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Newsletter n°3: July - August 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

Before the Summer break, the EJP RD coordination team would like to share with you some of our latest news from inside the EJP RD and from some of our "EJP RD friends". We are happy to provide you a broad spectrum of information on the rare diseases field spanning from more basic science to advocacy and policy.

Relevant EJP RD training courses on "Organizing & maximizing rare disease biological sample data in biobanks" and on "Quality assurance, variant interpretation, and data management in the NGS diagnosis era" are listed, together with some interesting patient-centered initiatives such as a newly released paper on patient preferences on data sharing and protection, an open forum, and new document on how patient engagement expectations are met. Moreover, a panel discussion on health biotech around Europe, a film festival on rare diseases, and a new drug for a RD available in EU...then a bit of "pure science" too with the first ever nanoscale video on the immune system in action! Also, the EJP RD serves as official partners for the 10th European Conference on Rare Diseases & Orphan Products (ECRD 2020)!

"Save the Date(s)" for the upcoming **Rare Disease Perspectives in Central - Eastern Europe workshop** and for the joint **RE(ACT) Congress 2020 and IRDiRC Conference!**

***Last but not least, the latest [interview](#) to our Coordinator, Daria Julkowska, on the EJP RD project has just been released!***

So, check out our EJP RD newsletter, website, and twitter for details, additional info and links!

Get involved and join our growing #EJPRDcommunity!



## Rare disease perspectives in Central - Eastern Europe

On September 16th 2019, the workshop "[Rare disease perspectives in Central - Eastern Europe](#)", twinned to the General Assembly of EJP RD, will be organized in Gdansk, Poland. The main aims of this conference are discuss challenges and opportunities of rare disease research in Central - Eastern Europe countries and areas of involvement within EJP RD for multiple stakeholders (including researchers, clinicians, authorities, and patient organizations). Presentations on rare disease policies, clinical research and innovation, data ecosystem, Orpha coding, biobanks, rare disease research education, European Reference Networks and other important topics will be given by the major EJP RD partners and representatives of local rare disease research communities.

The deadline for [registration](#) is September 1st, 2019!

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

The BLACKSWAN Foundation and IRDiRC, the International Rare Diseases Research Consortium, are glad to announce the joint [RE\(ACT\) Congress 2020 and IRDiRC Conference, Berlin, Germany, 11th -14th March 2020](#). This joint event will continue the RE(ACT) Congress series (6th edition) and IRDiRC Conference series (4th edition). It 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.

Registration will open soon!

### More EJP RD news:

#### Organizing & maximizing rare disease biological sample data in biobanks

This training course will take place in Vilnius, Lithuania, on October 29th - 30th 2019.

The participants to the first training course in Milan, Italy, highly appreciated the use of the problem-based learning methodology and enjoyed the courses.

**Registration will open soon!**

[Read more >](#)

#### Quality assurance, variant interpretation, and data management in the NGS diagnosis era

This training course will take place in Leuven, Belgium, on October 23rd - 25th 2019.

The main objectives of the training are decrease RD data fragmentation and increase data quality.

**Registrations are still open!**

[Read more >](#)

#### 10th European Conference on Rare Diseases & Orphan Products (ECDRD2020), Stockholm, Sweden, 15th - 16th May 2020

European Joint Programme on Rare Diseases serves as official partners for the 10th European Conference on Rare Diseases & Orphan Products (ECDRD 2020) taking place at the Stockholmsmässan Congress Center in Stockholm, Sweden on 15 - 16 May 2020. The overarching theme for the conference is: The rare disease patient journey in 2030.

[Read more >](#)

## Related News & Opportunities

### **INVEST IN THE FUTURE OF HEALTH BIOTECH, BRUSSELS, BELGIUM, 23RD SEPTEMBER 2019.**

As part of the European Biotech Week, EuropaBio will host a panel discussion on the 23rd of September in Brussels. Experts from the biotech financing ecosystem in Europe will discuss the future for access to finance in the EU and present different approaches to keep up in the race with biotech front-runners such as China and the US.

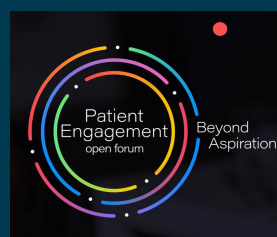
[More information here](#)



### **PATIENT ENGAGEMENT OPEN FORUM, BRUSSELS, BELGIUM, 18th - 19th SEPTEMBER 2019.**

PARADIGM, PFMD and EUPATI will organise the Patient Engagement Open Forum, which aims to provide a holistic perspective of patient engagement, the landscape and actors, and foster collaboration and co-creation while breaking down fragmentation and silos that are often present in patient engagement work. The agenda of the two days offer a deep dive into some ongoing patient engagement work done by many collaborative initiatives.

