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External Newsletter n°4: September - October 2019

Welcome to the EJP RD NEWSLETTER

Every month, latest news of the biggest European rare diseases initiative, bringing together funders, researchers, patients and clinicians!

EDITO

Welcome back to our first fall edition of the EJP RD Newsletter. In this edition, we invite you to the RE(ACT) Congress - IRDiRC Conference and the next EJP RD workshop Hackathon on molecular pathways for rare disease (FAIR) data analysis, registrations are open. Also, we share with you the important Pillar 2 guidance document on interoperability considerations pertinent to the development and operation of ERN registries in collaboration with the EJP RD and some job openings at the EJP RD partner institutions. On the "Related news and opportunity" section, you will find the historical declaration of the UN where it is stated that persons with rare diseases deserve to be included in the Universal Health Coverage, the call for new European Reference Networks (ERNs) members as well as the registrations for the EURO-NMD 3rd Annual Meeting and for the Info and Brokerage event day organized by TUBITAK to facilitate international collaborations for the EJP RD JTC 2020 call. Finally, we reported short updates regarding the 1st EJP RD General Assembly meeting and those related to other events where EJP RD has participated triggering innovation on RD research, funding, and education across Europe.



EJP RD General Assembly

The EJP RD held its [first general assembly meeting](#) on 17 - 19 September at the

Medical University of Gdansk, Poland. More than 200 members and delegates coming from European countries, plus Israel, Turkey, and Canada, gathered together to discuss about the achievements reached in these nine months of activity, since the kick off meeting in January 2019. This 3-days meeting has been an extremely important opportunity for all EJP RD members to strength internal cooperation, define new cross-collaborations between the members and institutions, and consolidate activities and projects for the upcoming year. The EJPR RD tackles challenges in the rare disease field to trigger the impact patients expect. The consortium validated the working plan for the second year of the project and all EJP RD members confirmed their high commitment and motivation.

Within 9 month of its existence the EJP RD already demonstrates how the centralised collaboration between different stakeholders advances rare disease (RD) research for the benefit of patients. The bridges built between expanded RD research community and the European Reference Networks allow the advancement of RD Virtual Platform and common approach to standards, registries, data and FAIRification. Integration of patients in all activities and close collaboration with funders leads to the long-expected mindset changes on patient-centered research. The grounds put by actions dedicated to innovation in clinical trials, support for translation of research results and Rare Diseases Research Challenges partnership with industry, pave the way to EU competitiveness and faster delivery of therapies. Finally, the support received by the EJP RD from the members of its Policy Board and initiation of National Mirror Groups confirms the willpower to fully integrate RDs in national strategies ensuring seamless connection to EU and international strategy levels with special attention to CEE countries.



RE(ACT) Congress 2020 and IRDiRC Conference Berlin, Germany. 11 - 14 March 2020

EARLY REGISTRATION IS OPEN UNTIL 30 NOVEMBER
SUBMIT YOUR ABSTRACT!

The co-joint [RE\(ACT\) Congress 2020 and IRDiRC Conference, Berlin, Germany, 11th -14th March 2020](#) aims to bring together scientific leaders and experts and young scientists from a variety of breakthrough scientific fields to present cutting-edge research, exchange ideas and discuss policies related to rare diseases research. Patients and patient organizations, who are committed to research, will also be in attendance to share their experiences and perspectives.

EJP RD TRAINING COURSE

EJP RD Training Course. Workshop and Hackathon: Molecular pathways for rare disease (FAIR) data analysis

Link to the training on the EJP RD website
<http://www.ejprarediseases.org/index.php/workshop-and-hackathon-molecular-pathways-for-rare-disease-fair-data-analysis/>

26 - 29 November 2019



EUROPEAN JOINT PROGRAMME
RARE DISEASES

Maastricht University



[Registration](#) for the “Workshop and Hackathon: Molecular pathways for rare disease (FAIR) data analysis” in Maastricht on 26 – 29 November 2019, is still open.

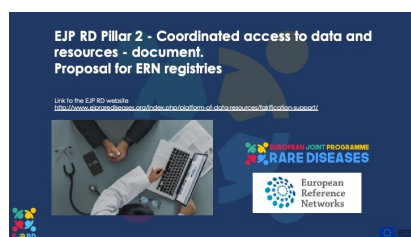
The workshop and registration are free of charge. Everyone interested is invited to attend, particularly EUTOxrisk and ERN researchers. The latter can claim cost reimbursement through the ERN Coordinator's institution provided their and the Coordinator's institution have a bilateral ERN Collaboration Agreement that makes them a Linked Third Party. For more information on these conditions please, get in touch with [secretariat-bigcat\(at\)maastrichtuniversity.nl](mailto:secretariat-bigcat(at)maastrichtuniversity.nl)

The topics will be focused on:

1. Pathway creation and curation – especially rare disease and adverse outcome pathways inspired by needs of ERNs and tox community (day 1-2)
2. FAIR data preparation (prep for pathway and network analysis) – define needs of data analysis and do first steps towards solving (day 2-3).

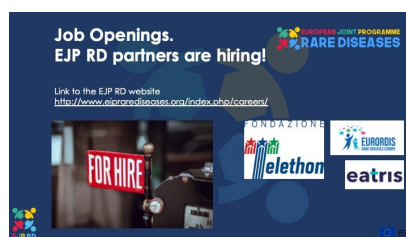
After the 3 days of workshop, a hackathon will take place in which the participants will start together solving the identified problems and test different approaches.

