the implementation of care pathways in national systems including referral systems to ERNs (T6.5) 2) Implementation of pilots for care pathways in national systems including referral systems to ERNs (T6.5)	1) 1 workshop (M33) 2) 4 pilot projects implemented (M36)
Specific Objective	6.3 To develop guidelines for the incorporation of CPMS advice into patients' care
Process Indicator(s)	**Target**
1) Development of recommendations for the organisation of national care pathways, referral systems to ERNs and incorporation of CPMS advice for rare and complex diseases (T6.4, D6.3)	1) 1 set of recommendations (M34)
Output Indicator(s)	**Target**
1) Toolkit of best practices in the implementation of care pathways, including the incorporation of CPMS advice for rare and complex diseases (D6.4.)	1) 1 Toolkit (M35)
Outcome/Impact Indicator(s)	**Target**
1) Capacity building for the implementation of care pathways in national systems including the incorporation of CPMS advice for rare and complex diseases (T6.5) 2) Implementation of pilots for care pathways in national systems including the incorporation of CPMS advice for rare and complex diseases (T6.5)	1) 1 workshop (M33) 2) 4 pilot projects implemented (M36)
Specific Objective	7.1 To support capacity building in MS for the development of national reference networks (NRN), or equivalent strategies, for rare and complex diseases and their integration with ERN
Process Indicator(s)	**Target**
1) Presentation of work progress in the internal newsletter of the JA 2) Presentation of work progress in Steering Committee meetings 3) Regular work package meetings held	1) ≥ 4 contributions to internal newsletter at M36 2) Presentations in ≥90% of meetings (M36) 3) n ≥ 18 (M36)
Output Indicator(s)	**Target**
1) Report on results of state-of-the-art analysis of existing structures, models and initiatives for national reference networks, or equivalent strategies on member state level (T7.1, D7.1) 2) Document on recommendations for models and recommendations for national reference networks, or equivalent strategies in Europe (T7.2, D7.2) 3) Report on online capacity building workshops for member states (T7.3)	1) n = 1 (M10) 2) n = 1 (M21) 3) n = 5 (M35)
Outcome/Impact Indicator(s)	**Target**

1) Number of pilot implementations (T7.3, all categories) at the end of the project 2) Number of MS intending to implement or maintain national reference networks (to be assessed in a survey together with other WP) 3) Number of additional MS having started to establish national reference networks at the end of the project (to be assessed in a survey together with other WP) 4) Recommendations (T7.2, D7.2) and implementation report (T7.3, D7.7) published and available as starting point for MS health care systems	1) n = 7 (M36) 2) n ≥ 5 (M35) 3) n ≥ 2 (M35) 4) All documents available at M36
Specific Objective	7.2 To develop structures and procedures for undiagnosed patients closely linked to ERN on a national and European level
Process Indicator(s)	**Target**
1) Presentation of work progress in the internal newsletter of the JA 2) Presentation of work progress in Steering Committee meetings 3) Regular work package meetings held	1) ≥4 contributions to internal newsletter at M36 2) Presentations in ≥90% of meetings (M36) 3) n ≥ 18 (M36)
Output Indicator(s)	**Target**
1) Report on results of state-of-play analysis UDP (T7.4, D7.1) 2) Document on recommendations for UDP in Europe (T7.5, D7.3) 3) Document on recommendations for a registry of undiagnosed patients in Europe (T7.7, D7.4) 4) European SOP for assigning ORPHAcode 616874 (undiagnosed RD) in CoE and ERN-HCP (T7.8, D7.5) 5) Document on recommendations for national patient organizations (PO) for undiagnosed patients (T7.10, D7.6)	1) n = 1 (M10) 2) n = 1 (M21) 3) n = 1 (M21) 4) n = 1 (M21) 5) n = 1 (M21)
Outcome/Impact Indicator(s)	**Target**
1) Number of pilot implementations (T7.11, all categories) at the end of the project 2) Number of MS intending to implement or maintain the proposed structures for UDP after the end of the project (to be assessed in a survey together with other WP) 3) Number of MS intending to implement or maintain the proposed structures for PO for undiagnosed patients after the end of the project (to be assessed in a survey together with other WP) 4) Number of additional MS having started to establish UDP at the end of the project (to be assessed in a survey together with other WP) 5) Recommendations (T7.5, T7.10, D7.3, D7.6) and implementation report (T7.11, D7.7) published and available as starting point for MS health care systems	1) n ≥ 4 (M36) 2) n ≥ 2 (M35) 3) n ≥ 2 (M35) 4) n ≥ 1 (M35) 5) All documents available at M36
Specific Objective	8.1 To develop recommendations ensuring the interoperability of data structures on MS level (local, regional, national) and ERN level
Process Indicator(s)	**Target**
1) Identification of the main organisational, technical, and legal barriers to integration of data management between National Health Systems and ERNs for the purpose of RD data sharing (T8.1.2)	1) 2 surveys conducted (M12)
Output Indicator(s)	**Target**

1) Report on the current status quo of RD data collection and sharing (T8.1.1) 2) Report on the existing solutions and identification of current barriers to RD data sharing (T8.1, D8.1) 3) Roadmap for the FAIR management of MS (national, regional, and local) and ERN data structures (T8.3) 4) "Practical solutions" for implementation in an extensive interoperability of MS data structures (local, regional, national) and ERN data structures (T8.2 – T8.4, MS11-13, D8.2)	1) n = 1 (M12) 2) n = 1 (M15) 3) 1 set of recommendations by M35 4) 1 strategic document by M35
Outcome/Impact Indicator(s)	**Target**
1) Increase in the number of countries having implemented the standardised common RD dataset (and ORPHAcodes) in health information systems compared to the baseline assessed by the survey. 2) Demonstration of successful implementation of interoperability/data sharing mechanisms between EHR and registries, EHRs and CPMS, EHRS and monitoring systems.	1) ≥ 3 countries by M36 2) 3 demonstrators by M35
Specific Objective	9.1 To collect and analyse good practices and mechanisms to provide support to ERN-hosting healthcare providers at national level as well as to individual ERN centres at the hospital (HCP) level
Process Indicator(s)	**Target**
1) Analytical reports corresponding to tasks T9.1.1 – T9.1.5 (MS13)	1) n = 5 (M24)
Output Indicator(s)	**Target**
1) Report on mechanisms (existing, partly existing, missing) to support ERN-related activities of ERN-HCPs (D9.1)	1) n = 1 (M18)
Outcome/Impact Indicator(s)	**Target**
1) Analysis completed and reports available as a basis to elaborate recommendations (T9.1, MS13, D9.1)	1) All documents available at M24
Specific Objective	9.2 To develop specific recommendations for: 1. national support to healthcare providers participating in ERNs, 2. the hospital support to individual ERN centres, 3. the CPMS service and reimbursement models
Process Indicator(s)	**Target**
1) Online meetings held to create recommendations (T9.2) 2) Final consensus conference held (T9.2)	1) n = 5 (M36) 2) n = 1 (M36)
Output Indicator(s)	**Target**
1) Guidance document on the requirements for the national support of ERN-HCPs (T9.2, D9.2)	1) n = 1 (M36)
Outcome/Impact Indicator(s)	**Target**
1) Recommendations published and available as starting point for relevant stakeholders (T9.2, D9.2, D9.3)	1) All documents available at M36

2.6 Cost effectiveness and financial management

Cost effectiveness and financial management

Describe the measures adopted to ensure that the proposed results and objectives will be achieved in the most cost-effective way.

Indicate the arrangements adopted for the financial management of the project and, in particular, how the financial resources will be allocated and managed within the consortium.

⚠ *Do NOT compare and justify the costs of each work package, but summarize briefly why your budget is cost effective.*

The cost effectiveness of the project will be ensured by specific transversal measures, as described in section 2.1, such as the structured sharing of software, expertise etc. in a common pool of resources open to the whole consortium and organizing face-to-face meetings as satellite meetings back-to-back with the annual consortium meetings. In most instances, we have replaced face-to-face meetings by virtual meetings to save travel costs. We will arrange inter-WP conference calls from the very beginning in order to allow for seamless updates and exchange of information, using synergies between WP and tasks (which we already systematically identified during the preparation phase of the proposal but will continuously monitor and revise if necessary), harmonizing our activities to avoid duplication of efforts, and to provide advice and share expertise to support everyone in working as efficiently as possible. Accordingly, we will synergize report writing by including several tasks in one larger report where applicable, and by reducing deliverables to a minimum as advised in the instructions in this template. Wherever possible, we will build on resources (data, structures etc.) that have already been established by other projects and actions; to do this most efficiently, we have identified these during the writing process, and our large consortium ensures direct links to many of these activities.
A dedicated, experienced financial officer will be hired by the coordination of the JA to provide day-to-day support to the participants on budget issues and to keep up a close link with the financial administration of the lead institution (MUW), as well as HaDEA. Expenditures and possible difficulties of the partners will be assessed at least once half-way through the project in order to be able to re-allocate funds and guarantee fulfilment of duties if necessary. One month prior to the compilation of the financial and technical reports, all partners will be asked to send in their financial documents and the budget template filled in for their institution with clear instructions (first quality control level). Upon reception, the financial officer will perform the second step of quality control to ensure compliance with the grant regulations and the original budget agreement, revise the documents in agreement with the partners if necessary, and submit them to the financial department of the MUW, which will compile them in order to provide HaDEA with a consolidated budget (third round of quality control). The MUW financial department will also manage the payments to the partners. The financial officer will inform the partners upon each payment to ensure the reception of such payment. He/she will work in close collaboration with the project manager and the project coordinator so that in case of problems, amendments to the contract etc., the steering committee will be informed in a timely manner.

The project will be receiving financial contributions from third parties amounting to 120.000 Euro as registered in the budget.

#§FIN-MGT-FM§# #@RSK-MGT-RM@#

2.7 Risk management

Critical risks and risk management strategy

Describe critical risks, uncertainties or difficulties related to the implementation of your project, and your measures/strategy for addressing them.

Indicate for each risk (in the description) the impact and the likelihood that the risk will materialise (high, medium, low), even after taking into account the mitigating measures.

***Note:** Uncertainties and unexpected events occur in all organisations, even if very well-run. The risk analysis will help you to predict issues that could delay or hinder project activities. A good risk management strategy is essential for good project management.*

Risk No	Description	Work package No	Proposed risk-mitigation measures
1	Partner withdrawal from the JA (high impact, medium likelihood)	1-9	Prevention: tight collaboration between coordination and WP leads to recognize /avoid problems; close monitoring of all progress (laid down in consortium agreement) in order to be able to move tasks to another partner Remedy: find another partner to step in in case of actual withdrawal
2	Conflicts between partners (medium impact, medium likelihood)	1-9	Prevention: open communication, clarifying tasks and objectives and the role of each partner Remedy: mediation by coordinator
3	Conflict of interest of participants serving potentially opposing institutions (medium impact, high likelihood)	1-9	Prevention: clear role definition for survey completion, recommendation formulation Remedy: consultation with responsible authority, abstain from voting
4	Low quality of written deliverables (medium impact, low likelihood)	1-9	Prevention: continuous follow-up and feed-back by responsible WP leads Remedy: revision of documents by the steering committee
5	Delays in submitting deliverables (medium impact, medium likelihood)	1-9	Prevention: continuous follow-up by WP leads, coordinator, and WP3 on the progress of WPs Remedy: support by the steering committee, withdraw funding from partner in case of failure to deliver
6	A meeting cannot be delivered face-to-face due to unforeseen external factors (low impact, medium likelihood)	1-9	Remedy: workshop/ meeting will be converted to on-line format
7	Insufficient information, interest, participation, support from MS (medium impact, high likelihood)	1-9	Prevention: close monitoring of partners by WP leads and reporting to steering committee and coordination Remedy: consultation with

			National Policy Contact Points; continuous discussions with policy makers, ministries, other stakeholders on JA objectives
8	Low participation in planned surveys (high impact, medium likelihood)	4-9	Prevention: tight collaboration with coordination, competent authorities, National Policy Contact Points; careful choice of methodology (user-friendly, clearly formulated, motivatory introduction) Remedy: repeatedly approach stakeholders, contact stakeholders personally
9	Consensus among MS on recommendations and models difficult to achieve / not achieved (high impact, medium likelihood)	4-9	Prevention: keep partners informed through regular communication of progress; personal communication with partners; adaptation of recommendations considering factors stratifying MS Remedy: consensus finding repeated until common agreement is achieved
10	Implementation hindered by insufficient commitment or objection of MS (high impact, medium likelihood)	2, 4-9	Prevention: tight collaboration with coordination, competent authorities; contact the national policy contact point of the MS Remedy: selection of another MS for the pilot
11	Board of National Authorities incomplete (non-participation of some MS) (medium impact, high likelihood)	4	Prevention: encouragement for participation sent through ERN BoMS; participation of the relevant ERN BoMS member will be asked for Remedy: identification and recruitment of board member(s) repeated until a solution is found
12	Implementation steps not completed within the proposed timeframe (high impact, medium likelihood)	2, 4-9	Prevention: tight monitoring of implementation steps by WP leads, coordination, and steering committee Remedy: apply for no-cost extension of the JA

#§RSK-MGT-RM§# #§QUA-LIT-QL§# #@IMP-ACT-IA@#

3. IMPACT

3.1 Impact and ambition

Impact and ambition — Progress beyond the state-of-the-art

Define the short, medium and long-term effects of the project.

