

# How European Joint Programme on Rare Diseases can support paediatric research

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**ePTRI**  
EUROPEAN PAEDIATRIC TRANSLATIONAL RESEARCH INFRASTRUCTURE

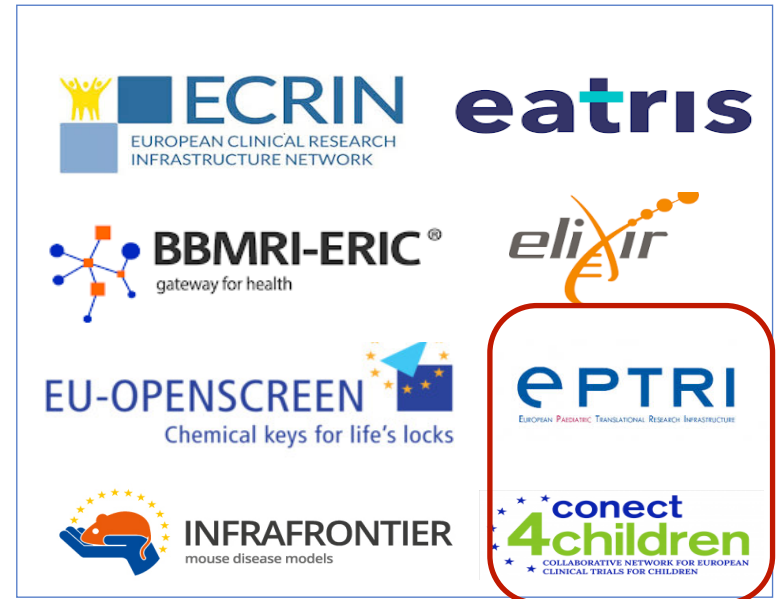
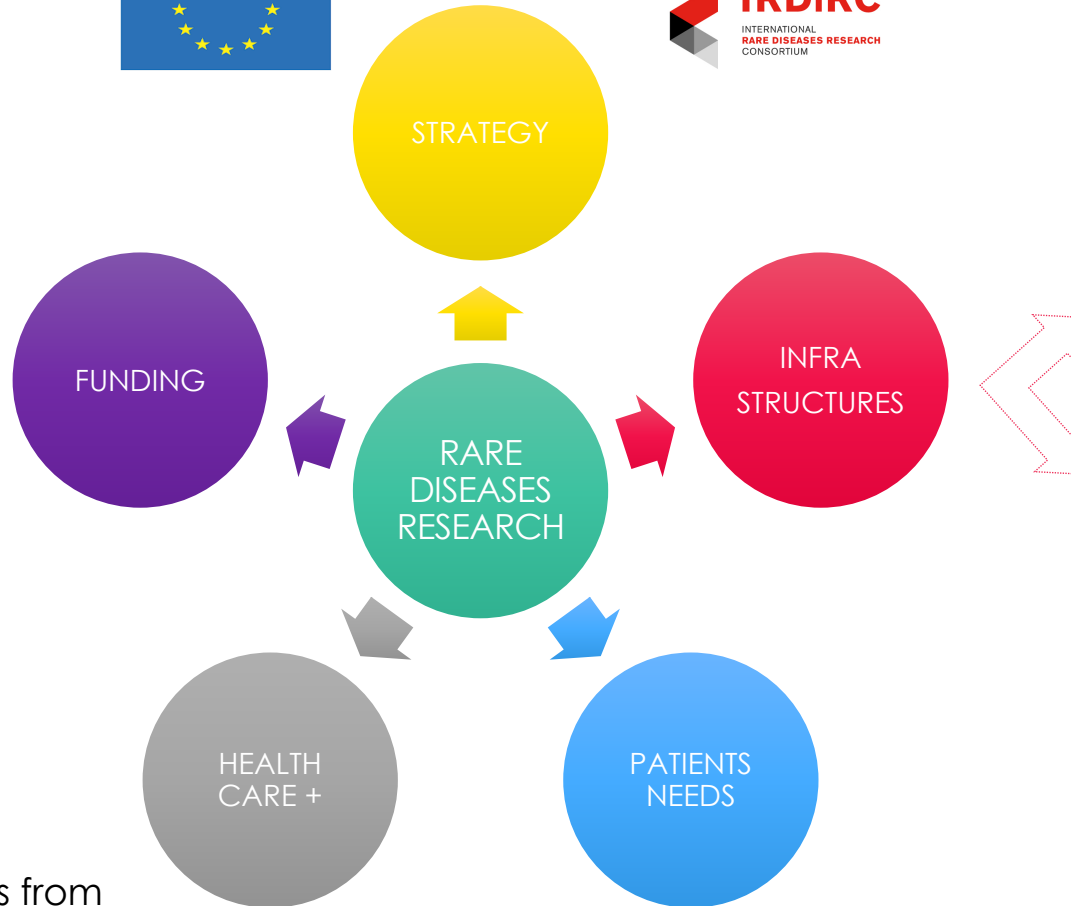
EPTRI Roundtable  
09/07/2020, Online event