More EJP RD news



Coordinated access to FAIR data: Proposal for ERN registries

The EJP RD - Pillar 2 group provided an elaborated guidance document describing interoperability considerations on the development and operation of ERN registries. The aim is to offer organizational advice on how to make a registry more interoperable in collaboration with the EJP RD. The suggestions provided are equally relevant to groups setting up new registries or improving existing registries, and relate to all types of registries (primary resources, aggregation focused, clinical objectives, research objectives, etc.).



Careers

The following EJP RD partners are hiring:

1. **Telethon Italia** for a Technology Transfer Associate
2. **ECRIN** a Project Manager (deadline 31 October 2019)

[Read more](#)

Here we are!



Daria Julkowska
Assistant Director
Thematic Institute of Genetics, Genomics & Bioinformatics, INSERM

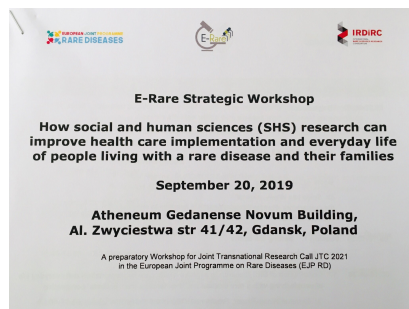
European Research & Innovation Days

The EJP RD Coordinator, Dr. Daria Julkowska, was invited to give a talk at this first annual policy event of the European Commission on how Horizon Europe, the successor to the current research Horizon 2020, should innovate and implement the future of rare disease research. Dr. Julkowska was part of the "Tackling Rare diseases" session together with Anne Paoletti (French Research Ministry, France), Andrzej Rys (European Commission, DG SANTE, Belgium), and Anna Lonnroth (European Commission, Belgium). Click [here](#) to watch the recording video and for more.



eHEALTH Forum

The EJP RD had the opportunity to develop and lead a specific session on rare diseases where Dr. Schreier, Dr. Mangir, and Dr. Chrzanowska presented relevant elements such as the importance of interoperability, FAIRness, Clinical Patient Management System, traceability of rare diseases patients in health information systems, and the role of ORPHAcodification. The EJP RD represented a valid voice of the European interest on advancing the rare disease research.



E-Rare Strategic Workshop

The EJP RD took part at the "*How social and human sciences (SHS) research can improve healthcare implementation and everyday life of people living with a rare disease and their families*" workshop held in Poland in September. Patients, scientists, healthcare providers, ethicists, and policy makers defined common research strategies to advance distinct social sciences aspects associated to rare diseases healthcare. The conclusions will be used to define the next related topics for the Joint Translational Call 2021.



The International Summer School on Rare Disease Registries and FAIRification of Data

The [workshop](#) held in Rome under the guidance of Prof. Domenica Taruscio and with the collaboration of Dr. Claudio Carta, gathered together patient representatives, researchers, healthcare professionals, and students coming from all over Europe who brainstormed around FAIR registry and on how establish it in a high-quality level.

Related News & Opportunities

Historical moment for the rare disease community worldwide! For the first time ever, in a UN official political declaration document was stated that persons with rare diseases deserve to be part of the universal health coverage (UHC). The EJP RD is very enthusiastic about this decisive turning point for the entire rare disease community and even more motivated to make a difference in the field working together hand by hand with patient organizations, the scientific community, and the other stakeholders.

[More information here](#)



Training needs on "Orphanet nomenclature and rare disease ontologies for RD research" (ORDO/HOOM)

Please answer this [EJP RD survey](#) about training needs for rare disease nomenclature and ontologies, if you are working with RD patients or data and involved in research activities, or if you supervise people in this case. Please answer it and forward it to your colleagues involved in rare disease research, it takes only **6 - 9 minutes**, **deadline is Friday 18 October**. Do not hesitate to forward the link to other professionals in your network who might be involved in rare disease research.



Call for new members to join the existing 24 European Reference Networks (ERNs)

The first call for new members to join existing 24 ERNs has been launched on 30 of September 2019 **until 30 of November 2019**.

HOW TO APPLY TO BECOME A MEMBER OF AN ERN? Click [here](#) for the details.



EJP RD on Rare Diseases Info Day and Brokerage Event. 7 November 2019. Istanbul, Turkey.

TUBITAK (The Scientific and Technological Research Council of Turkey) committed ~1,5 M Euro for the EJP RD 2020 Call to support the research partners from Turkey in the funded projects. In order to facilitate international collaborations for the EJP RD JTC2020 Call, together with Sabancı University, TUBITAK is organizing a Rare Diseases Research Info Day and International Networking Event on 7 November 2019, in Istanbul, at Sabancı University Nanotechnology Research and Application Center (SUNUM). Details on who can participate, travel grant, and registration links can be found [here](#).

More information [here](#).



EURO-NMD 3rd Annual Meeting. 6 - 8 November 2019. Ferrara, Italy.

The European Reference Network for Rare



Neuromuscular Diseases
(ERN EURO-NMD)

Neuromuscular Diseases Euro-NMD in collaboration with the University Hospital Cona in Ferrara, Italy and the Azienda Ospedaliero Universitaria di Ferrara organize the 3rd EURO-NMD Annual Meeting.
Registration is now open!
Click [here](#) for more info.

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EJP RD is coordinated by the National Institute for Health and Medical Research (INSERM)



CONTACT US:

EJP RD
Hôpital Pitié-Salpêtrière
4 rue des Petites Loges
75013 PARIS
[communication\(at\)ejpredirectediseases.org](mailto:communication(at)ejpredirectediseases.org)

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