Who are the target groups? How will the target groups benefit concretely from the project and what would change for them?

Does the project aim to trigger change/innovation? If so, describe them and the degree of ambition (progress beyond the status quo/state-of-the-art).

Although principles for organization of services for RD are substantially different from non-rare diseases, there is currently no guidance for MS how to ensure high quality, safe and integrated care for persons living with a RD, comparable to, e.g., guidance developed by the relevant EU or international bodies (as the European Observatory on Health Systems and Policies, OECD or WHO) on common non-communicable diseases or acute communicable diseases. In 2017, pioneer ERNs have been established and have already demonstrated their potential. However, as described in section 1.2, inappropriate organization of services leads to inefficient use of the benefits of the ERN in national health systems. This JA will provide a strong basis for the development of guidance for MS on how to ensure provision of high quality, safe and integrated care for rare and low-prevalence complex diseases, integrated with a cross-border framework of ERNs. The main target groups of this JA are national authorities, professionals providing care to people living with rare and low-prevalence complex diseases, and the patients themselves. In the short and medium term, the JA will provide national authorities and professionals with the evidence-based resources for the organization of services for rare and low-prevalence complex diseases and sustainment of ERN-related resources at a national level, as well as evaluation and monitoring of these services for further improvement. Through the best practice sharing and capacity building activities, the JA will empower MS to apply these resources and to implement developments of this JA. This will lay the foundation for a mature, all-encompassing ERN ecosystem eventually fulfilling the overall vision of ERNs "to provide the framework for healthcare pathways […] through a high level of integrated expertise", and eventually serve all patients with rare and low-prevalence complex diseases, "including those in the process of seeking a diagnosis or in whom a final diagnosis is not yet confirmed" [1, 2]. By achieving this, in the long term, access to timely and accurate diagnosis, provision of high quality, safe and integrated care, health indicators, health-related quality of life of patients, and sustainability of health systems will be improved.

References: [1] European Union Committee of Experts on Rare Diseases, "Addendum to EUCERD Recommendations on European Reference Networks of January 2013," 2015; [2] EURORDIS - Rare Diseases Europe, "Recommendations to achieve a mature ERN system in 2030" (December 2020).

#§IMP-ACT-IA§# #@COM-DIS-VIS-CDV@#

3.2 Communication, dissemination and visibility

Communication, dissemination and visibility of funding

Describe the communication and dissemination activities which are planned in order to promote the activities/results and maximise the impact (to whom, which format, how many, etc.). Clarify how you will reach the target groups, relevant stakeholders, policymakers and the general public and explain the choice of the dissemination channels.

Describe how the visibility of EU funding will be ensured.

Communication and dissemination (C&D) activities will be at the core of the JA. The consortium will build a potent team of dedicated staff at their institutions able to liaise with either downstream to the different ERN communications teams or upstream towards the relevant stakeholders. Activities will have a double reference: 1. Coordination and mirroring of the ERNs' own C&D activities to promote synergies, expand audiences, and deepen the impact; 2. the progress of the JA itself, focusing on the WPs' activities and driving the C&D to both the ERN and the identified stakeholders. The final Blueprint document will be the key around which a particular dissemination effort will be released. Visibility of EU funding for this JA will be clearly marked in every resource developed (web page, leaflets, etc.) either separately or with the EU funding granted to the particular ERN.

C&D will be structured in WP2, composed of seven different tasks that will cover the continuous flow of: 1) communication of the JA itself (objectives, activities, participants), 2) strengthening of the ERNs' communication (mirroring, clustering, coordination), 3) dissemination of ERNs' outputs (successful use cases, achievements, highlighting of synergies between them), 4) dissemination of JA outputs (analyses, reports, blueprint). For doing so, the JA will aggregate a team of C&D professionals (specialized journalists with management of social media and corporate communication). The team will

comprise staff from every country integrated in the JA that will be coordinated by WP2 leader (FIBHULP, ES). They will meet regularly using virtual channels and build together the new materials required in all the EU-MS official languages that apply. English will be the default language for operations, with each participating country being in charge of providing valid translations of the contents generated for C&D implementation (posts, reports, summaries, etc.). The workflow will start by analyzing the current situation of the ERNs' impact in terms of knowledge and integration into the national health systems, effective international cooperation, and maximization of success. This analysis will help to identify specific C&D objectives and needs for the target stakeholders (national authorities, health policy makers, hospital managers, public and private healthcare providers) and key audiences (clinicians, patient organizations, biomedical insurance companies, etc.). For the target stakeholders a contact list will be arranged to help organize meetings and e-newsletters with non-confidential information.
The JA webpage will be a centerpiece of the C&D strategy, available in its core in all the required languages. This JA will avoid the duplication of efforts with the existing ERN and contribute to the minimization of the information noise on the internet. The webpage will mostly aggregate and boost the ERNs' C&Ds activities alongside with the JA's own performance linked to our specific progress. We will coordinate social media posting and press releases for upcoming achievements as well as re-circulate archived material that contributes to raising awareness of the work of the ERN and their potential to the improvement of the clinical attention of rare and complex clinical conditions. Use of social media will be focused to generate engagement in a broad concerned audience. Taking into account the continuously evolving social media landscape, the JA will tailor posts to those networks that ensure a higher quality of engagement for the message to be delivered (such as LinkedIn towards professionals, TikTok towards patient organizations). Besides digital informative products, the C&D team will work collaboratively to produce and refine physical elements for dissemination (brochures, leaflets etc.).
The C&D team will lead the organization of national and international meetings to communicate ERN services and solutions towards the target stakeholders. These meetings will travel a double phase: (i) gathering initial expectations and needs for ERN integration into the national health systems; (ii) presentation and dissemination of JA outputs, mostly the blueprint for ERN sustainability.
In addition to the activities in WP2, direct, systematic communication channels will be established and maintained via the structures outlined in section 2.4. Also, the JA aims to cooperate closely with the CBHC national contact points to ensure appropriate and complete display of ERN-related information on their respective websites.

#§COM-DIS-VIS-CDV§# #@SUS-CON-SC@#

3.3 Sustainability and continuation

Sustainability, long-term impact and continuation

Describe the follow-up of the project after the EU funding ends. How will the project impact be ensured and sustained?

What will need to be done? Which parts of the project should be continued or maintained? How will this be achieved? Which resources will be necessary to continue the project? How will the results be used?

Are there any possible synergies/complementarities with other (EU funded) activities that can build on the project results?

During the course of the Joint Action, we will develop a series of measures to promote the sustainability and continuation of the project activities after termination of JARDIN. These measures can be subdivided into structural elements on the European and national level and into activity/output elements from the different work packages.

The major structures to ensure sustainability at a European level are:

a) The Board of Member States for ERNs with its "Working Group on ERN Integration into National Systems" that was set-up by the Board in 2016 and later also joined by several ERN coordinators. In the past, this working group on integration prepared key documents our project will be based on. Its activities are paused during the preparation and duration of JARDIN, but it will be re-established after our project has finished and is intended to continue some of the activities of JARDIN within the frame of the BoMS including continuation and up-dating of the JARDIN website with all its elaborated documents and monitoring of the ERN integration process based on the national integration indicators developed in work package 5. Many members of the working group are direct partners in JARDIN, and those not participating as CA or AE in the project will be linked to JARDIN at the management board level via an additional advisory group, the "ERN Working Group on Integration Extension Group" (see figure 1).
Of note, the Board of Member States, being a continuous body, legally based on the Commission Implementing Decision 2014/287/EU, consisting of national representatives from all EU/EEA Member States, delegated by relevant authorities (Ministry of Health or any other relevant), and from the EC,

exerts its supportive sustainability function also on the national level (see below).

b) The ERNs which are instrumental for the continuation of certain pilots foreseen in this JA as a proof-of-concept (including the development of reference care pathways for further adoption/ adaptation in the national systems, the ERN framework for solving undiagnosed cases and measures for FAIRification and interaction of ERN datasets with national/regional datasets).

The main structures for the sustainability at the national level are:

a) The Board of Member States for ERNs with its linking function to the national health authorities (see above).

b) The "National Policy Contact Point Group", a subgroup of the multi-stakeholder advisory group (see figure 1) that will be set-up specifically for the actions of this JA. It will be composed of one higher level representative of the Health Ministry in each Member State with an overview of and close links to all the divisions within the ministries that are targeted by the different activities of our project. The profile of the members of this group ("higher level") should include a certain level of policy mandate and capacity. All sustainable elements of WPs 2 and 5-9 with relevance to the health authorities will be developed in close coordination with the National Policy Contact Point group, introducing all relevant topics early during the term of the JA into the health policy discussion level and planning in all member states, thereby ensuring as good as possible the national sustainability and further implementation of the projects' activities and results.

c) The "Hospital Manager Advisory Group", a further subgroup of the multi-stakeholder advisory group (see figure 1) that will also be set-up specifically for the actions of this JA. It comprises the hospital managers of all ERN coordination centres, as well as one hospital manager representative from every participating member state not covered by the hosting an ERN coordination centre. Similar to the previous subgroup, the Hospital Manager Advisory group will be involved in the development of all sustainable elements falling into the decision competence and implementation power of hospital managers in all partner countries, again aiming to ensure as good as possible the national sustainability and further implementation of the projects' activities and results.

On the work package level, the sustainability of the projects' activities and results will be promoted via the several channels including:

a) WP2:
- the sustainability of the blueprint for a national dissemination strategy on ERNs with the elaboration of specific materials, as leaflets, videos, etc., will be promoted through the placement of these materials onto the websites of ERNs, HCPs that belong to the ERNs or comprise national networks, national authorities/ relevant institutions and the EC and will be available beyond the project duration.

b) WP4:
- sustainable elements of the six relevant WPs (2, 5-9) will be developed together with the National Policy Contact Point group to set the grounds for further implementation beyond the duration of the project;
- two strategic concept papers (D4.1 "Compendium of sustainability models and solutions"; D4.2 "Recommendations for better integration of JA actions and sustainable elements into the National RD Plans / Strategies") will be widely disseminated through European and national conferences and meetings, including a workshop involving multinational multi-stakeholder communities under WP4, and placed onto the websites of ERNs, national authorities/ relevant institutions and EC, where relevant.

c) WP5:
- a framework for the collection of national monitoring data will be piloted in selected countries and included into the existing ERN monitoring system for the implementation across all EU and EEA Member States.

d) WP6:
-model (reference) care pathways for RD or groups of RD, developed by ERNs, will be placed onto ERN websites;
- besides, these model care pathways will serve as a proof-of-concept for the further development of model care pathways beyond the duration of the JA;
- recommendations and guidelines for the implementation of care pathways will be published in the websites of ERNs, HCPs that belong to the ERNs or comprise national networks, national authorities / relevant institutions and EC and will be available beyond the project duration (including D6.2 "Compendium (blueprint) of model care pathways for RD or groups of RD", D6.3 "Recommendations for the organisation of national care pathways, referral systems to ERNs and incorporation of CPMS advice for rare and complex diseases" and D6.4 "Toolkit of best practices in the implementation of care pathways").

e) WP7:

- an expert panel for undiagnosed cases comprising all 24 ERN will be incorporated into day-to-day activities of ERNs, including supplementation of CPMS system;
- an SOP for assigning ORPHA code 616874 (undiagnosed RD) will be implemented in ERNs, HCPs that are members of ERNs and other Centres of Excellence in the national rare disease networks and will be used beyond the duration of the JA.

f) WP8:
- a visualisation of RD Expert Centres with ERNs and NRNs will be developed based on a harmonization of the Orphanet and SE-ATLAS data sets and data models, and subsequently implemented in interested pilot countries via individual national search start pages per country. Depending on its evaluation, success and national interest, this pilot visualisation could be available beyond the duration of the JA and could also be extended to other partner countries;
- a key recommendation (D8.2 "Recommendations for integration of data management between National Health Systems and ERN") will be placed onto the websites of ERNs, HCPs that belong to the ERNs or comprise national networks, national authorities / relevant institutions and EC and will be available beyond the project duration.

g) WP9:
- best practice and guidance documents (including D9.1 "Report on existing mechanisms to support ERN-related activities of ERN-HCPs in MS", D9.2 "Guidance document on the requirements for national support of ERN-HCPs" and D9.3 "Recommendations for CPMS reimbursement models") will be placed onto the websites of ERNs, HCPs that belong to the ERNs or comprise national networks, national authorities / relevant institutions and EC and will be available beyond the project duration.

