European Joint Programme on Rare Diseases (EJP RD)
Bringing opportunities to Rare Diseases community

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INSERM

EJP RD coordination team

EUROPEAN JOINT PROGRAMME
RARE DISEASES

VASCERN Days 2019
2019, November 7-8
Brussels, Belgium

Main facts about the EJP RD

Jan 2019 → **Dec 2023**

Total budget (min. submitted): **101 M€** (→ expected > 110 M€)

Union contribution: **55 M€** (70% reimbursement rate)

27 EU MS (AT, BE, BG, CZ, DE, DK, ES, EE, FI, FR, GR, HU, HR, IE, IT, NL, LT, LV, LU, MT, PL, PT, RO, SE, SK, SI, UK), 7 associated (AM, CH, GE, IL, NO, RS, TK) and CA

89 beneficiaries

- 31 research funding bodies/ministries
- 12 research institutes
- 24 universities/hospital universities
- 10 hospitals
- 5 EU infrastructures (BBMRI, EATRIS, ECRIN, ELIXIR, INFRAFRONTIER) + EORTC
- EURORDIS
- 5 charities/foundations (FTELE, AFM, FFRD, FGB, BSF)

+ 50 Linked Third Parties

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Objectives

Main objective:

Create a research and innovation pipeline "from bench to bedside" ensuring rapid translation of research results into clinical applications and uptake in healthcare for the benefit of patients

Mode of action:

Large programme that integrates existing infrastructures, trainings, funding programmes and tools, expands them and develops new essential ones to offer harmonized (and centralized) RD research ecosystem that is easy to use for scientists and produces benefits for patients in the most efficient way

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EJP RD STRUCTURE

Coordinated by Inserm
La science pour la santé
From science to health

COORDINATION
& TRANSVERSAL ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

SUSTAINABILITY

ETHICAL & REGULATORY

COMMUNICATION

1 FUNDING

2 COORDINATED ACCESS TO DATA & SERVICES

3 CAPACITY BUIDLING & EMPOWERMENT

4 ACCELERATING TRANSLATION OF RESEARCH & THERAPY DEVELOPMENT

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WP1 COORDINATION & MANAGEMENT

WP2 STRATEGY	WP3 SUSTAINABILITY	WP4 ETHICS, LEGAL, REGULATORY & IPR	WP5 COMMUNICATION & DISSEMINATION

- WP6 Joint Transnational Calls
- WP7 Networking scheme
- WP8 RDR Challenges
- WP9 Monitoring of funded projects

- WP 10 User-driven strategic planning for P2
- WP 11 Virtual Platfform for data & resources
- WP 12 Enabling sustainable FAIRness
- WP 13 Holistic approaches for rare disease diagnostics and therapeutics

- WP 14 Training on data management & quality
- WP 15 Capacity building and training of patients and researchers
- WP 16 Online Academic education course
- WP 17 ERN RD training and support programme
- WP 18 Development and adaptation of training activities

- WP 19 Facilitating partnerships and accelerating translation
- WP 20 Validation , use and development of innovative methodologies for clinical studies

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Pillar 1: Collaborative research funding

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Pillar 1: Activities

WP6: Joint Transnational Calls for collaborative research projects

Open to research teams from countries with funders involved – min of 4 teams from 4 countries. Topics spanning from pre-clinical, translational to clinical research.