[More information here](#)



### **NEW RESULTS OF #RareBarometer SURVEY ON PATIENT'S PREFERENCES ON RARE DISEASE DATA SHARING & PROTECTION.**

A new EURORDIS-led article has just been published in the Orphanet Journal of Rare Diseases.

The peer-reviewed article is freely accessed. The article presents a series of **recommendations** to inform and support stakeholders (policy makers, researchers, funders and patient organisations) in shaping relevant, ethical and responsible data-sharing initiatives that would respect preferences from people living with a rare disease.

[More information here](#)



### **EFPIA, EATRIS, ELIXIR, BBMRI, ECRIN STATEMENT ON THE ROLE OF RESEARCH INFRASTRUCTURES TO BOOST PATIENT-CENTRED RESEARCH AND INNOVATION IN EUROPE.**

The European Union has invested heavily in health research. Since the early 2000's it supported the creation of European Research Infrastructure like BBMRI, EATRIS, ECRIN, ELIXIR, and funded the Innovative Medicines Initiative (IMI), the world's largest public private partnership (PPP) in health. The combination of PPPs such as the IMI and European infrastructures has contributed significantly to the boosting of Europe's translational research power – turning science in to new treatments. The next step for Europe is to



bring these two success stories in health research closer and design a new path together, to enable European researchers to make more and more new treatments available to patients.

[More information here](#)

#### **EPTRI EU STAKEHOLDERS ROUNDTABLE SUMMARY REPORT RELEASED!**

Representatives from paediatric networks, research infrastructures (RIs), European Reference Networks (ERNs), charities, patients' associations, Young Persons Advisory Groups (YPAGs), companies' federations, Governments and regulatory Authorities had a very productive discussion during the stakeholders roundtable held in the framework of the 3rd General Assembly of EPTRI (European Paediatric Translational Research Infrastructure) in Madrid, Spain, on March 27th 2019.

[More information here](#)



#### **THE CALL FOR NOMINATIONS FOR THE EURORDIS BLACK PEARL AWARDS 2020 IS STILL OPEN.**

The Awards are presented to patient advocates, organisations, policy makers, scientists, companies and media for their major achievements and outstanding commitment to the rare disease cause.

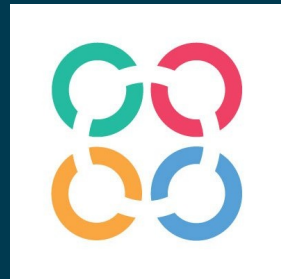
[More information about the award and how to nominate your candidate here](#)



#### **D1.2 CRITERIA TO ASSES HOW PATIENT ENGAGEMENT EXPECTATIONS ARE MET**

This report describes the work that was done as part of PARADIGM's work package 1, which aimed to identify and define stakeholders' preferences, needs and expectations in patient engagement through an online survey and focus group consultations that were conducted in 2018. The objectives of this work were two-fold: firstly to prioritise those needs and expectations identified, and secondly, with the help of topic experts, turn those prioritised needs into a set of criteria for a good patient engagement activity.

[More information here.](#)



#### **UNO SGUARDO RARO - A RARE GLANCE -.**

The rare disease international film festival is the first and only film festival on rare diseases in Europe. It collects and promotes the best video works on rare diseases and social inclusion through an international competition and works with public and private partners, both national and international, the rare diseases community, disability advocates, entertainment and film culture professionals. It is a cultural project created by Claudia Crisafio, actress and author, and Serena Bartezzati, a rare patient and professional in the field of communication.

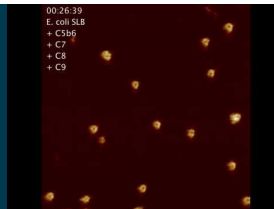
[More information here.](#)



## FIRST EVER NANOSCALE VIDEO ON THE IMMUNE SYSTEM IN ACTION.

New Frontier Research - Health article by Horizon Magazine on a video at nanometre resolution -first time ever-, where scientists filmed how the immune system kills bacteria by poking holes into it. Professor Georg Fantner tells "We want to watch what is going on at the nanoscale inside living cells, which has never been done. You can't use a normal optical microscope to do this because photons (or the wavelength of light) are too big: in essence, we are blind. So instead of trying to see something with light, we try to feel the surface - like a blind person using a stick".

[More information here.](#)



## NEW MEDICINE FOR A RARE DISEASE AVAILABLE IN EUROPE.

EMA's human medicines committee (CHMP) recommends authorisation of new treatment for phenylketonuria, a rare inherited metabolic disease.

[More information here](#)



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



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