The broad dissemination of the project activities and outputs via different channels should help to generate a public interest among various stakeholders in each member state in the project achievements and their continuation, creating a supportive framework to promote the further implementation and sustainability of the project results on a national level. One element in this context is the elaboration and implementation of new or revised national action plans or strategies for rare diseases, incorporating all the relevant documents, results and pilot implementations from JARDIN, a development that the JA should help to initiate in each member state towards the end of the project, further strengthening the sustainability of all achievements.

#§SUS-CON-SC§#

#@WRK-PLA-WP@#

4. WORKPLAN, WORK PACKAGES, ACTIVITIES, RESOURCES AND TIMING

4.1 Work plan

Work plan

Provide a brief description of the overall structure of the work plan (list of work packages or graphical presentation (Pert chart or similar)).

The activities of this project are structured in 9 work packages (WP): 4 transversal ones dedicated to the coordination, dissemination, evaluation, and sustainability of the JA, and 5 technical ones aiming to cover all aspects of the integration of ERN into national health systems. Of note, WP 2 "Dissemination" will not only take care of disseminating information on the JA, but also on ERN and resources linked to them in general. The individual WP of this JA are **WP1:** Coordination, **WP2:** Dissemination, **WP3:** Evaluation, **WP4:** Sustainability, **WP5:** National governance and quality assurance models, **WP6:** National care pathways and ERN referral systems, **WP7:** National reference networks and undiagnosed disease programmes or equivalent strategies interlinked with ERN, **WP8:** Data management, **WP9:** National support options for ERN-HCP.

4.2 Work packages, activities, resources and timing

WORK PACKAGES

Work packages

This section concerns a detailed description of the project activities.

*Group your activities into work packages. **A work package means a major sub-division of the project**. For each work package, enter an objective (expected outcome) and list the activities, milestones and deliverables that belong to it. The grouping should be logical and guided by identifiable outputs.*

Projects should normally have a minimum of 2 work packages. WP1 should cover the management and coordination activities (meetings, coordination, project monitoring and evaluation, financial management, progress reports, etc) and all the activities which are cross-cutting and therefore difficult to assign to another specific work package (do not try splitting these activities across different work packages). WP2 and further WPs should be used for the other project activities. You can create as many work packages as needed by copying WP1.

For very simple projects, it is possible to use a single work package for the entire project (WP1 with the project acronym as WP name).

Work packages covering financial support to third parties (⚠ only allowed if authorised in the Call document) must describe the conditions for implementing the support (for grants: max amounts per third party; criteria for calculating the exact amounts, types of activity that qualify (closed list), persons/categories of persons to be supported and criteria and procedures for giving support; for prizes: eligibility and award criteria, amount of the prize and payment arrangements).

⚠ *Enter each activity/milestone/output/outcome/deliverable only once (under one work package).*

⚠ *Ensure consistence with the detailed budget table (if applicable).*

Objectives

List the specific objectives to which the work package is linked.

Activities and division of work (WP description)

Provide a concise overview of the work (planned tasks). Be specific and give a short name and number for each task.

*Show who is participating in each task: Coordinator (COO), Beneficiaries (BEN), Affiliated Entities (AE), Associated Partners (AP), indicating **in bold** the task leader.*

Add information on other participants' involvement in the project e.g. subcontractors, in-kind contributions.

Note:

In-kind contributions: In-kind contributions for free are cost-neutral, i.e. cannot be declared as cost. Please indicate the in-kind contributions that are provided in the context of the work package.

The Coordinator remains fully responsible for the coordination tasks, even if they are delegated to someone else. Coordinator tasks cannot be subcontracted.

If there is subcontracting, please also complete the table below.

Milestones and deliverables (outputs/outcomes)

***Milestones** are control points in the project that help to chart progress (e.g. completion of a key deliverable allowing the next phase of the work to begin). Use them only for major outputs in complex projects, otherwise leave the section empty. Please limit the number of milestones by work package.*

Means of verification are how you intend to prove that a milestone has been reached. If appropriate, you can also refer to indicators.

***Deliverables** are project outputs which are submitted to show project progress (any format). Refer only to major outputs. Do not include minor sub-items, internal working papers, meeting minutes, etc. Limit the number of deliverables to max 10-15 for the entire project. You may be asked to further reduce the number during grant preparation.*

For deliverables such as meetings, events, seminars, trainings, workshops, webinars, conferences, etc., enter each deliverable separately and provide the following in the 'Description' field: invitation, agenda, signed presence list, target group, number of estimated participants, duration of the event, report of the event, training material package, presentations, evaluation report, feedback questionnaire.

For deliverables such as manuals, toolkits, guides, reports, leaflets, brochures, training materials etc., add in the 'Description' field: format (electronic or printed), language(s), approximate number of pages and estimated number of copies of publications (if any).

For each deliverable you will have to indicate a due month by when you commit to upload it in the Portal. The due month of the deliverable cannot be outside the duration of the work package and must be in line with the timeline provided below. Month 1 marks the start of the project and all deadlines should be related to this starting date.

The labels used mean:

- *Public — fully open (⚠ automatically posted online on the Project Results platforms)*
- *Sensitive — limited under the conditions of the Grant Agreement*
- *EU classified — RESTREINT-UE/EU-RESTRICTED, CONFIDENTIEL-UE/EU-CONFIDENTIAL, SECRET-UE/EU-SECRET under Decision 2015/444. For items classified under other rules (e.g. national or international organisation), please select the equivalent EU classification level.*

Work Package 1

Work Package 1: Project management and coordination			
Duration:	M1 – M36	**Lead Beneficiary:**	MUW
Objectives			
1) Establish an effective and efficient governance, 2) Monitor and guide the activities and ensure the quality of the JA implementation, 3) Ensure effective communication and information exchange among the JA partners and governance and advisory bodies, 4) Provide day to day administrative support to the partners, 5) Ensure appropriate financial administration with regard to budgeting, planning, and accounting, 6) Ensure all communication with HaDEA and DG SANTE, including timely presentation of all deliverables and technical and financial reports, 7) Ensure appropriate risk management			
Activities and division of work (WP description)			

Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)
			Name	Role (COO, BEN, AE, AP, OTHER)	
T1.1	Organize the kick-off meeting (KOM)	Organize KOM including work plan, logistics, constitution/approval of governing/ management structure. Publish report/executive summary	MUW	COO	No
T1.2	Establish the advisory bodies of the JA	Set up hospital managers, data management, and patient organizations advisory groups	MUW	COO	No
T1.3	Monitor the progress of the JA, ensure the quality of the implementation together with the Steering Committee and WP3	Monitor the progress, ensure the quality of the JA implementation, and address potential difficulties and opportunities arising during the project	MUW all WP (co-)leads	COO, BEN, AE	No
T1.4	Ensure communication and information exchange among project participants	Issue a regular internal newsletter together with WP2, organize regular conference calls of the consortium	MUW	COO	No
T1.5	Provide day-to-day administrative support, ensure appropriate financial	Dedicated project manager will provide day-to-day administrative support to participants. Dedicated	MUW	COO	No

	management	financial manager will manage all financial aspects			
T1.6	Report to HaDEA and DG SANTE	Perform reporting according to the official deadlines. The coordinator will act as the official representative towards HaDEA and DG SANTE	MUW	COO	No
T1.7	Ensure appropriate risk management	Perform risk management in accordance with risks laid out in section 2.7 and newly identified risks	MUW all WP (co-)leads	COO, BEN, AE	No

Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description		Due Date (month number)	Means of Verification
Deliverable No (continuous numbering linked to WP)	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month number)	Description (including format and language)
D1.1	JARDIN interim report	1	MUW	R	PU	18	Project report in English
D1.2	JARDIN final report	1	MUW	R	PU	36	Project report in English
D1.3	KOM report	1	MUW	R	PU	4	Meeting report including participants list, summary of presentations, decisions etc.
D1.4	Annual meeting 1 report	1	MUW	R	PU	23	Meeting report including participants list, summary of presentations, decisions etc.
D1.5	Annual meeting 2 report	1	MUW	R	PU	33	Meeting report including participants list, summary of presentations,

							decisions etc.
D1.6 – D1.11	Internal newsletter produced and edited on M6, M12, M18, M24, M30, and M36	1	MUW	R	PU	6, 12, 18, 24, 30, 36	Internal newsletters directed at JA participants prepared

Estimated budget — Resources
See detailed budget table (annex 1 to Part B).

Work Package 2

Work Package 2: Dissemination			
Duration:	M1 – M36	**Lead Beneficiary:**	FIBHULP
Objectives			
1) To achieve efficient and effective visibility, awareness, and acceptance of the JA to internal and external stakeholders, 2) To support national ERN-specific dissemination activities, 3) To develop a blueprint for national dissemination strategies on ERNs			
Activities and division of work (WP description)			

Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)
			Name	Role (COO, BEN, AE, AP, OTHER)	
T2.1	Set communications objectives of the JA,	Agree dissemination plan with Steering committee and dissemination contacts from the participants.	FIBHULP	COO,	No

	including both internal and external communications	Develop and agree sign-off process for later stages of communications strategy development. Hold Communications Planning session with Coordination to work through objectives and key elements below to ensure these reflect and respond to the objectives set out by the other WP	all competent authorities (except for Germany: NAMSE, Norway: OUS)	BEN, AE	
T2.2	Define the audience needs, especially patients and clinicians	Identify and segment internal and external audiences for the JA, through the analysis of updated dissemination impact from the different ERNs/MS. Conduct stakeholder analysis, segmenting audience into levels of interest and influence in readiness for development of channel strategy and key messages. Stakeholders will cover the target groups, especially clinicians and patients, as well as other actors which will be identified at National level. Hold communications workshops/meetings with other WP, MS, and ERNs communications leads to understand their specific needs and challenges (e.g. system maturity, technology, etc.). Identify language needs and consider any specific cultural issues that will help or hinder effective communications.	FIBHULP all competent authorities (except for Germany: NAMSE, Norway: OUS)	COO, BEN, AE	No
T2.3	Key messages	Produce and agree a set of key messages for each stage of the JA which reflect the work of the WPs: Tailor and translate messages, where appropriate. Adapt messages to suitable channels	FIBHULP all competent authorities (except for Germany: NAMSE, Norway: OUS)	COO, BEN, AE	No
T2.4	Channels	Identify a channel mix, considering language requirements or whether a single overarching multilingual channel is more appropriate. Compile existing ERN and MS own channels. Develop a channel strategy (actions on visibility and awareness). Commission program branding to run through all channels and other JA documentation. Commission a bespoke website for the JA, aligned with the existing ERNs. Set up appropriate social media channels, tailored to the stakeholders' use	FIBHULP all competent authorities (except for Germany: NAMSE, Norway: OUS)	COO, BEN, AE	No

T2.5	Dissemination planning and implementation	Develop C&D plan incorporating the above elements, including tailored communications toolkits for use by WP leads. Work with other WP and ERN to ensure coordinated dissemination of specific tasks/activities. Implement C&D plan. Pay specific attention to the dissemination of JA final blueprint document, with the elaboration of specific materials (leaflets, videos, etc.)	FIBHULP all competent authorities (except for Germany: NAMSE, Norway: OUS)	COO, BEN, AE	No
T2.6	Measurement and evaluation of communications activity	Develop further qualitative and quantifiable communications evaluation indicators (apart from those outlined in chapter 2.5) for all WP2 activities in close cooperation with WP3 (shared task lead) from the beginning of the joint action. Handover plan for decommissioning/ closure, including plans for sustainability after the end of the JA. This will require linking in with a range of existing dissemination structures from within MS and will be done in collaboration with WP4.	FIBHULP, UHCZ all other competent authorities (except for Germany: NAMSE, Norway: OUS)	COO, BEN, AE	No
T2.7	Draft blueprint pilot	A draft blueprint will be produced by M18, taking into consideration the two main target groups: patients and clinicians. The pilot will be tested in three countries (IE, IT, RO) from M18 - M24. The evaluation phase would be on M25 - M30, in collaboration with WP3 (including surveys and a pilot evaluation report). The final blueprint will be produced from M31 to M35.	MUW, FIBHULP, HSE, IRCCS IOR, ISS, ECCHC	COO, BEN	No

Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description	Due Date (month number)	Means of Verification
MS1	Stakeholder analysis	2	FIBHULP	Report on stakeholder analysis	12	Presentation of results to Steering Committee
MS2	Dissemination progress	2	FIBHULP	Follow up implementation according to Dissemination Plan	24	Presentation of results to Steering Committee

Deliverable No (continuous numbering linked to WP)	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month number)	Description (including format and language)
D2.1	Communication and dissemination plan	2	FIBHULP	R	PU	M6	English document on JA website
D2.2	Website and social media channels	2	FIBHULP	R	PU	M9	English document on JA website
D2.3 – D2.8	Internal newsletter disseminated on M6, M12, M18, M24, M30, and M36	2	FIBHULP	R	PU	M36	English document delivered to JA partners
D2.9	National stakeholder analysis in three countries	2	FIBHULP	R	PU	M12	English document on JA website
D2.10	Final blueprint	2	FIBHULP	R	PU	M35	English document on JA website
D2.11	Pilot evaluation report	2	FIBHULP	R	PU	M32	English document on JA website
D2.12	Interim Dissemination Report	2	FIBHULP	R	PU	M18	English document on JA website
D2.13	Final dissemination report	2	FIBHULP	R	PU	M36	English document on JA website

Estimated budget — Resources
See detailed budget table (annex 1 to Part B).

