

# Alignment of national rare diseases strategies with the European Joint Programme on Rare Diseases

Strategy Meeting

8 July 2021  
ONLINE



## Introduction

The European Joint Programme on Rare Diseases (EJP RD) is based on a joint effort reflected by common initiatives (like for example funding opportunities or trainings) but also through implication of national contributions (for example to funding or infrastructures). Thus, the coordinated undertaking of creation of a RD research European ecosystem must be translated at both national (or even regional) and European (or even international) levels. Therefore, it is expected that the European dimension of the EJP RD should have impact on the activities of the EJP RD itself but also on the undertakings at national and EU level. To monitor and enhance this dimension, the EJP RD Work Package entitled “Integrative Research and Innovation Strategy” (WP2) developed different type of actions. One of the most important activities consists of collection of information from EU Member States (via a survey targeting key persons identified in each country) in relation to the development and implementation of the national plans or strategies on rare diseases.

The **objective of the survey** is to identify relevant/complementary RD-related actions performed at national level, with specific focus put on EU-13 countries with respect to their specific needs, obstacles and advancements.

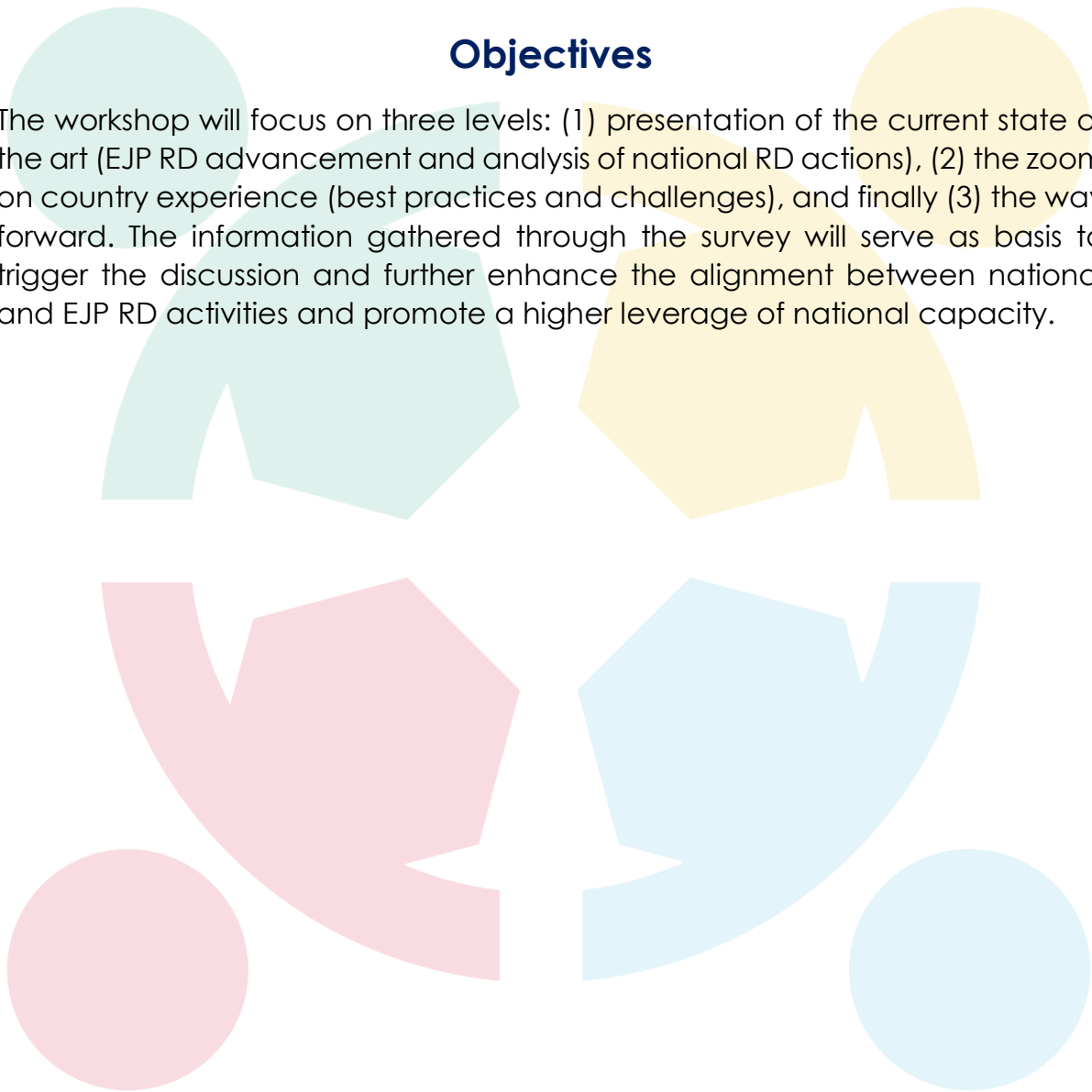
The analysis of the survey allows on identification of points of national alignment with EJP RD, as well as specific gaps that require further actions (on EJP RD