JTC2020

- About the **development of novel therapies in a preclinical setting**
- Consortium submitting a proposal: In 2019 the following conditions were applied (likely to be the same for JTC2020): must involve **4 to 6 eligible partners** from **at least four different participating countries**
- Preliminary dates
  - **November 18, 2019: pre-announcement,**
  - **February 12, 2020: deadline of pre-proposals application**

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Pillar 1: Activities

- **WP7: Networking to share knowledge on rare diseases**
- encourage sharing of knowledge on rare diseases
  - to support health care professionals, researchers and patient advocacy organizations with a networking grant to re-organize themselves into transnational (clinical) research networks
  - that focus on a (group re disease(s), a (group of) rare cancer(s) or on cohorts of undiagnosed patients that are suspected of suffering from a rare disease
- Small support schemes for networking (workshops/events/share of knowledge) – 30K€ max
- open all year long – open to all countries involved in EJP RD
- **1st Networking scheme call will open Q4 2019**

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Pillar 1: Activities

WP8: Rare disease research challenges

Public-private (small scale) partnerships – challenges set by industry and validated by EJP RD – short term (max. 18 months) projects - open to all countries involved in EJP RD

- **pre-launched by the end of 2019 (matching event Jan 14 2020 in Paris)**

WP9: Monitoring of funded projects

Monitoring of all projects funded through EJP RD and previous E-Rare projects

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Pillar 2: Innovative coordinated access to data and services for transformative rare diseases research

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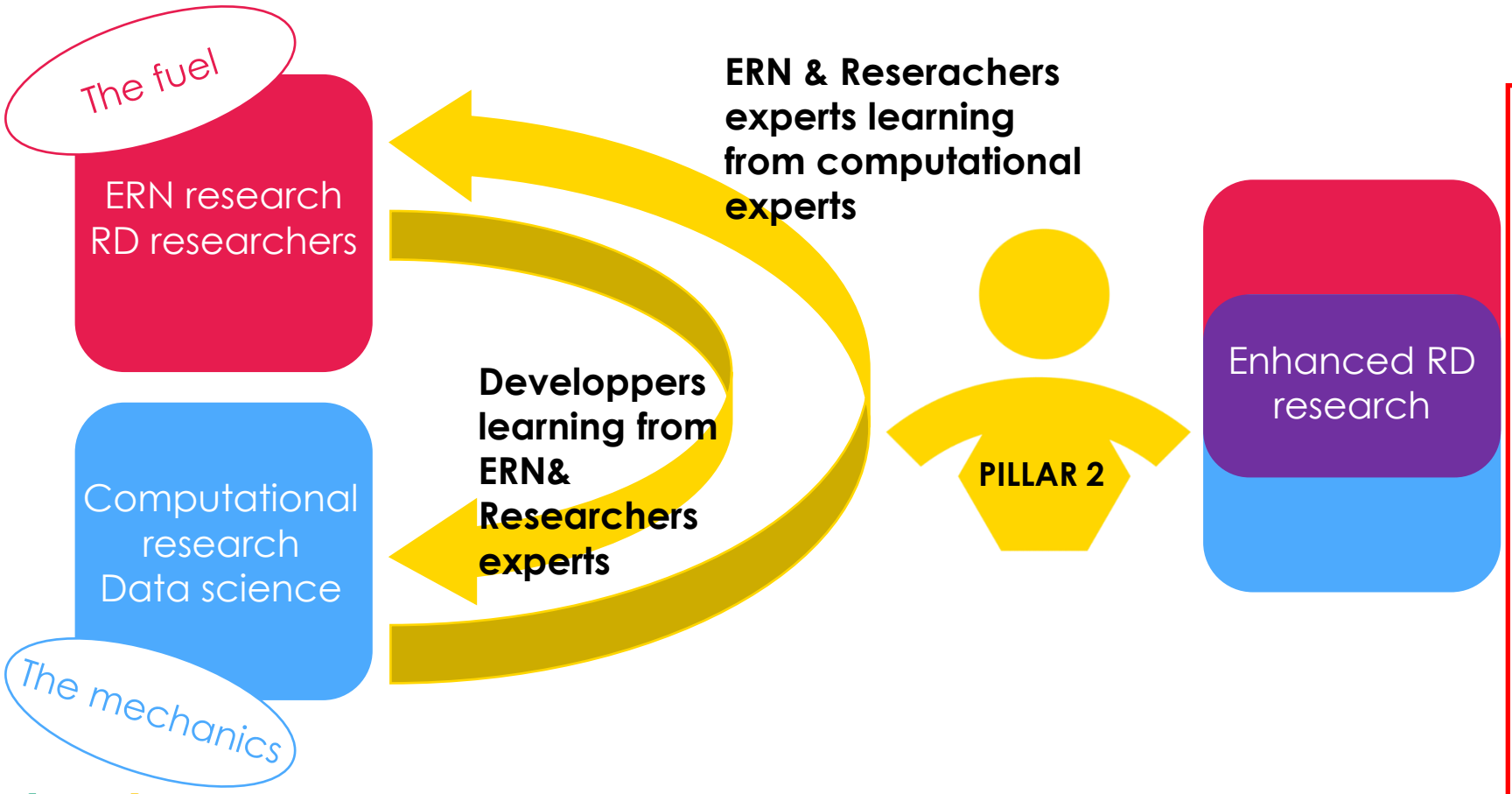
Pillar 2 target: FAIR-based virtual platform