# Rare Diseases Landscape in Europe

1.4 billion € for RD research in FP7 & Horizon 2020



25 funders from 17 countries  
Over 120 M€ invested in RD research



More than 900 units from 300 hospitals covering 26 countries



European Reference Networks



869 patient organisations from 76 countries



**85%** of  
**European RD community**  
(directly or indirectly)  
involved in **EJP RD**

**750** people:

**650** Scientifics

**100** Admin

**35**

participating  
**countries**



**87** Beneficiaries:

**9** hospitals

**12** research institutes

**31** research funding bodies/ministries

**24** universities/hospital universities

**5** EU infrastructures

**5** charities/foundations

EURORDIS

+

**50**

**Linked Third Parties**

**And 100% of the associated networks**

**EURORDIS:**

884 RD patient  
organisations  
72 countries

**24 ERNs:**

300 institutions  
>950 healthcare units  
26 countries

**ECRIN-ERIC**

12 main national nodes

**EATRIS-ERIC**

13 main national nodes

**ELIXIR**

220 research  
organisation  
23 partners

**INFRAFRONTIER**

23 partners  
15 countries

**BBMRI-ERIC**

1 international partner  
21 main national nodes  
20 countries

# EJP RD STRUCTURE

Coordinated by



**COORDINATION  
& TRANSVERSAL ACTIVITIES**

**INTEGRATIVE RESEARCH STRATEGY**

**SUSTAINABILITY**

**ETHICAL & REGULATORY**

**COMMUNICATION**

**1**

**FUNDING**

**2**

**COORDINATED  
ACCESS TO  
DATA &  
SERVICES**

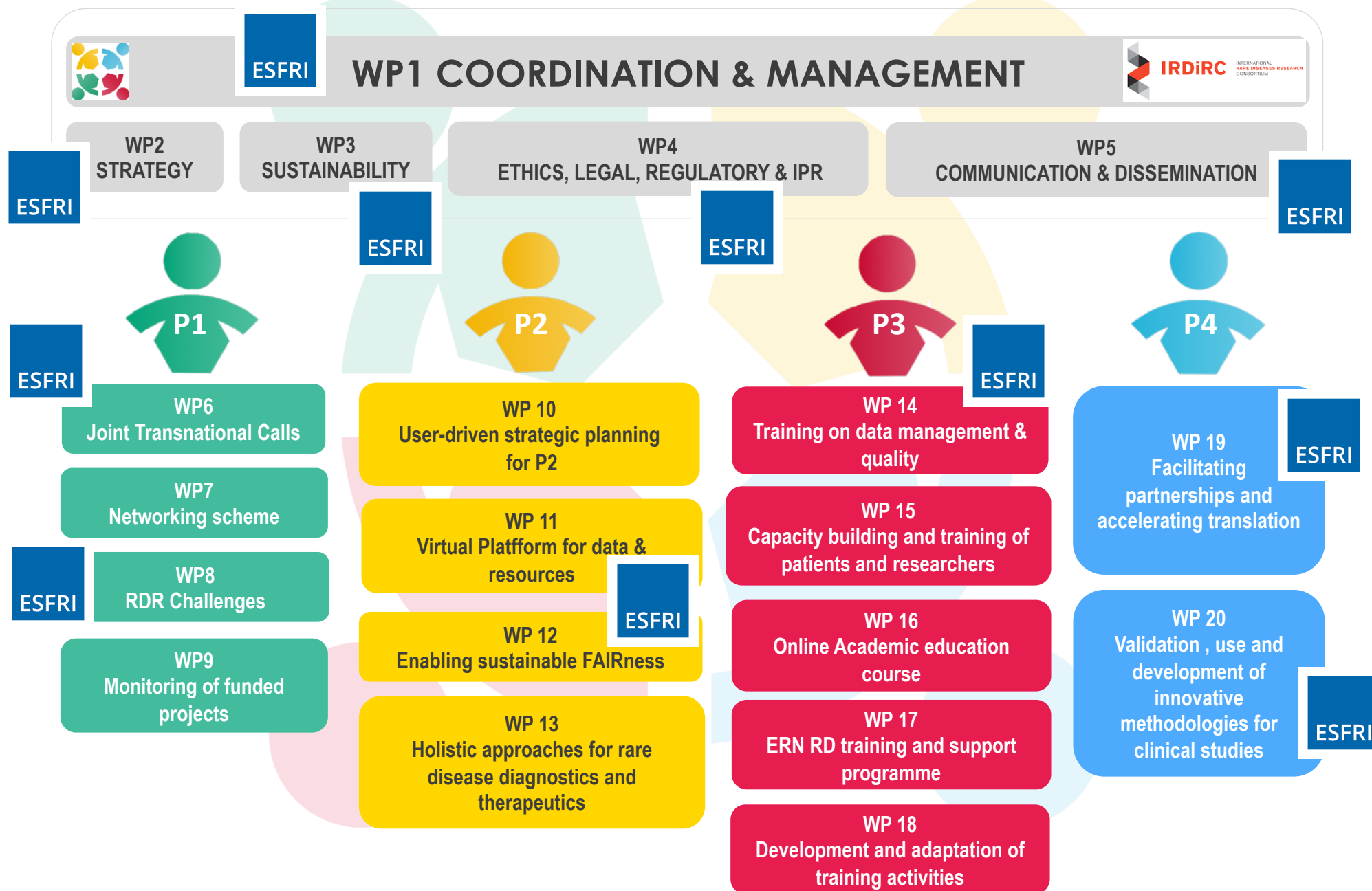
**3**

**CAPACITY  
BUILDING &  
EMPOWERMENT**

**4**

**ACCELERATING  
TRANSLATION  
OF RESEARCH &  
THERAPY  
DEVELOPMENT**

# Why together is better than separately?



# Support for paediatric research in EJP RD (current status)

- ✿ EJP RD Central Helpdesk → support any incoming demand including paediatric research related issues and redirects to specific experts (e.g. EPTRI, C4C, ERNs)
- ✿ Advisory, Regulatory, Ethics Board of EJP RD → supports demands in relation to paediatric research (again internal expertise of involved partners is sought when needed)
- ✿ Pillar 1 funding activities (WP6, WP7, WP8) → no targeted calls but paediatric research on rare diseases can be financed as any other research project
- ✿ Pillar 3 training (WP15) → dedicated training for young patients will take place starting from year 3
- ✿ Pillar 4 innovation management toolbox → any contents specific to paediatric considerations for rare diseases will be described and included
- ✿ Pillar 4 clinical trials support office → run by ECRIN and in close collaboration with C4C and PaedCRIN avoids duplications and ensures paediatric expertise in clinical trials

# EJP RD survey

# Rare disease research community – who?



**European Reference Networks (ERNs)** are virtual networks involving healthcare providers across Europe with the aim to tackle complex or rare diseases. There are 24 thematic networks: over 900 specialised healthcare units located in 313 hospitals in 25 Member States (plus Norway).