Work Package 3

Work Package 3: Evaluation			
Duration:	M1 – M36	**Lead Beneficiary:**	UHCZ

Objectives

1) To verify if the project is being implemented as planned and reaches the objectives and to evaluate whether the JA as a whole has produced planned results, delivered expected benefits, and made the desired change (evaluation of process, outputs, and outcomes). Other objectives include: to evaluate if project processes are going according to plan; to evaluate whether the participants (WP leaders, stakeholders, MS representatives, etc.) are satisfied with the project processes; to assess the outcomes of the JA; to monitor whether deliverables are produced on time and following the proposed objectives; to evaluate the ability to implement the new findings of the JA in MS

Activities and division of work (WP description)

Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)
			Name	Role (COO, BEN, AE, AP, OTHER)	
T3.1	Evaluation plan	Development of a cohesive evaluation strategy including the creation of surveys that are going to be sent out to the participants after key meetings	UHCZ, all partners	COO, BEN, AE	Yes, external expert
T3.2	Workshop	Workshop with WP leaders to gain a deeper insight into satisfaction with JA processes and the direction of development of the JA	UHCZ all WP (co-)leads	COO, BEN, AE	No

T3.3	Checklist	Creation of a checklist of process, output, and outcome indicators for WPs 1-2 & 4-9	UHCZ, all partners	COO, BEN, AE	Yes, external expert

Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description	Due Date (month number)	Means of Verification
MS3	Indicators	3	UHCZ	List of indicators prepared and confirmed for WPs 1-2 & 4-9	2	SMART indicators accepted by the Steering Committee
MS4	WP Leader's Workshop	3	UHCZ	Workshop to ascertain qualitative interviews and in-depth insight into WPs and stakeholders; conclusions will be used to complete the list of indicators	17	Workshop Report

Deliverable No (continuous numbering linked to WP)	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month number)	Description (including format and language)
D3.1	Evaluation plan	3	UHCZ	R	PU	4	Key strategic document containing all basic elements of process evaluation
D3.2 – D3.4	Survey	3	UHCZ	R	PU	12, 24, 34	Key interim strategic document based on an internal survey for the evaluation of the JA, available in English on the JA website
D3.5	Evaluation final report	3	UHCZ	R	PU	36	Report on the results of process evaluation, outcomes, outputs, implementation level of the JA

Estimated budget — Resources
See detailed budget table (annex 1 to Part B).

Work Package 4

Work Package 4: Sustainability of outcomes and outputs of the Joint Action			
Duration:	M1 – M36	**Lead Beneficiary:**	VULSK

Objectives
1) To develop the JA sustainability strategy including a) sustainability of JA actions at MS level and b) mechanism for sustainability/accountability at the EU level 2) To support capacity building in MS for the elaboration of new/updated National Plans / Strategies for RD (in terms of sustainability of JA actions).

Activities and division of work (WP description)					
Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)
			Name	Role (COO, BEN, AE, AP, OTHER)	
T4.1	Sustainable elements and sustainability mechanisms	**4.1.1:** Establishment of sustainability mechanisms: a National Policy Contact Point Group will be set up as a renewable list of contact persons within national authorities with the mandates to support sharing of responsibility, implementation and sustainability of outcomes and outputs of this JA. **4.1.2:** Identification of sustainable elements per 6 WPs (2, 5-9). Two sets of sustainable elements will be identified: i) minimal requirements across all MS (methodology: interviews and workshops with WP leaders and CA of this JA; consultation	VULSK all competent authorities (except for Germany: NAMSE, Norway: OUS; Italy IOR & ISS)	COO, BEN, AE	No

		with members of the multistakeholder group and the National Policy Contact Point group; common agreement through Nominal Group Technique - consensus conference); ii) diverse set of solutions across MS and regions that enable implementation and sustainability of JA actions across 6 WPs beyond minimal requirements (methodology: interviews, workshops and case studies with the aim to reflect five groups of MS, grouped according to health system organisation, population / country size and other factors: Large/medium federal health systems: DE, FR, BE, NL. Large/medium regionalized health systems: AT, IT, ES. Small/medium EU-14 health systems: DK, FI, GR, IE, LU, NO, PT, SE. Large/medium EU-13 health systems: PL, HU, CZ, BG, RO. Small EU-13 health systems: HR, CY, EE, LV, LT, MT, SK, SI			
T4.2	Support for better integration of sustainable elements into the National legislation, including National RD Plans / Strategies	**4.2.1:** Evaluation of ERN integration aspects in the existing National RD Plans/Strategies. Methodology: surveys and interviews, supported by the evaluation of available legal acts and literature, to identify current patterns and practices for legal definition of ERN integration into national systems. **4.2.2:** Development of recommendations for better integration of sustainable elements into the national legislation, including National RD Plans/Strategies: based on the results of T4.1.2 and T4.2.1 - identified sustainable elements, best practices for legal definition of ERN integration into national systems. Consultation with members of the multistakeholder group and the National Policy Contact Point group; common agreement through Nominal Group Technique - consensus conference. **4.2.3:** Support for capacity building for the elaboration of new / updated National Plans / Strategies for RD (in terms of sustainability of JA actions): workshop involving multinational multistakeholder communities to support common agreement, responsibility sharing and capacity	VULSK all competent authorities (except for Germany: NAMSE, Norway: OUS; Italy IOR & ISS)	COO, BEN, AE	No

		building for the implementation and sustainability of JA actions at a national level.			

Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description	Due Date (month number)	Means of Verification
MS5	National Policy Contact Point Group	4	VULSK	Establishment of a renewable list of contact persons within national authorities with mandates to support implementation and sustainability of actions, outcomes and outputs of the JA	M4	(Confidential) renewable list of national policy contact points in the internal database of the JA

Deliverable No (continuous numbering linked to WP)	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month number)	Description (including format and language)
D4.1	Compendium of sustainability models and solutions	4	VULSK	R	PU	33	Strategic concept paper published as a Pdf document in English on the project website
D4.2	Recommendations for better integration of JA actions and sustainable elements into the National RD Plans / Strategies	4	VULSK	R	PU	34	Strategic concept paper published as a Pdf document in English on the project website

Estimated budget — Resources

See detailed budget table (annex 1 to Part B).

Work Package 5

Work Package 5: National governance and quality assurance models						
Duration:	M1 – M36	**Lead Beneficiary:**	IOR; co-leads MUW, AOUP			
Objectives						
1) To develop proposals for national governance models and practices for rare and complex disease HCPs and care pathways, fully interoperable with ERNs 2) To develop a proposal for national quality assurance models for rare and complex diseases						
Activities and division of work (WP description)						
Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)	
			Name	Role (COO, BEN, AE, AP, OTHER)		
T5.1	Mapping of existing national governance models for ERN-HCPs and care pathways in MS for a complete understanding of national governance systems and RD policies	Explore the current situation in the different MS through Survey and mapping of existing national governance models for ERN-HCPs. Collect information on national networks per country (link with WP7.1), the flow of information regarding monitoring activity (link with WP8), national procedures for endorsing centres of expertise, role, and status of RD expert centres on national level, ERN-HCP coverage per country as compared to national RD prevalence, ERN clinical practice guidelines (link with WP6) or ERN policy documents.	IOR, MUW, Erasmus MC, AOUP, ISS, HSM, Veneto Region, ASUFC, FPG, HSO, NAMSE, DGS, GUH, FPS HFCSE, MoH-SR	BEN, AE	Yes; AOP	
T5.2	Identification of existing best practices, gaps, and deficiencies	Based on the results of 5.1, we will proceed to the identification of existing best practices, gaps, and	IOR, MUW, Erasmus MC, AOUP, ISS, HSM, Veneto Region,	BEN, AE	No	

		unmet needs/barriers; identify weaknesses and strengths in governance models for federated and centralized MS; evaluate different perspectives including the patient perspective. For this, we will organise a dedicated meeting with all relevant stakeholders (health care representatives including professional and authorities, patient advocacy groups, ERN coordinators group representatives and members) focusing on the identification of the best governance models. These will be identified by the analysis of the best practices, gaps, and deficiencies of each of the governance models.	ASUFC, FPS HFCSE, OUS, MoH-SR		
T5.3	Identification of elements and means to align national and European ERN policies	(a) evaluate all elements and means to align national and European ERN policies; (b) compare different national regulations (including the administrative procedures) in order to identify the barriers to the alignment of the national and European ERN policies	IOR, MUW, Erasmus MC, AOUP, ISS, HSM, Veneto Region, ASUFC, FPS HFCSE, GUH	BEN, AE	No
T5.4	Definition of possible systems that could be applied to different national contexts	Perform an analysis utilising the case study approach that allows for in-depth, multi-faceted explorations of complex issues in their real-life settings and help to understand and explain causal links and pathways resulting from a new policy initiative or service development this methodology uses to capturing information on how the intervention is being implemented and received on the ground, what gaps exist in its delivery, why one implementation strategy might be chosen over another. Data collection and elaboration.	IOR, MUW, Erasmus MC, AOUP, ISS, HSM, Veneto Region, ASUFC, UKT, FIBHULP, FPS HFCSE, GUH	BEN, AE	No
T5.5	Mapping of national quality assurance models and good practices in rare and complex diseases in MS	Identify the target population that will be invited to fill the survey. Develop and distribute a survey of national quality assurance models and good practices in rare and complex diseases in MS (including national / international similar networks). Data analysis. The survey will be articulated in:	AOUP, IOR, Erasmus MC, ISS, HSM, Veneto Region, ASUFC, FPG, NAMSE, UKT, LUMC, MUW, FPS HFCSE,	BEN, AE	No

		Step 1: mapping of already existing indicators on national level used by healthcare governance systems, this should include all MS; Step 2: identify relevant indicators specifically for RD	GUH, MoH-SR		
T5.6	Identification and elaboration of selected and meaningful indicators for a national monitoring of rare and complex diseases primarily focussing on care pathways	Based on the results of the case study, we will identify the best indicators for measuring the impact of each HCP on the care pathways and, ideally, on the patients' outcomes. Each indicator will include at least the criterion, a measurable element, the target to be achieved and the possible exceptions. We will also identify the steps needed for the adoption of ERN-developed/ evidence-based CPGs (link to WP6) and the starting of the process that will conduct to the ERN integration into national healthcare systems.	AOUP, IOR, Erasmus MC, ISS, HSM, Veneto Region, ASUFC, FPG, DGS, UKT, MUW, FPS HFCSE	BEN, AE	No
T5.7	Development of a framework for the collection of national monitoring data	A framework for the collection of national monitoring data will be developed. This will complement the existing collection of monitoring data in the frame of the ERN monitoring process.	AOUP, IOR, Erasmus MC, ISS, HSM, Veneto Region,MUW, ASUFC,NAMSE,OUS UKFFM, UKT, DGS, HSO,FPS HFCSE	AE, BEN	No
T5.8	Collection of national indicators	First national data collection in at least one selected MS including, if applicable, the definition of a collecting institution also responsible for the quality check of the data. Development of a harmonised monitoring / evaluation / certification process of ERN-HCP on the national level across all MS.	AOUP, IOR, Erasmus MC, ISS, HSM, Veneto Region, ASUFC, DGS, UKT, MUW, FPS HFCSE	AE, BEN	No
T5.9	Development of monitoring indicators for ERN integration	Development of national monitoring indicators aimed at mapping the level of integration of the ERNs into the national healthcare systems in Europe. A maximum of 3 indicators will be co-designed together with the ERN Continuous Monitoring and Quality Improvement Working Group and with the involvement of patient representatives from the EURORDIS Amequis	AOUP, IOR, Erasmus MC, ISS, HSM, Veneto Region, ASUFC, FPG, UKT, MUW, FPS HFCSE	AE, BEN	No