A powerful substrate for translational research:

- **Centralized services for collections (resource-level)**
  - Sample, biobanks, registries, infrastructures and tools catalogue
  - Analysis platform for omics data
  - Curated rare disease-centered information and data

- **Federated services for data elements (record-level)**
  - FAIR 'at source'
  - Data, patients, and samples - linked and discoverable
  - Consents and data use conditions also represented

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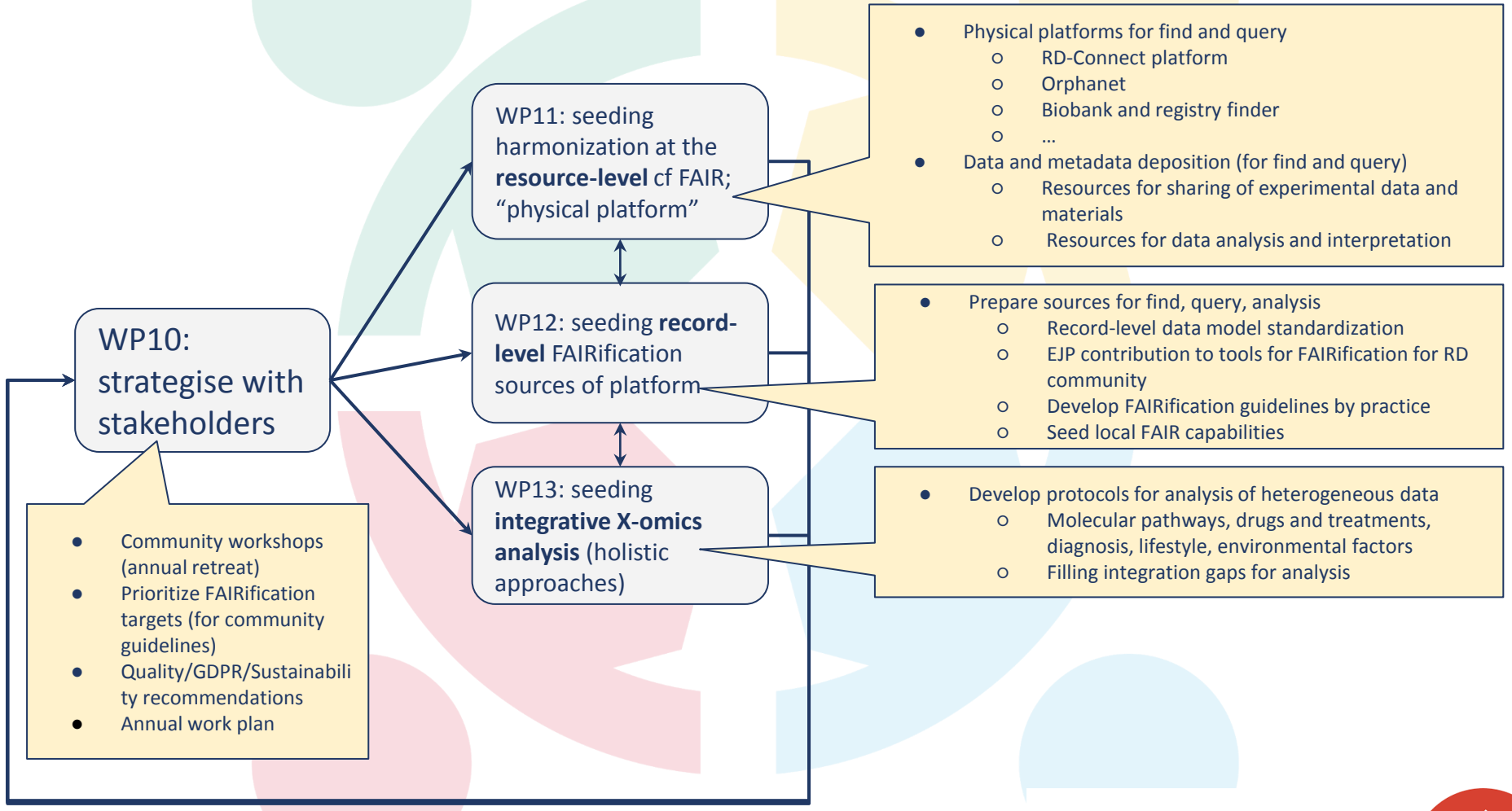
PILLAR 2 WORKFLOW