Source: [https://ec.europa.eu/health/ern\\_en](https://ec.europa.eu/health/ern_en)



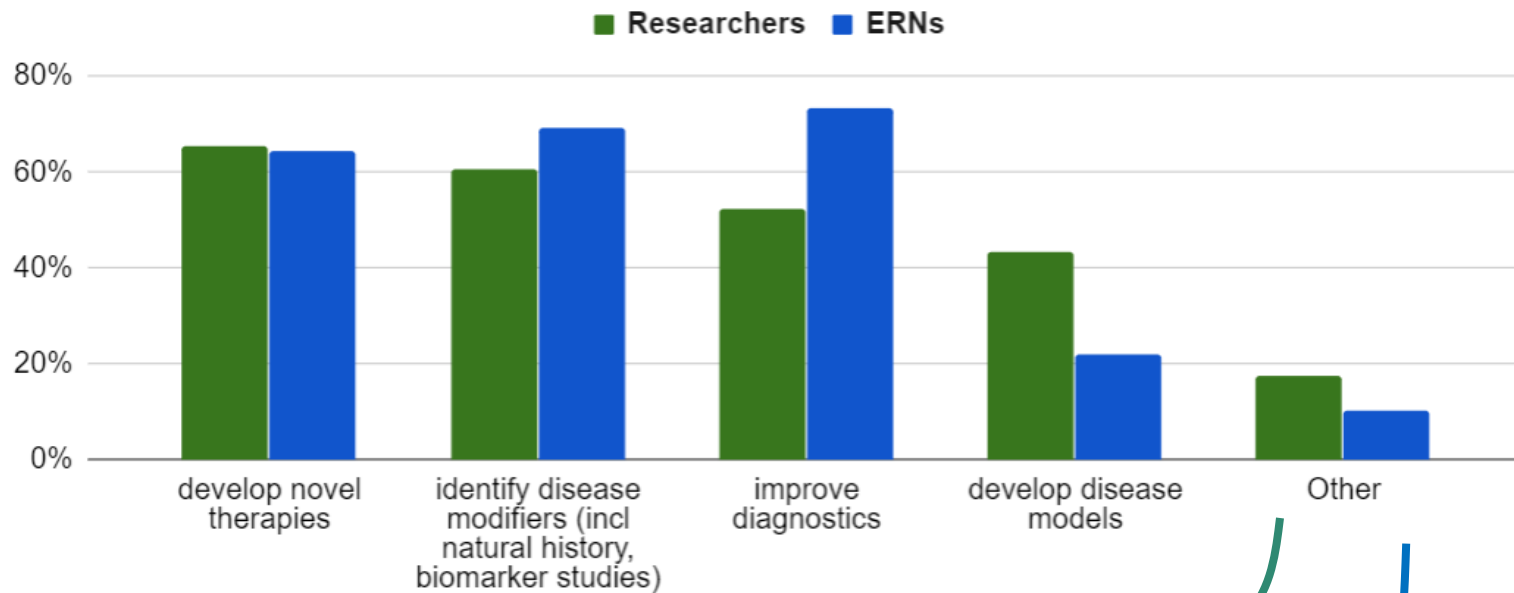
**Rare Disease researchers** are academic scientists in universities and research centres working on any research projects related to rare diseases. They can be working to understand disease mechanisms using disease models as well as translational research. Some EU-level research projects are funded by E-Rare and EJP RD Joint Transnational Calls.