		Task Force. The co-design will take into account the previous experience gathered in the ERN Continuous Monitoring data collections, in the current ERN Evaluation as well as in the results of the Amequis project. An initial draft of the putative indicators will be designed with the support of the ERN Continuous Monitoring and Quality Improvement Working Group and with the patient's representatives in a dedicated workshop. A Delphi process will be organised among the ERN Coordinators, the ERN BoMS, with the EURORDIS Amequis Task Force, as well as with DG SANTE to agree on the indicators.			
T5.10	Implementation of the integration indicators into different MS	Pilot implementation of the indicators developed will be organised in at least 3 different MS with different healthcare systems. After the data collection, a refinement of the indicators will be performed to take advantage of the experience gathered in the pilot implementation and to develop the final version of the indicators. The final set of validated indicators will be implemented in at least 8 MS. Based on this, a guidance document will be developed for future data collections, and also for the eventual integration of new indicators dedicated to the integration of the ERNs into the national healthcare systems in the future ERN Evaluation.	AOUP, IOR, Erasmus MC, ISS, HSM, Veneto Region, ASUFC, FPG, UKT, MUW, FPS HFCSE	AE, BEN	No

Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description	Due Date (month number)	Means of Verification
MS6	Actual national governance models	5	IOR	Collection and evaluation of the existing national governance models in the different healthcare systems	14	Report document
MS7	First compendium	5	AOUP	Development of national monitoring	19	Report document

	of complementary indicators			indicators aimed at mapping the level of integration of the ERNs into the national healthcare systems			
Deliverable No (continuous numbering linked to WP)	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month number)	Description (including format and language)
D5.1	Interim progress report (WP5)	5	IOR	R	PU	24	Electronic, English
D5.2	Report including recommendations for national governance models adapted to the different types of national health systems in Europe	5	IOR	R	PU	35	Electronic, English
D5.3	Report including recommendations for quality assurance models adapted to the different types of national health systems in Europe	5	AOUP	R	PU	35	Electronic, English
D5.4	Final implementation report WP5	5	IOR	R	PU	36	Electronic, English

Estimated budget — Resources
See detailed budget table (annex 1 to Part B).

Work Package 6

Work Package 6: National care pathways and ERN referral systems						
Duration:	M1 – M36	**Lead Beneficiary:**	VULSK; co-leads HSE, MUW			
Objectives						
1) To develop recommendations for the organisation of national care pathways for rare and complex diseases interfacing with ERNs, including the recognition of and preferably full compliance with ERN-elaborated evidence-based resources (like Clinical Practice Guidelines), 2) To develop a proposal for referral systems to ERNs; 3) To develop guidelines for the incorporation of CPMS advice into patients' care						
Activities and division of work (WP description)						
Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)	
			Name	Role (COO, BEN, AE, AP, OTHER)		
T6.1	Sign posting of expertise and multidisciplinary care pathways	Development of a sign-posting tool for national expertise and multidisciplinary team access, and linkages to ERNs including a tool to identify pathways for conditions not covered by the country's expertise and/or not included in ERNs	HSE, DGS, MUW, INSERM, THL	BEN, AE	Yes, subcontracting	
T6.2	Model (reference) care pathways for RD or groups of RD	**6.2.1:** Identification of RD or groups of RD for the development of model (reference) care pathways: a survey of patient representatives and ERNs. **6.2.2:** Development of model (reference) care pathways for identified RD or groups of RD using methodology developed by Ward AJ et al, 2022 (https://pubmed.ncbi.nlm.nih.gov/35410222/), that takes into account national, patient-centred	HSE, NAMSE, UKHD, FIBHULP, DGOS, Veneto Region, VULSK, ZonMw, MUW, GUH, THL	BEN, AE	Yes, subcontracting	

		perspective and encompass all health system levels from primary to highly-specialised (to interregional) to cross-border care, intersectoral collaboration, holistic and integrated approach; **6.2.2.1:** Mapping of patients' care trajectories by leveraging on the expertise collected from i) ERNs (highly-specialised expertise) and ii) patient representatives (patient experience-based expertise); **6.2.2.2:** Design and consensus on optimised care pathways.			
T6.3	BEAM (barriers/ enablers/ accelerators/ motivators) for the implementation of care pathways	Identification of BEAM (barriers/ enablers/ accelerators/ motivators) for the implementation of care pathways in the national systems by using Systems Engineering Initiative for Patient Safety (SEIPS 3.0) framework. Methodology: i) surveys and workshops with the CA of this JA and members of the Multistakeholder Advisory Group; ii) interviews with selected national representatives (to reflect five groups of MS: Large/medium federal health systems: DE, FR, BE, NL. Large/medium regionalized health systems: AT, IT, ES. Small/medium EU-14 health systems: DK, FI, GR, IE, LU, NO, PT, SE. Large/medium EU-13 health systems: PL, HU, CZ, BG, RO. Small EU-13 health systems: HR, CY, EE, LV, LT, MT, SK, SI.)	VULSK, all competent authorities (except for Germany: NAMSE, Italy: IOR & Veneto Region)	BEN, AE	No
T6.4	Recommendations and guidelines for the implementation of care pathways	Development of recommendations and guidelines for the implementation of care pathways that take into account diversity across MS (5 groups of MS as per 6.3); **6.4.1:** Development of recommendations for the implementation of care pathways, referral systems to ERNs and models for case management/ care coordination (CA of this JA). Consultation with the National Policy Contact Point Group.	VULSK, all competent authorities (except for Germany: NAMSE; Italy: IOR & Veneto Region)	BEN, AE	No
T6.5	Capacity building and implementation of pilots for care pathways in national	**6.5.1:** Capacity building for the implementation of care pathways in national systems: a workshop	VULSK, all competent	BEN, AE	No

	systems	involving multinational multistakeholder communities; **6.5.2:** Implementation of pilots for care pathways in national systems. Pilots will be structured for care pathways developed in T6.2; participating countries will choose them according to their needs. Participating countries: DE, PT, HU, LT, IE, FI, CZ, RO	authorities (except for Germany: NAMSE; Italy: IOR & Veneto Region)		
T6.6	Capture the lessons learned and develop recommendations for early emergency response as it relates to the needs of people living with a RD	We will analyse common elements and differences of potential scenarios with huge impact on the national healthcare system (including pandemic, local war, earthquake). To this end, we will conduct webinars with patients and advocacy organisations working in Ukraine and other countries. In addition, we will analyse aid agencies preparedness and, based on the results of the analyses, develop recommendations for clinical preparedness in the context of RD and ERN, including sustainment of healthcare support and care pathways.	MUW, HUS, ASUFC, VULSK	BEN, AE	Yes, EURORDIS
T6.7	Capacity building to establish and operate a RD hub in the Ukraine providing national support, as well as links to the ERN level for people living with a RD	We will promote capacity building activities for the new Ukrainian RD hub including training activities, development of procedures to sustain elementary care and treatment needs for people living with a RD in Ukraine to enable the hub to support patients and families nationally or to coordinate medical advice via ERNs including physically cross-border care, where necessary. A report will be prepared with lessons learned regarding the integration of the hub into the ERN system (including care pathways) and provision of cross-border healthcare in a nationwide disaster situation.	MUW, HUS, ASUFC, VULSK,	BEN, AE	Yes, Children's hospital 'Ohmatdyt', EURORDIS

Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description	Due Date (month number)	Means of Verification

MS8	Report including recommendations from T6.6 and 6.7	6	MUW, HUS	A report on lessons learned and recommendations derived from this will be prepared.		35	Report available
Deliverable No (continuous numbering linked to WP)	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month number)	Description (including format and language)
D6.1	Tool for sign-posting of national expertise and linkage to ERN pathways	6	HSE	DEM	PU	25	Sign-posting tool
D6.2	Compendium (blueprint) of model care pathways for RD or groups of RD	6	HSE	R	PU	34	Electronic, English
D6.3	Recommendations for the organisation of national care pathways, referral systems to ERNs and incorporation of CPMS advice for rare and complex diseases	6	VULSK	R	PU	34	Electronic, English
D6.4	Toolkit of best practices in the implementation of care pathways	6	VULSK	R	PU	35	Electronic, English

Estimated budget — Resources
See detailed budget table (annex 1 to Part B).

Work Package 7

Work Package 7: National reference networks and undiagnosed disease programmes or equivalent strategies interlinked with ERN			
Duration:	M1 – M36	**Lead Beneficiary:**	UKT; co-lead MUW

Objectives
1) To support capacity building in MS for the development of NRN or equivalent strategies for rare and complex diseases and their integration with ERN 2) To develop structures and procedures for undiagnosed patients closely linked to ERN on a national and European level

Activities and division of work (WP description)

Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)
			Name	Role (COO, BEN, AE, AP, OTHER)	
T7.1	To perform a state-of-the-art analysis of existing structures, models, and initiatives on member state level	Perform a state-of-the-art analysis of existing structures, models and initiatives for NRN or equivalent strategies on MS level. Analyse results regarding Identification of addressed needs / unmet needs, Interface to ERNs (including how the national and European dimension is (not) balanced), Structure and elements of NRN, or equivalent strategies, National guidance/ governance structure, Key and optional activities, Involvement of patient organisations into national networks and their links to the existing ePAGs,	UKT, MUW, GUH, AUH, NAMSE, ISS, NNGYK, TUH, OUS, DGOS, SoS	BEN, AE	Yes, ACHSE

		funding of networks, assignment process and criteria of network members, Inclusion of non-ERN CoEs and Best practice for MDT establishment and work.			
T7.2	To develop models and recommendations for NRN, or equivalent strategies, for rare and complex diseases and their integration with ERNs	Do a multi-stakeholder consensus process will be implemented to agree on models and re-commendations for NRN or equivalent strategies	UKT, MUW, GUH, AUH, NAMSE, NNGYK, TUH, OUS, DGOS, SoS	BEN, AE	Yes, ACHSE, EURORDIS
T7.3	Capacity building and pilot implementation for national reference networks, or equivalent strategies	Organise capacity building workshops based on the analysis and development status of different member states. Implement pilot steps for national networks, or equivalent strategies, which will focus on the establishment of national networks, or equivalent strategies, or on certain aspects directly linked to these structures.	UKT, MUW, GUH, AUH, NAMSE, ISS, NNGYK, TUH, OUS, DGOS, DGS, UKW, UKFFM	BEN, AE	Yes, ACHSE, EURORDIS, Medical University Innsbruck
T7.4	Perform a state-of-play analysis of existing structures and models for undiagnosed patients on a national and European/international level	Perform a survey in participating countries. Identify models in non-EU countries, supranational projects. Analyze results regarding structure, funding, national governance, embedding in national healthcare systems, capacity, inclusiveness, international context, outcome data	MUW,CCUH,ECCHC, ISS, NNGYK, RUMC, UHCZ, UKFFM, UMCL, UKT, AUH, UHCZ	BEN, AE	Yes, EURORDIS
T7.5	Develop recommendations for national UDP or equivalent strategies and their integration with ERN	Identify key and optional elements of UDP based on T7.4. Develop universally applicable models and tools. Develop recommendations for integration of UDP into national healthcare systems and international referral (including ERN and external resources) together with WP7.1 and 6, and for national governance with WP5.1.	MUW, CCUH, ECCHC, HSE, RUMC, ISS, ASUFC, NNGYK, SoS, UKFFM, UMCL, VULSK, UKT	BEN, AE	Yes, EURORDIS
T7.6	Develop a concept for an ERN-overarching expert panel for undiagnosed cases comprising all 24 ERN	Based on and extending Solve-RD structures, develop procedures for accepting patients for panel discussion, organizing regular case discussions, further referral within ERN and/or external resources for undiagnosed patients, and	MUW, CCUH, ECCHC, HSE, RUMC, NNGYK, SoS, UMCL, UKT, UHCZ	BEN, AE	Yes, EURORDIS

		follow-up and recall.			
T7.7	Define a core data set for a registry of undiagnosed patients labelled with ORPHAcode 616874, develop recommendations for a registry of undiagnosed patients in Europe	Adapt the European Rare Disease Set of Common Data Elements to accommodate undiagnosed patients by analyzing UML options, defining additional data elements, and consenting a core data set for undiagnosed patients in Europe. Develop recommendations for a European undiagnosed registry by analyzing pros and cons of central vs. de-centralized data storage and defining common data elements.	UKFFM, APHP, HSE, ISS, MUW, SoS, DGOS	BEN, AE	No
T7.8	Develop a European SOP for assigning ORPHAcode 616874 (undiagnosed RD) in CoE and ERN-HCP	Define minimal requirements for the use of this code in order to harmonize its application, specify the access criteria for an ERN-overarching expert panel for undiagnosed cases, and lay the foundation for a homogenous European undiagnosed patient cohort.	UKT, APHP, MUW, Veneto Region, DGOS	BEN, AE	No
T7.9	Identify requirements for national teleconsultation systems, monitor the usability of the new CPMS in expert panels for undiagnosed cases, develop recommendations for its use in national UDP	Define requirements for national teleconsultation systems by surveying all MS. Monitor the evolving new CPMS with respect to its utility in national and international expert panels for undiagnosed cases (including GDPR compatibility), develop recommendations for the use in national UDP. Accompany developments in participating countries to ensure compatibility with new CPMS	UKW, CCUH, ECCHC, MUW	BEN, AE	No
T7.10	Develop recommendations for national patient organizations (PO) for undiagnosed patients	Perform a state-of-play analysis of existing PO for undiagnosed patients in Europe. Identify specific needs of undiagnosed patients, elaborate recommendations for national PO with the input of the EURORDIS-led Community Engagement Task Force	MUW, SoS	BEN, AE	Yes, EURORDIS, VSOP
T7.11	Perform capacity building and pilot implementations	Capacity building workshops together with WP7.1; Pilot ERN-overarching expert panel: **1)** Identify 100 undiagnosed cases with suspected genetic RD via UDP in DE, EE, NL, RO **2)** re-analysis in data analysis hubs **3)** panel discussions **4)**	MUW, CCUH, ECCHC, HSE, RUMC, ISS, NNGYK, TUH, UKT, UKW	BEN, AE	Yes, EURORDIS, ACHSE, Pro Rare Austria

		evaluation of sustainability/ scalability; Pilot national PO for undiagnosed patients (AT, DE); Pilot national UDP (AT, LV, RO) incl. training at RD Center UKW.			

Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description		Due Date (month number)	Means of Verification
Deliverable No (continuous numbering linked to WP)	**Deliverable Name**	**Work Package No**	**Lead Beneficiary**	**Type**	**Dissemination Level**	**Due Date** (month number)	**Description** (including format and language)
D7.1	State-of-play report NRN / UDP	7	UKT	R	PU	10	Report on results of state-of-play analyses WP7 published on JA website
D7.2	Recommendations and models for NRN or equivalent strategies	7	UKT	R	PU	21	Recommendation document on JA website
D7.3	Recommendations for UDP in Europe	7	MUW	R	PU	21	Recommendation document on JA website
D7.4	Recommendations for a registry of undiagnosed patients in Europe	7	UKFFM	R	PU	21	English document on JA website
D7.5	European SOP for assigning ORPHAcode 616874 (undiagnosed RD) in CoE and ERN-HCP	7	UKT	R	PU	21	English document on JA website

D7.6	Document on recommendations for national patient organizations (PO) for undiagnosed patients	7	MUW / Eurordis	R	PU	21	English document on JA website
D7.7	Implementation report WP7	7	UKT	R	PU	35	Report on pilot implementations WP7

Estimated budget — Resources
See detailed budget table (annex 1 to Part B).

Work Package 8

Work Package 8: Data management			
Duration:	M1 – M36	**Lead Beneficiary:**	DGOS; co-lead VWS
Objectives			
1) To develop recommendations ensuring the interoperability of data structures on MS level (local, regional, national) and ERN level. A supplementary graphic representation of WP8 activities is provided at the end of the description of this work package.			
Activities and division of work (WP description)			

Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)
			Name	Role (COO, BEN, AE, AP,	

				OTHER)	
T8.1	Identification of current barriers to RD data sharing and inventory of existing solutions	**8.1.1:** Baseline study of the journey of RD data for primary and secondary use at HCP, national, and ERN level. The inventory will address the following stakeholders: ERN/NRN HCPs, national/regional authorities, ERN/national RD registries, ongoing European and international health data projects. It will cover the extent of structured data capture by HCPs, the existence, brand and format of EHRs, the use of existing data standards and tools, the level of data FAIRness, the implementation status of Orphacodes, the EUCERD Minimum Data Set and the JRC Common Data Elements, the use of CPMS and other teleconsultation tools, and the legal and regulatory provisions in place for primary and secondary data use at EU/national/local level. **8.1.2:** Identification of the main organisational, technical and legal barriers and existing candidate solutions to integration of data management between National Health Systems and ERNs for RD data sharing based on the results of 8.1.1	GUH, Sciensano, LUMC, INSERM, UKW, DGS, AUH, BfArM, UKHD, RUMC, OUS, HSO, UMCL, THL	BEN, AE	
T8.2	Implementable solutions to improve semantic accuracy and interoperability of RD health data	Propose/develop functional specifications for the implementation of a standardised common RD dataset in health information systems (HIS). **8.2.1:** inventorise and recommend semantic standards to unambiguously capture data (based on previous projects, on agreed Minimum Data Set/Core Data Elements, and results of 8.1), to maximise its use across national healthcare systems and registries for ERNs. Semantic (data standards and data models) and implementation specifications will be defined based on outputs and guidelines from other European projects and eHealth Network guidelines, to which this task will contribute. We will hold 2 workshops with MS representatives focusing on reality-checking the proposed specifications for which feedback from demonstrators (T8.4) will be taken into account. **8.2.2:** will facilitate the implementation of	APHP, AUH, RUMC, LUMC, UKFFM, Sciensano, TUH, UKW, BfArM, DGS, INSERM, Veneto Region, GUH, OUS, HSO, UMCL, THL	BEN, AE	Yes, subcontracting

		ORPHAcodes in HIS by encouraging MS health authorities to use the support and tools available from OD4RD2 and T8.3 to ultimately harmonise European data sharing and analysis. Close interaction between this task and the OD4RD2 project will ensure that OD4RD2 support will be organised according to the concrete implementation roadmap in each MS as decided within this JA, even for those not directly involved in OD4RD2.			
T8.3	Propose and develop implementable solutions to overcome organisational, technical, and legal barriers to integration of national health systems and ERN data management	Following the analysis of T8.1, existing solutions will be assessed for their implementability. Solutions for different settings will be prepared in an agile manner in collaboration with T8.4. Solutions are prepared for IT personnel and data stewards at HCPs and national infrastructures in three main areas: **8.3.1:** technical solutions (meta-data and data models, software specifications, APIs, IT tools) facilitating the communication between HCP data management systems and RD registries, eHealth tools, and health data spaces for RD, building on FAIR principles and work of for instance EJP-RD on distributed data resources and X-eHealth on exchange of EHR-type data; **8.3.2:** instruments facilitating legitimate (re)use of data, encompassing common consent elements and standard data sharing and transfer agreements for primary and secondary use in accordance with EU and national data protection regulations, also including methods to create and apply digitised access policies; **8.3.3:** ERN-supporting data management policies and procedures for mitigating organisational barriers faced by HCP and national data stewards.	INSERM, UKFFM, APHP, BfArM, DGS, UKW, Sciensano, Veneto Region, GUH, LUMC, UKHD, UMCL, THL	BEN, AE	Yes, subcontracting
T8.4	Testing and implementing integration solutions in agile mode	3 pilot projects will ensure that the deliverables of 8.2 and 8.3 are relevant and can be fully deployed in an easy manner: **8.4.1:** Implementing T8.2 and 8.3 outputs across samples of HCPs among 4 pilot ERNs (EpiCARE, EURO-NMD, ERN-RND,	APHP, HCL, UKT, GUH, LUMC, RUMC, Sciensano, DGS, Veneto Region, UKHD, AUH,		Yes, subcontracting

		ITHACA). A call for volunteers will be launched among HCPs involved in the 4 ERNs and the corresponding National Health Data Spaces (NHDS) using T8.1 to ensure that selected HCPs sufficiently represent the variety in Europe. **8.4.2:** Communicating data from EHR datasets for use in CPMS. In parallel to the development of the new CPMS, data required for case discussions (i.e., data from the RD common data set, DICOM files, etc.) will be mapped to available sources in EHRs of ERN members and national clinical case consultation systems. A transfer of data from EHRs of HCPs into the new CPMS will then be piloted in 6-7 HCPs from 4-5 countries, in addition to transfer from the German clinical case consultation system KONSIL-SE. Data transfers will be adjusted as the new CPMS evolves. National privacy preserving requirements and GDPR will be followed. **8.4.3:** Use of implemented RD datasets for monitoring HCP and ERN activities and for data exchange with ERN registries. This demonstrator will focus on (a) the re-use of prospective and retrospective data to assess the added-value of ORPHAcode implementation in measuring HCPs and ERNs KPIs; (b) the implementation of recommendations issued from X-eHealth for EHRs data extraction for registries. Conducted in Denmark and open to voluntary countries. It will follow 4 steps: 1) technical implementation, 2) coding capacity building (Y1); 3) actual data coding (Y2); 4) data extraction and analysis (Y3).	INSERM, UKW, MoH SR, SoS		
T8.5	Visualisation of RD Expert Centres with ERNs and NRNs	Visualisation of data about RD HCPs and patient organizations will be adapted to target people living with a RD, based on the harmonization of the data set and data model of Orphanet with those of SE-ATLAS. An individual entry point per country will be created. **8.5.1:** Mapping the Orphanet expert centres, networks and patient organisations/alliances data set with the generic	UKFFM, INSERM, Sciensano, MUW, HSE, DGS, VULSK, OUS, BfArM, HSO		

		SE-ATLAS data model. An alignment of the database of SE-ATLAS and Orphanet DE, e.g. through addition of optional fields in the Orphanet Database, will be evaluated for allowing a mapping of the data elements for data set alignment. **8.5.2:** Demonstration and evaluation of national visualisation in interested countries. User-centred conceptualization and adaptation of the search tool based on SE-ATLAS technology for patients & experts, considering T6.1 specifications. Visualisation of national ERN and NRN members. Evaluation of transferability of health care driven SE-ATLAS structure to other European countries. Include retrieving feedback from patients and doctors with the help of Focus Group Meetings, including Orphanet national teams in participating countries. **8.5.3:** Definition of data transfer between Orphanet and SE-ATLAS and definition of individual national search start pages. API to be checked and adapted according to the requirements. Implementation of a feedback loop for quality assurance: User feedback to editors. Individual national entry points, including a translation engine for enabling barrier free access. **8.5.4:** Patient-centred easy/fast-to-use design. In addition to the search possibilities via search bar and ERN/NRN-overviews, a specialised search opportunity primarily for ultra-rare diseases will be conceptualised and developed (using thematic groupings). Work will consider the concepts from T6.1. **8.5.5:** Demonstration and evaluation of national visualisation in interested countries. Individual entry points per country in national language, including customised background information and links to relevant websites will be realised and evaluated. Data entry and curation are demonstrated and evaluated, as well as accuracy in data visualisation and providing country-specific views via the SE-Atlas successor and Orphanet. **8.5.6:** Determine models and strategies for long-term sustainability.			