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Global Alliance for Genomics & Health
Collaborate. Innovate. Accelerate.

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Use Case Work Focus

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The use case work focus…

- *Provides a service*
  - for pillar 2 development
  - **for converting stakeholders' expectations to VP requirements**
- *Describes* use cases, not implement or perform them
- *Helps identify specific stakeholders to engage* more deeply in Pillar 2 R&D on case-to-case basis
- *Takes input from*
  - *stakeholders* (unsolicited use cases)
  - *Pillar 2 developers* (solicited use cases)

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What are use case descriptions?

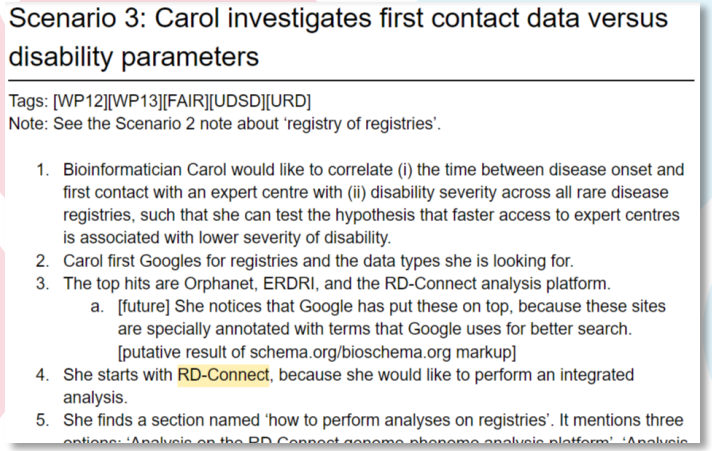
Template questions

As a …, I would like to …, such that I can …

"*As a* **TransplantERN researcher**, *I would like to* **estimate the number of patients** that I could eventually collect, *such that I can* **retrieve a prospective collection of blood samples from a biobank** at the right time point for a project"
courtesy Eduardo Granados