**Stakeholders** being involved for Pillar 2 Use Cases Work Focus:

- ERNs
- Researchers
- Patient representatives
- Funding agencies

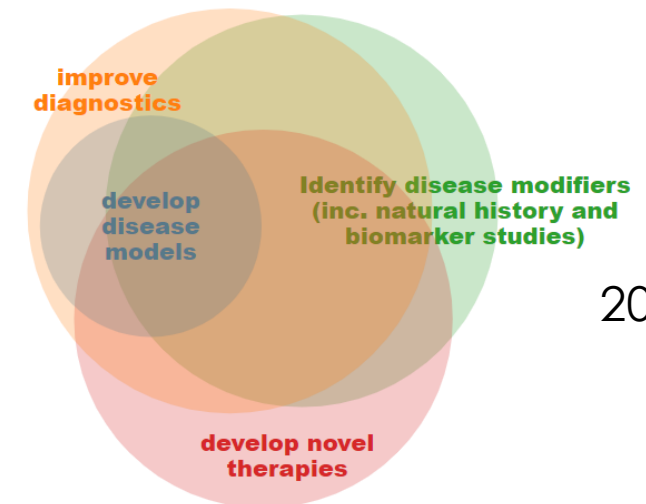
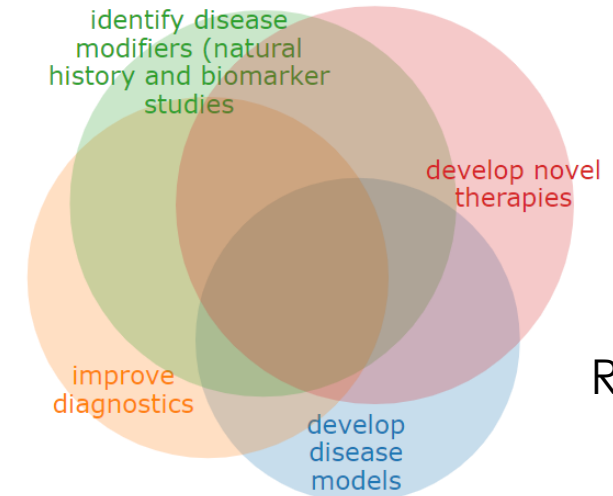