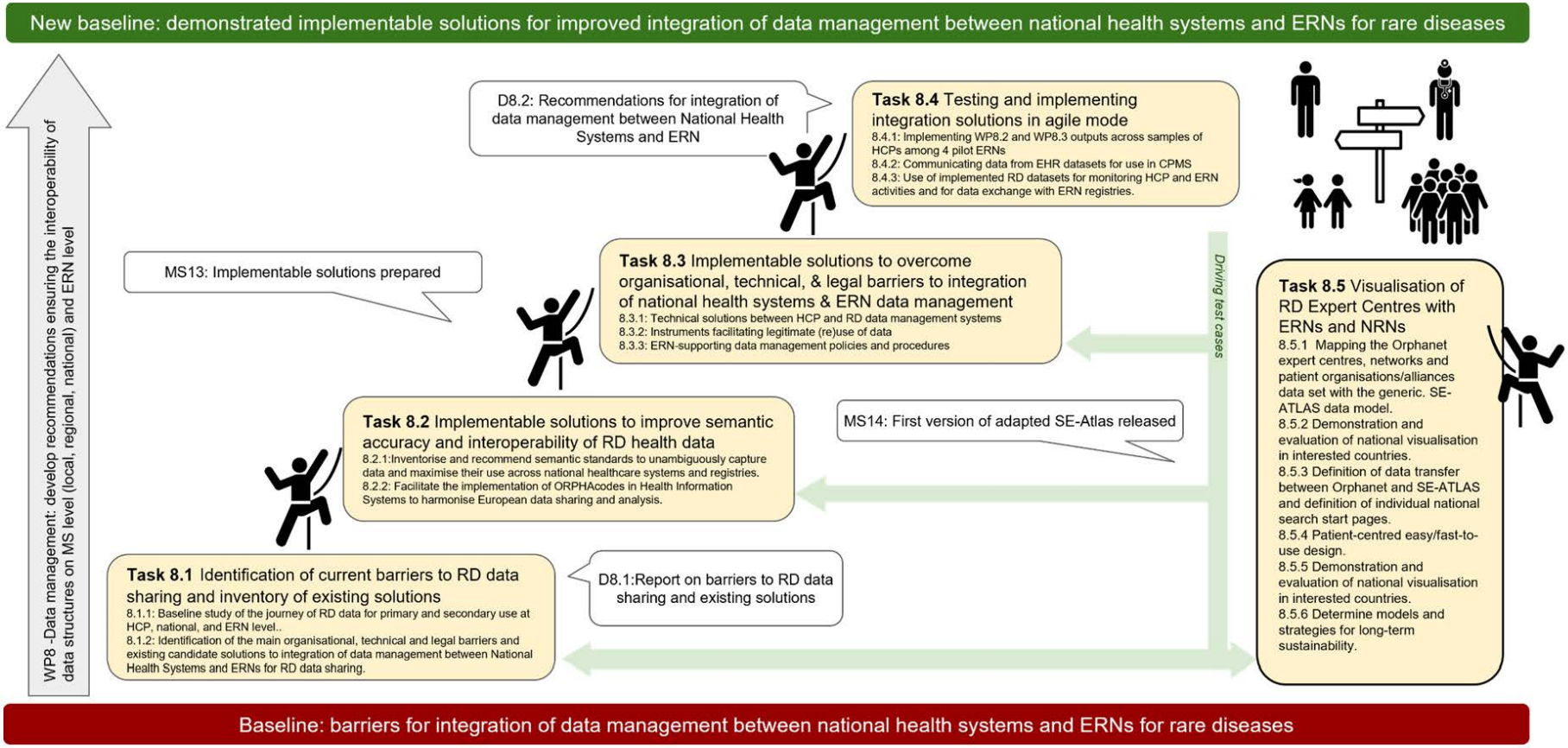
Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description	Due Date (month number)	Means of Verification
MS9 – M11	Implementable solutions prepared	8	LUMC, APHP, INSERM	Preparation of implementable solutions in MS including implementation guidelines issued in 3 release cycles	12, 21, 30	Review by designated WP8 reviewers
MS12	First version of adapted SE-Atlas released	8	UKFFM	SE-ATLAS with national entry points and inclusion of Orphanet data is published and accessible through Orphanet	24	SE-Atlas accessible through Orphanet for at least one country

Deliverable No (continuous numbering linked to WP)	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month number)	Description (including format and language)
D8.1	Report on barriers to RD data sharing and existing solutions	8	UKHD	R	SEN	15	Documentation report available in English
D8.2	Recommendations for integration of data management between National Health Systems and ERN	8	LUMC, INSERM	R	PU	35	Strategic concept paper published in English in the JA website

Estimated budget — Resources

See detailed budget table (annex 1 to Part B).



Supplementary Figure 2: Work Package 8. The objective of WP8 is to develop recommendations ensuring the interoperability of data structures on MS level (local, regional, national) and ERN level. WP8 follows a process that acknowledges the diversity between member states. Current barriers for integration per MS are analysed and existing solutions to address them are inventoried (Task 8.1, D8.1); existing solutions are tested and where needed reprocessed and documented to become implementable solutions for health systems in MS (Tasks 8.2, 8.3; MS13); improvement upon the baseline is tested and demonstrated in MS and HCPs selected using the baseline study in close collaboration with ERNs (Task 8.4). In parallel, a tool that helps patients and non-specialised medical professionals to find RD experts and expert centres is deployed to work in all MS (Task 8.5; MS14). Integration stewards will play an important role in developing guidance making WP8 recommended solutions implementable in national health systems of MS (Tasks 8.1-8.5; D8.2). The driving test cases (Task 8.4) are used throughout WP8, from helping define scope and requirements (Task 8.1) to testing and refining solutions (Tasks 8.2-8.5).

Work Package 9

Work Package 9: National support options for ERN-HCP			
Duration:	M1 – M36	**Lead Beneficiary:**	GUH
Objectives			
1) To collect and analyse good practices and mechanisms to provide support to ERN-hosting healthcare providers at national level as well as to individual ERN centres at the hospital (HCP) level, 2) To develop specific recommendations for: 1. the national support to healthcare providers participating in ERNs, 2. the hospital support to individual ERN centres, 3. the CPMS service and reimbursement models			
Activities and division of work (WP description)			

Task No (continuous numbering linked to WP)	Task Name	Description	Participants		In-kind Contributions and Subcontracting (Yes/No and which)
			Name	Role (COO, BEN, AE, AP, OTHER)	
T9.1	To collect and analyse good practices and mechanisms to provide support to ERN-hosting healthcare providers at national level as well as to individual ERN centres at the hospital level	**9.1.1:** Survey and mapping of existing mechanisms to support ERN-related activities of ERN-centres at national/regional/hospital level. To be coordinated with WP 5, 6 and 8.1. **9.1.2:** Identification of ERN-related activities requiring support at a national/regional/local level (administrative, organisational and/or financial) and of potential mechanisms to strengthen the sustainability of ERNs: An exhaustive inventory of items. **9.1.3:** In-depth analysis of previous work on CPMS utility and reimbursement models, in particular analysis of the recent ERN pilots, initiated and supported by the European Commission, using different reimbursement strategies / concepts. Feasibility of CPMS in real-world clinical practice. **9.1.4:** Analysis of legal ways for the implementation of transnational	GUH, UKHD, UKFFM, DGOS, NAMSE, ASUFC, Veneto Region, VULSK, TUH, VWS, MoH-SR	BEN, AE	No

		CPMS reimbursement models based on existing national reimbursement mechanisms. **9.1.5:** General analysis of cost-effectiveness/economic impact within the healthcare system by integrating ERN HCPs, reality-based cost analysis / developing of pilot health economic model.			
T9.2	To develop specific recommendations	**9.2.1:** Development of specific recommendations for the national support to healthcare providers participating in ERNs – selection from the 9.1.2 inventory reached by multistakeholder consensus. **9.2.2:** Development of specific recommendations for the hospital support to individual ERN centres - selection from the 9.1.2 inventory reached by multistakeholder consensus. **9.2.3:** Development of specific recommendations for the CPMS service and reimbursement models. Based on the results of 9.1.3 and 9.1.4.	GUH, UKHD, UKFFM, DGOS, MUW, NAMSE, TUH, NPHO, DGOS, ASUFC, Veneto Region, MOH-HR, HSE, VULSK, MFH, MOH-PL, MOH-SR, FIBHULP, all competent authorities	BEN, AE	No

Milestones and deliverables (outputs/outcomes)

Milestone No (continuous numbering not linked to WP)	Milestone Name	Work Package No	Lead Beneficiary	Description	Due Date (month number)	Means of Verification
MS13	Analysis completed	9	GUH	Analysis (9.1.1-9.1.5) report	24	Report available

Deliverable No (continuous numbering linked to WP)	Deliverable Name	Work Package No	Lead Beneficiary	Type	Dissemination Level	Due Date (month number)	Description (including format and language)
D9.1	Report on existing mechanisms to support ERN-related activities of ERN-HCPs in MS	9	UKHD, UKFFM	R	PU	18	Electronic, English
D9.2	Guidance document on the	9	GUH	R	PU	36	Electronic, English

	requirements for national support of ERN-HCPs						
D9.3	Recommendations for CPMS reim-bursement models	9	GUH	R	PU	35	Electronic, English

Estimated budget — Resources
See detailed budget table (annex 1 to Part B).

Subcontracting

Subcontracting

Give details on subcontracted project tasks (if any) and explain the reasons why (as opposed to direct implementation by the Beneficiaries/Affiliated Entities).

Subcontracting — Subcontracting means the implementation of 'action tasks', i.e. specific tasks which are part of the EU grant and are described in Annex 1 of the Grant Agreement.

***Note:** Subcontracting concerns the outsourcing of a part of the project to a party outside the consortium. It is not simply about purchasing goods or services. We normally expect that the participants have sufficient operational capacity to implement the project activities themselves. Subcontracting should therefore be exceptional.*

Include only subcontracts that comply with the rules (i.e. best value for money and no conflict of interest; no subcontracting of coordinator tasks).

Work Package No	Subcontract No (continuous numbering linked to WP)	Subcontract Name (subcontracted action tasks)	Description (including task number and BEN/AE to which it is linked)	Estimated Costs (EUR)	Justification (why is subcontracting necessary?)	Best-Value-for-Money (how do you intend to ensure it?)
General remark: Due to the page/space limitation in Form B the description of subcontracting is kept to a minimum. For more detailed information on the activities covered by the subcontracts (including task explanations) please refer to the budget tables of the related BEN/AE (chapter subcontracting: description of activities and background).						
WP1	S1.1	Translation service	Translation of all docu-ments developed in the Action from all WPs (DGS)	12 000,00	National necessity to provide all documents in native language	Choose best offer

WP2	S2.1, S2.2, S2.3	Blueprint pilots	Organisation of information campaigns about ERNs (HSE, IOR, ECCHC)	10 000,00 HSE + 39 000,00 IOR + 38 000,00 ECCHC	Test of newly developed blueprints for ERN information campaigns	Choose best offer
WP3	S3.1	Evaluation methodology expert	Bojana Gundic providing expertise to whole WP (UHCZ; see also CV BG)	43 106,00	Specific expertise in project management and evaluation activities	High expertise; in addition strengthening the link between BEN and AE
WP5	S5.1	AOP contract	Italian umbrella patient organisation providing services for T5.1 (IOR)	20 000,00	Patient expert contribution and feedback is essential.	Unique expertise
WP6	S6.1	EURORDIS contract care pathways	European patient organisation for RD providing services to T6.2 and T6.6-T6.7 (MUW)	115 450,00	Patient expert contribution and feedback is essential; no eligibility as AE.	Unique expertise
	S6.2	Capacity Building RD Hub Ukraine	Supporting the capacity building of the Ukrainian RD Hub T6.7 (MUW)	82 569,50	Financial contribution to capacity building essential; subcontractors not eligible as AE	Unique expertise
	S6.3	Bethesda Children's Hospital (BCH) contract	Experts from BCH providing services to the Hungarian CA in WP2 activities, T2.1-2.6 (NNGYK)	8 250,00	Senior staff members and junior staff members from BCH will provide services for the equivalent of 1,5 PM and 2 PM, respectively, in WP2 activities: Teaching activities, patient safety, quality assurance, leaflets, webpages, teaching videos, good clinical practice.	Offer by BCH (no competitor in Hungary available)
	S6.4	Care and ERN integration contract	Educational programs for primary care; RD patients' perspective to integration,	5 000,00	Complementing the NNGYK expertise; inclusion of essential	Offer by HUFERDIS (no competitor in Hungary available)