'Alice and Bob' scenarios

Expected experience of stakeholders while working with imagined EJPRD products



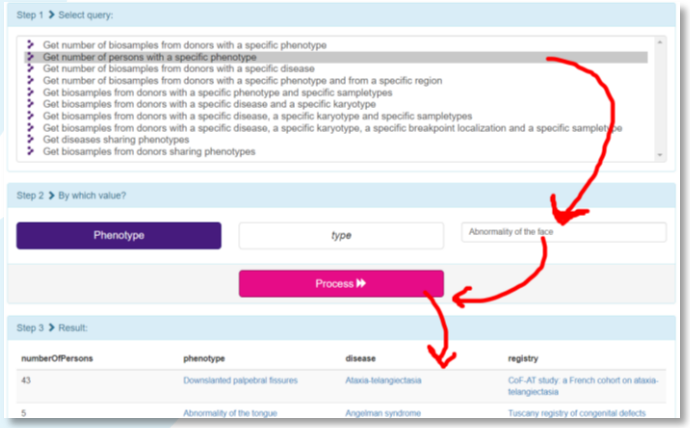
Scenario 3: Carol investigates first contact data versus disability parameters

Tags: [WP12][WP13][FAIR][UDSD][URD]
Note: See the Scenario 2 note about 'registry of registries'.

1. Bioinformatician Carol would like to correlate (i) the time between disease onset and first contact with an expert centre with (ii) disability severity across all rare disease registries, such that she can test the hypothesis that faster access to expert centres is associated with lower severity of disability.
2. Carol first Googles for registries and the data types she is looking for.
3. The top hits are Orphanet, ERDRI, and the RD-Connect analysis platform.
   a. [future] She notices that Google has put these on top, because these sites are specially annotated with terms that Google uses for better search. [putative result of schema.org/bioschema.org markup]
4. She starts with RD-Connect, because she would like to perform an integrated analysis.
5. She finds a section named 'how to perform analyses on registries'. It mentions three

Presentations & mock-ups

Visualisation of what a stakeholder imagines an experience with EJPRD products would look like



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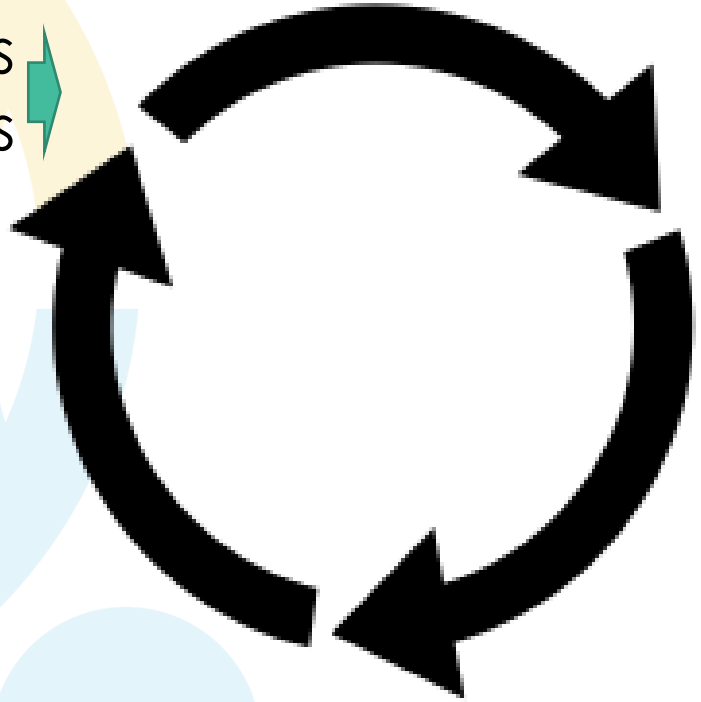
Role of use cases

Use case descriptions
Volunteer stakeholders

Requests for use cases
Results for feedback



Stakeholder representatives
+ P2 mediators
(Use case work focus)

Pillar 2 agile development cycles
(all work foci)

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Request from Pillar 2

We need a simple use case for developing a first 'proof-of-concept' virtual platform

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Levels of voluntary involvement

- fully engaged, volunteer stakeholder-in-the-loop
- involved in defining some use cases
- informed, but not more
- not involved

- Why should you get involved?
  - **converting your expectations to VP requirements**