# The main purpose of the current research

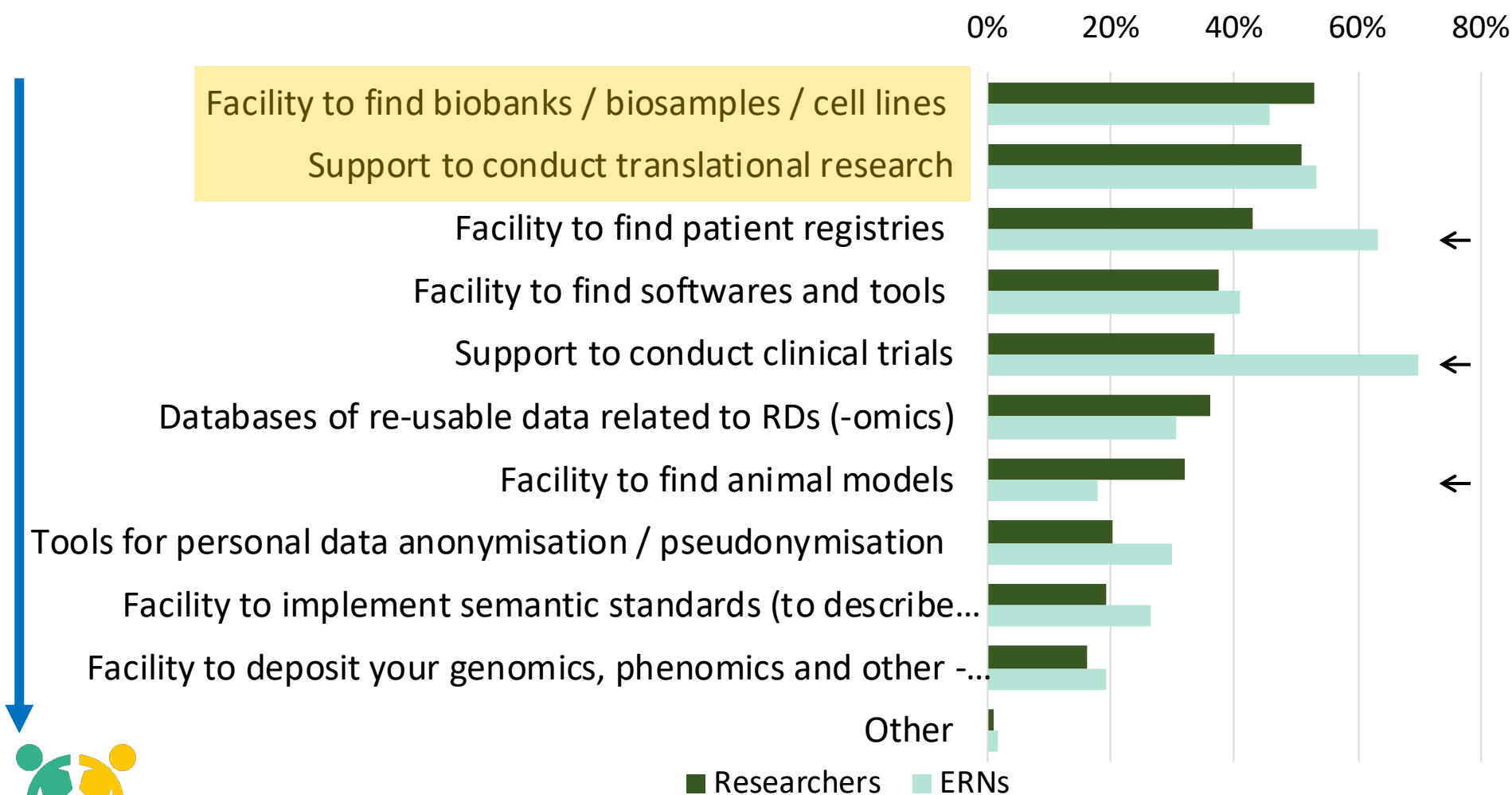


Understanding disease mechanism

Innovative/improve care



# Which of these services are of utmost importance for your research?



Researchers and ERNs require different services, this is most likely due to the differences in the type of research they perform.

# Are these *infrastructures or databases* of utmost importance for your research?

0% 20% 40% 60% 80% 100%

0% 20% 40% 60% 80% 100%

## disease data & experts

Orphanet | Orphadata  
Orphanet | RD expert resources



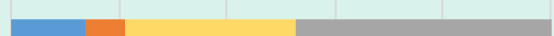
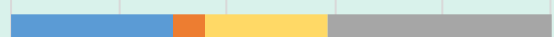
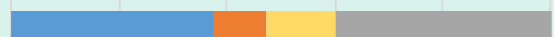
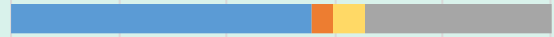
## Registry related

ERDRI  
RaDiCo



## Registry related

International/national registries  
Orphanet catalog of RD resources  
ERDRI  
RaDiCo



## Biobanks, Biological samples

EuroBioBank  
BBMRI-ERIC Directory of Biobanks  
RD-Connect Sample Catalogue  
RD-Connect Registry and Biobank Finder  
BBMRI-ERIC Negotiator  
Cellosaurus  
HPSCreg



## Bioinformatics

ELIXIR EGA  
ELIXIR bio.tools



## Other RIs

INFRAFRONTIER  
ECRIN  
EATRIS



The RD community often **do not** know about existing infrastructures or resources

■ Yes ■ No ■ I don't know this resource ■ No answer

2019 ERNs

# Research infrastructures within EJP RD

# Actions implemented by EJP RD to enhance the usage of research infrastructures by RD community

- ✿ Strong encouragement to use RIs and links to respective infrastructures in EJP RD calls