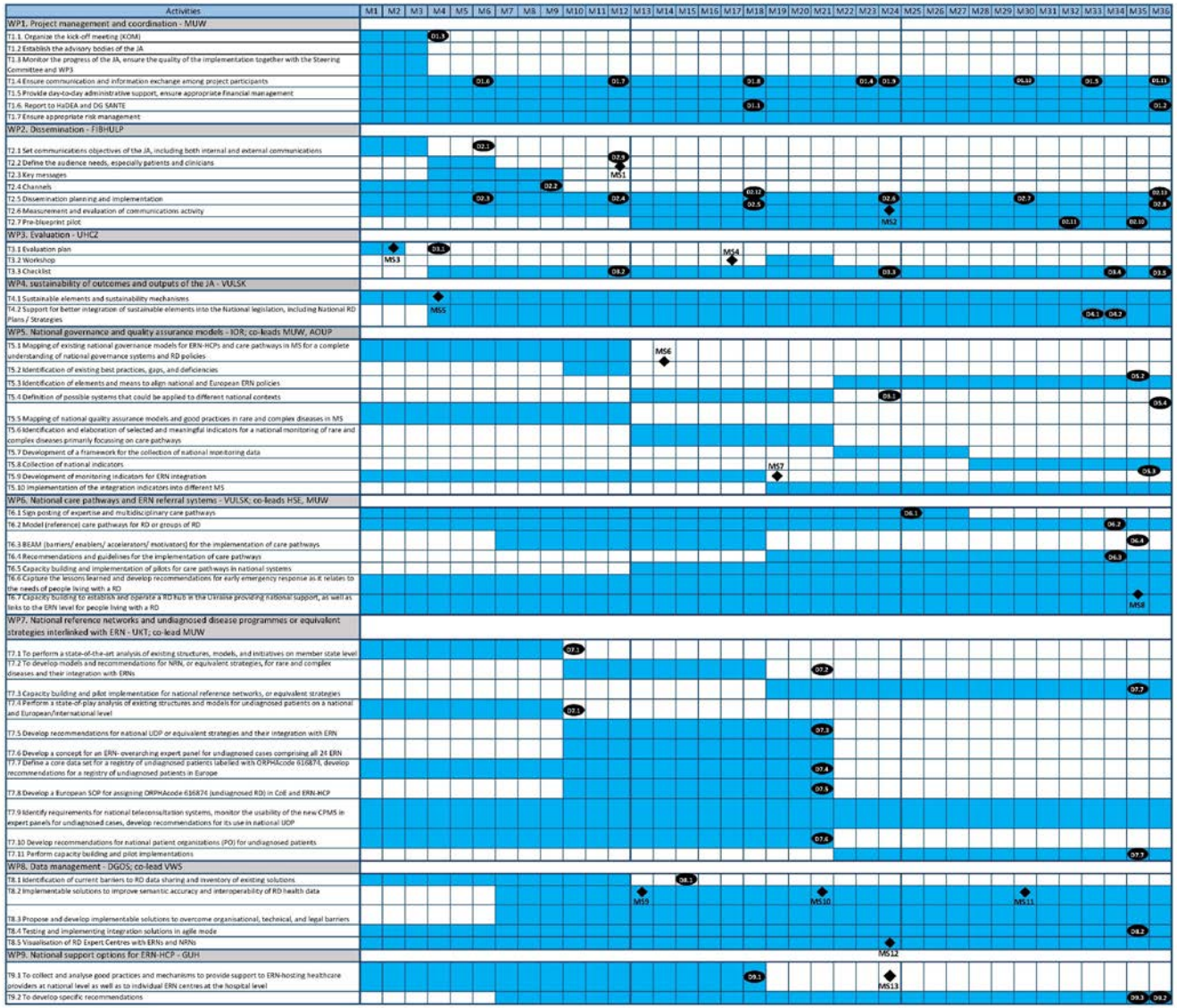
			T6.1-6.2 (NNGYK)		patient expertise	
	S6.5	Rare Disease Ireland contract	Irish umbrella patient organisation providing services to T6.1-6.2 (HSE)	6 000,00	Patient expert contribution and feedback is essential; no eligibility as AE.	Offer by Rare Disease Ireland (no competitor in Ireland available)
	S6.6	Care Pathways Website	Development of a microsite for T6.2 (HSE)	94 000,00	IT service needed for BEN	Choose best offer
	S6.7	International patient leads contract	Providing services to T6.1 and T6.2.2 (HSE; see budget table)	49 500,00	Patient expert contribution and feedback is essential.	Unique expertise
WP7	S7.1	EURORDIS contract UDP	EURORDIS providing services to T7.4-7.6, T7.10-7.11 (MUW)	67 500,00	Patient expert contribution and feedback is essential; no eligibility as AE.	Unique expertise
	S7.2	Pro Rare Austria contract	Austrian umbrella patient organisation providing services to T7.2-7.3 and T7.11; also services to T6.1-6.2 (MUW)	100 000,00	Patient expert contribution and feedback is essential; no eligibility as AE.	Choose best offer and unique expertise
	S7.3	NRN contract Med. University Innsbruck	Capacity building and pilot implementation of 2 NRNs, T7.3 (MUW)	80 000,00	Financial contribution to capacity building and pilot implementation.	Selection of best suited candidates with existing network structures
	S7.4	EURORDIS contract national networks	EURORDIS providing services to T7.2 (UKT)	39 100,00	Patient expert contribution and feedback is essential; no eligibility as AE.	Unique expertise
	S7.5	ACHSE contract	German umbrella patient organisation providing services to T7.1-7.3, T7.12 (UKT)	175 212,00	Patient expert contribution and feedback is essential; no eligibility as AE.	Choose best offer and unique expertise
	S7.6	Rare Diseases Greek contract	Greek umbrella patient organisation providing services to T7.2-3 and	15 000,00	Patient expert contribution and feedback is essential;	Unique expertise

			T7.11 (EODY)		no eligibility as AE.	
WP8	S8.1-S8.4	Demonstrator for agile implementation	Implementation of IT solutions in ERN-HCP in T8.4.1 (HCL [Epicare], UKT [ERN RND], APHP [Neuro-NMD, ITHACA)	100 000,00 (HCL) + 100 000,00 (UKT) + 200 000,00 (APHP [NeuroNMD, ITHACA])	Financial contribution to IT development essential; subcontractors not eligible as AE	Identification of best candidates through survey in T8.1.1-T8.1.2
	S8.5	Demonstrator for data transfer to CPMS	Implementation of IT solutions in ERN-HCP in T8.4.2 (UKW)	120 000,00	Financial contribution to IT essential; subcontractors not eligible as AE	Identification of best candidates through survey in T8.1.1-T8.1.2
	S8.6	EURORDIS contract data management	EURORDIS providing services to T8.1.2 and T8.3.2 (DGOS)	40 000,00	Patient expert contribution and feedback is essential; no eligibility as AE.	Unique expertise
	S8.7	Codevence contract	Contribution to software interfaces for data management systems integration T8.3.1 (LUMC)	60 000,00	Known experience in development of custom tailored IT interface solutions	Choose best offer
Other issues: *If subcontracting for the project goes beyond 30% of the total eligible costs, give specific reasons.*			Insert text			

Timetable

In response to the evaluators' request number 8, we provide on the next page a more detailed Gantt chart of our project, including the visualisation of all deliverables and milestones. For a higher resolution version of this Gantt chart, please refer also to the Other Annexes section (annex 5 to Part B) of this grant proposal.

JARDIN - Time Table



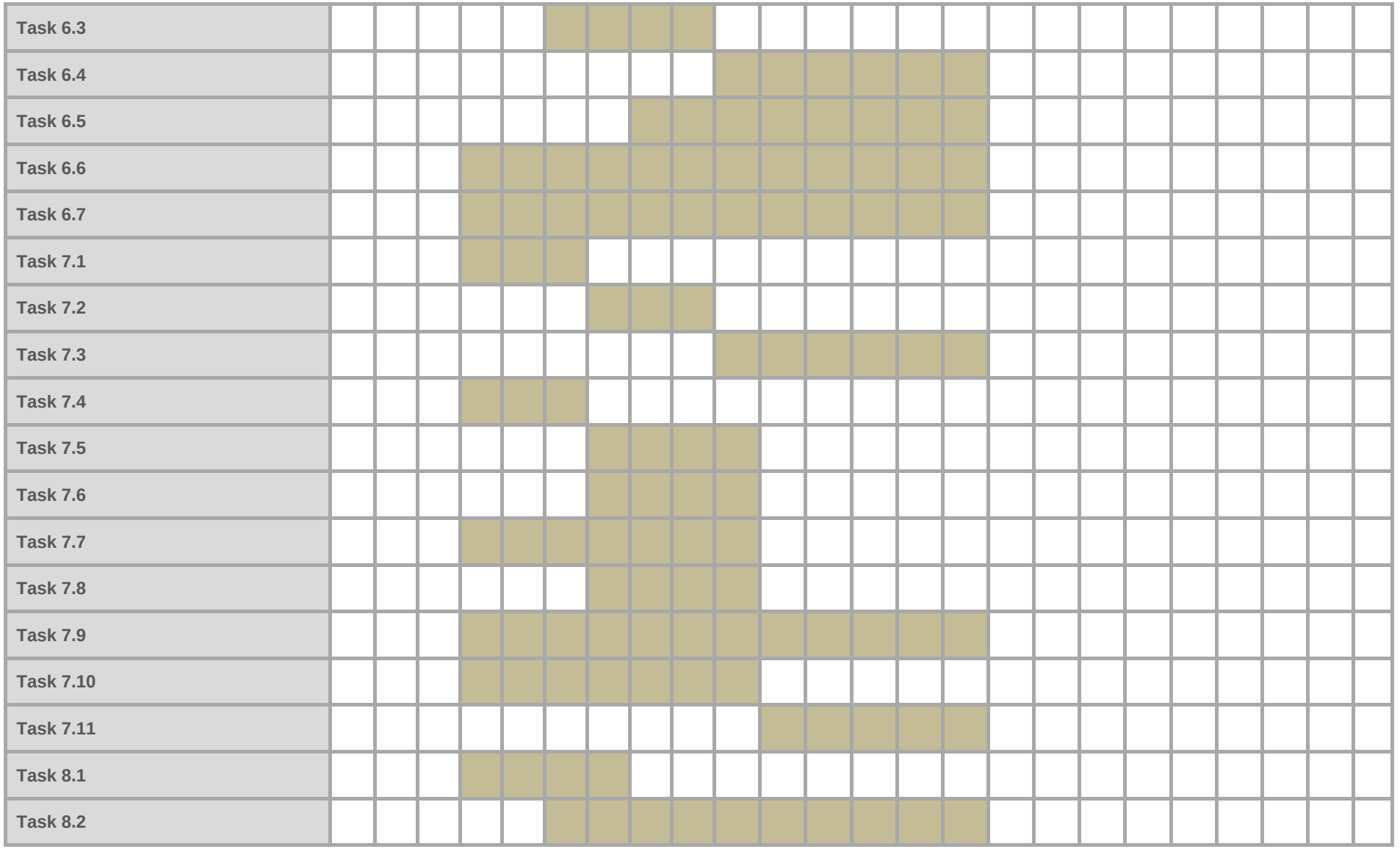
Timetable (projects of more than 2 years)

Fill in cells in beige to show the duration of activities. Repeat lines/columns as necessary.

Note: *Use actual calendar years and quarters. In the timeline you should indicate the timing of each activity per WP. You may add additional columns if your project is longer than 6 years.*

ACTIVITY	2023				2024				2025				2026				YEAR 5				YEAR 6			
	Q 1	Q 2	Q 3	Q 4	Q 1	Q 2	Q 3	Q 4	Q 1	Q 2	Q 3	Q 4	Q 1	Q 2	Q 3	Q 4	Q 1	Q 2	Q 3	Q 4	Q 1	Q 2	Q 3	Q 4
Task 1.1																								
Task 1.2																								
Task 1.3																								
Task 1.4																								
Task 1.5																								
Task 1.6																								
Task 1.7																								
Task 2.1																								
Task 2.2																								
Task 2.3																								
Task 2.4																								
Task 2.5																								
Task 2.6																								

Task 2.7																								
Task 3.1																								
Task 3.2																								
Task 3.3																								
Task 4.1																								
Task 4.2																								
Task 5.1																								
Task 5.2																								
Task 5.3																								
Task 5.4																								
Task 5.5																								
Task 5.6																								
Task 5.7																								
Task 5.8																								
Task 5.9																								
Task 5.10																								
Task 6.1																								
Task 6.2																								


Task 6.3
Task 6.4
Task 6.5
Task 6.6
Task 6.7
Task 7.1
Task 7.2
Task 7.3
Task 7.4
Task 7.5
Task 7.6
Task 7.7
Task 7.8
Task 7.9
Task 7.10
Task 7.11
Task 8.1
Task 8.2

Task 8.3																								
Task 8.4																								
Task 8.5																								
Task 9.1																								
Task 9.2																								

#§WRK-PLA-WP§#

#@ETH-ICS-EI@#

5. OTHER

5.1 Ethics

Ethics *If the Call document contains a section on ethics, describe ethics issues that may arise during the project implementation and the measures you intend to take to solve/avoid them.*
N.A.

#§ETH-ICS-EI§# #@SEC-URI-SU@#

5.2 Security

Security *If the Call document contains a section on security, describe security issues that may arise during the project implementation and the measures you intend to take to solve/avoid them.* *Indicate if there is need for EU classification of information (Decision 2015/444) or any other specific security measures.*
N.A.

#§SEC-URI-SU§# #@DEC-LAR-DL@#

6. DECLARATIONS

Higher funding rate (if applicable)	**YES/NO**
Do you fulfil the conditions set out in the Call document for a higher funding rate? If YES, explain and provide details.	YES
Bodies from all MS participate in this JA, Exceptional Utility Criteria (80% co-funding) apply.	

Double funding	
Information concerning other EU grants for this project ⚠ *Please note that there is a strict prohibition of double funding from the EU budget (except under EU Synergies actions).*	**YES/NO**
We confirm that to our best knowledge neither the project as a whole nor any parts of it have benefitted from any other EU grant *(including EU funding managed by authorities in EU Member States or other funding bodies, e.g. EU Regional Funds, EU Agricultural Funds, etc)*. If NO, explain and provide details.	YES
We confirm that to our best knowledge neither the project as a whole nor any parts of it are (nor will be) submitted for any other EU grant *(including EU funding managed by authorities in EU Member States or other funding bodies, e.g. EU Regional Funds, EU Agricultural Funds, etc)*. If NO, explain and provide details.	YES

Financial support to third parties (if applicable) *If in your project the maximum amount per third party will be more than the threshold amount set in the Call document, justify and explain why the higher amount is necessary in order to fulfil your project's objectives.*
N.A.

#§DEC-LAR-DL§#