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ERN registry Task Force

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ERN Registry Task Force' and the 'EJP-RD Registry Interoperability Work Focus'

- Set up to help in the preparation of the call on RD registries
- **Work on topics relevant to harmonizing existing and new registries**
- Further track and help coordinate activities on the metadata model, common data dictionary, digitizing consent, patient/data discoverability, API design, data obfuscation/anonymization, etc.
- **encourage ERNs to commit to adopting standards and policies emerging from all joint ERN-EJP efforts as they build their registries**

registryadvice@ejprarediseases.org

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Workshop and Hackathon

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Workshop and Hackathon: Molecular pathways for rare disease (FAIR) data analysis”

Maastricht on 26 – 29 November 2019

http://www.ejprarediseases.org/index.php/workshop-and-hackathon-molecular-pathways-for-rare-disease-fair-data-analysis/

Focused on:

- **Pathway creation and curation** – especially rare disease and adverse outcome pathways *inspired by needs of ERNs and tox community* (day 1-2)
- **FAIR data preparation** (prep for pathway and network analysis) – define needs of data analysis and do first steps towards solving (day 2-3).
- After the 3 days of workshop, a **hackathon** will take place in which participants will start together solving the identified problems and test different approaches.

- **Cost reimbursement** through the ERN Coordinator's institution
  - For more information on these conditions please get in touch with *secretariat-bigcat@maastrichtuniversity.nl*

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Pillar 3: Capacity building and empowerment

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Pillar 3: Activities

- **WP14: Trainings on data management & quality**

- **Objectives**: Decrease RD data fragmentation and increase data quality which will raise the level of capacities and help data sharing and networking within the RD community (existing and new courses)
- **Courses**: 1) Orphanet nomenclature – 2) standards & quality of genetics/genomics data in clinical practice – 3) strategies to foster undiagnosed diseases – 4) biobanks sample data management – 5) rare diseases registries & FAIRification at source – European Rare Diseases Registry Infrastructure

http://www.ejpraretdiseases.org/index.php/training-and-empowerment/

- **WP15: Capacity building & training of patients and researchers in rare diseases research and processes**

- **Objectives**: Improve RD research & innovation and enhance uptake of research results by building the capacity of the patient community and other key stakeholders (existing and new courses)
- **Courses:** 1) EURORDIS Summer school – 2) EURORDIS Winter School on scientific innovation and translation research– 3) EURORDIS Leadership School on Healthcare and Research – 4) education material and activities for paediatric patients (YPAGs led by Teddy network, EURORDIS participates)

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Pillar 3: Activities

- **WP16: Online academic education course**
- **Objectives:** Provide a EU-wide streamlined education programme on RD research to all interested stakeholders via an e-learning (brand new);
- Based on assessed needs of the RD community – in collaboration with universities – 10 to 12 modules with accreditation – e-learning format open to all – Future Learn platform

- **WP17: ERN RD training & support programmes**
- **Objectives**: research training programs for the ERNs & EJPRD Researchers focusing on cross-cutting and over-arching research themes Deliver (brand new)
- Based on four groups (Neuro, Neoplasm & malformation, Organs, Systemic) – preferences, needs and resources of ERNs – tailored for and performed by ERNs
  - **Research Workshop Topic**
  - **Research Mobility Fellowship → First year dedicated to ERNs**

- **WP18: Development and adaptation of training activities**
- **Objectives**: Ensure that activities within Pillar 3 address the developing education and training needs in RD research of key stakeholders across different EU countries (specific needs of EU 13 countries), according to progress of Pillars 2 & 4 and emerging needs of ERNs

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Pillar 4: Accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations

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Pillar 4: Activities

WP19: Facilitating partnerships and accelerating translation for higher patient impact

Innovation management toolbox – assessment and real time mentoring of translational projects – support in exploitation and follow-on funding – partnering support – roadmap for European investment platform for RD