- ✿ Promotion and contribution of RIs in EJP RD trainings (e.g. BBMRI-ERIC, ELIXIR)
- ✿ Direct services developed in EJP RD by participating RIs:
  - ✿ EATRIS mentoring service and support of innovation and translation of research results open to researchers financed by EJP RD or E-Rare and from ERNs
  - ✿ ECRIN clinical trials support office for RD community
- ✿ Direct involvement of respective RIs in the development of EJP RD Virtual Platform (of data, resources and tools)
- ✿ Organisation of webinars and workshops (at the occasion of EJP RD GA or in collaboration with ERNs and EC) to increase the knowledge about the RIs and their services

# Research infrastructures within EJP RD - NEXT STEPS

# Next steps to be implemented by EJP RD to enhance the usage of research infrastructures by RD community

- 🌐 EJP RD calls → from « encouragement » to « mandatory » (whenever possible) or direct inclusion of the service in the funding cycle (e.g. mentoring service to applicants and not only to funded projects)
- 🌐 More tailored and enhances trainings and communication campaigns
  - 🌐 Trainings developed with and for ERNs (WP17, WP20)
  - 🌐 Mandatory trainings for young researchers participating in funded projects
  - 🌐 Dedicated videos and use cases stories

# Thank you

[www.ejprarediseases.org](http://www.ejprarediseases.org)

[coordination@ejprarediseases.org](mailto:coordination@ejprarediseases.org)

[helpdesk@ejprarediseases.org](mailto:helpdesk@ejprarediseases.org)

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