and/or national side). The outcomes and identified needs are transmitted to the National policy makers, so these can translate the identified gaps into actions.

In this context, two “Strategic Workshops” with relevant policy stakeholders were planned since the inception of the EJP RD.

## Objectives

The workshop will focus on three levels: (1) presentation of the current state of the art (EJP RD advancement and analysis of national RD actions), (2) the zoom on country experience (best practices and challenges), and finally (3) the way forward. The information gathered through the survey will serve as basis to trigger the discussion and further enhance the alignment between national and EJP RD activities and promote a higher leverage of national capacity.



## Programme of the Workshop

09:30 Welcome address

Domenica Taruscio, Claudio Carta, Eva Bermejo, Manuel Posada

### 09:45 – 10:45 Session 1 - Setting the scene

09:45 Building the European ecosystem for rare diseases research – how EJP RD leverages the potential of every stakeholder

Daria Julkowska

10:15 Analysis of national state of play and alignment process with EJP RD

Domenica Taruscio, Eva Bermejo, Manuel Posada, Claudio Carta, Laura Lee Cellai, Marta De Santis

### 10:45-13:00 Session 2 - From national to European RD strategies - the country experience

10:45 The experience in Portugal

Carla Pereira

11:05 The experience in Germany

TBC

11:20 Break

11:40 The experience in Romania

Emilia Severin

12:00 The experience in Czech Republic

Milan Macek

12:20 Open Discussion

13:00 Lunch

### 14:00– 16:45 Session 3 – Capturing the national potential - many ways forward

14:00 From national/EU to international level: IRDiRC and Clinical Research Networks

Rima Nabbout, Lucia Monaco,

14:30 RARE2030 - how do we move forward taking stock of lessons learned

Victoria Hedley (TBC)

15:00 Development of the Dutch EJP RD National Mirror Group

Sonja van Weely

15:30 The added value of RD Partnerships

Daria Julkowska

16:00 Open Discussion

16:45 Remarks and Conclusions

Domenica Taruscio, Claudio Carta, Eva Bermejo, Manuel Posada, Daria Julkowska

17:00 End of the meeting

## Speakers

**Eva Bermejo**, Institute of Health Carlos III, Madrid, Spain

**Claudio Carta**, National Centre For Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

**Laura Lee Cellai**, National Centre For Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

**Marta De Santis**, National Centre For Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

**Victoria Hedley** John Walton Muscular Dystrophy Research Centre Newcastle University Translational and Clinical Research Institute International Centre for Life

**Daria Julkowska**, Intitute National de la Santé et de la Recherche Medicale, Paris, France

**Milan Macek**, Charles University-2nd Faculty of Medicine, Prague, Chechia

**Lucia Monaco**, Italian Telethon Foundation, Chair of the International Rare Diseases Research Consortium (IRDIRC), Italy

**Rima Nabbout**, Hôpital Necker Enfants malades, Université Paris Descartes, Institut Imagine, INSERM, France

**Carla Pereira**, Directorate-General of Health, Lisbon, Portugal

**Manuel Posada**, Institute of Health Carlos III, Madrid, Spain

**Emilia Severin**, Carol Davila University of Medicine and Pharmacy, Bucharest, Romania

**Domenica Taruscio**, National Centre For Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

**Sonja van Weely**, Zorgonderzoek Nederland Zon, The Netherlands