- **Support of a dedicated Innovation Manager (IM) (to E-Rare & ERNs)**
  - **for translation of research activities: help to get sponsor, access network of experts**
  - **Contact the EJP RD Central Helpdesk**

https://www.ejprarediseases.org/index.php/ejp-rd-helpdesk/

WP20: Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs

Key Task Force group - Support in design and planning of RD clinical studies with ECRIN – demonstration projects on existing statistical methodologies to improve RD clinical trials – innovative methodologies to improve RD clinical trials in limited populations (validation of outcomes from ASTERIX, IDeAI, InSPiRe).

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Pillar4 - Demonstration projects call (WP20)

- Aims to Show the usability and capability of the innovative statistical methodologies for clinical trials in RD, which have not been demonstrated on existing data for specific rare disease clinical trials yet.
  - re-evaluate data that lacked efficiency because it was analysed with classical statistical methodology (asterix; IDeAl; InSPiRe)

- Projects may concern a group of rare diseases or a single rare disease
- Must include one of the identified methodological experts (the list provided afterwards)
- Generally, data should of interventional trial. For most methodologies: data from one or more randomized clinical trials with at least two parallel treatment groups.
- The proposal should include, among other information, a confirmation letter that the consent and/or authorisation for data re-use is granted by the private owner of the data, if applicable (i.e. availability of data)
  - limited to partners from institutions beneficiaries of the EJP RD and their third linked parties

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Clinical Trials Support Office

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Clinical Trials Support Office

- Established in collaboration with ECRIN , composed of several experts
- Permanent contact point for the and EJP RD partners (ERN investigators and other researchers) :
  - Help in the design and planification of multinational clinical studies.
  - Collecting feedback from ERNs to improve processes and better adapt to RD community
- Contact via the the EJPRD Central Helpdesk:

https://www.ejprarediseases.org/index.php/ejp-rd-helpdesk/

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EJP RD
– WHAT IS THERE FOR ME?

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Opportunities for ERNs

- **Can apply to calls for multinational projects and networking scheme**
  - 2nd Joint transnational call will open in December 2019
  - 1st Networking scheme call will open Q4 2019 *
  - Rare Diseases Challenges will be pre-launched by the end of 2019 (matching event Jan 14 2020) *
- **Collaboration/implication of ERNs in creation of the Virtual Platform of data and resources (Pilar 2)**
  - Integration/support for existing & new registries of ERNs
  - **FAIRification support**
  - Access to data/resources/tools
  - **Use cases definition to guide the Virtual platform development**
- **Eligible for dedicated trainings proposed by EJP RD**
  - The agenda of trainings is available at: http://www.ejprarediseases.org/index.php/training-and-empowerment/
  - Specific training dedicated to patients, including forthcoming training for young patients, are grouped under WP15 and led by EURORDIS
  - Specific training dedicated to ERNs (WP 17 – opening Q4 2019)
  - Free access to the EJP RD e-learning platform (first training modules available in 2020)
- **Contribution/participation in internal call for demonstration projects (for units that are part of ERNs) on validation and new innovative methodologies for clinical studies (WP20)**
- **Access to Support Office of ECRIN** for multinational clinical trials in RDs
- **Access to the EJP RD Central Helpdesk → access to expertise and RD dedicated services (already available or to be developed under the run of the EJP RD): http://www.ejprarediseases.org/index.php/ejp-rd-helpdesk/**

*** Calls open only to researchers form countries participating in the EJP RD**

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Joint Congress

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

RE(ACT)®
INTERNATIONAL CONGRESS ON RESEARCH
OF RARE AND ORPHAN DISEASES

- WHEN: **11 – 14 of March 2020**
- WHERE: **Berlin, Germany**
- PROGRAMME: **dedicated workshops, scientific sessions, policy, patients**
- **SPECIAL RATE UNTIL NOVEMBER 30, 2019**

https://www.react-congress.org

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