

FROM THE COORDINATOR

Dear member, dear colleague,

In the third year of its existence, the ERN structure is expanding. We expect members from all EU member states. Not only new members will be welcomed to our RITA Network, also tasks put on us by the European Commission become clearer.

ERNs must prove to Europe, national health care authorities, pharma, health care insurance companies and the learned scientific societies that they are a vital structure for care for the rare across Europe. The European Commission requires clear output data indicative of our activities. This is of course challenging since much of our activities are “voluntary work”. But our existing societies supporting ERNs are strong and support RITA.

In this month we will learn how many centers have applied to the European Commission to become a new RITA member. We expect well over 50 new members. With this expansion we need to revise our governance structure with housekeeping rules. Our ERN contains 4 major disease streams. We will propose such a structure soon, and will discuss it with our current RITA board and our societies. As we discussed in the beginning of 2017 direct input from the councils of these societies is vital. We need to formalize this interaction. We also need a strong RITA network for future collaborative grant applications and discussions with the health care authorities.

I am happy to see there are vital patient/parent organizations for these 4 main areas. As they do in our scientific organizations such as PRES and ESID, patients should have more impact, for instance on relevant outcome measures meaningful for patients, setting priorities in research agendas, reviewing expertise centers, writing patient journeys, etcetera. In my own field, pediatric rheumatology, this has led to much stronger and vital interaction between health care providers of PRES and patients in ENCA. Also in the other streams I see vital organisations such as IPOPI. Let's get to work together.

The 17th and 18th of January 2020 we are organizing the annual general RITA meeting in Utrecht, the Netherlands. Details can be found on our website soon. We hope to welcome many of you there!

Nico Wulffraat,
Coordinator of RITA

■ RITA attended the ERN Coordinators meeting in Brussels

The ERN coordinators, project managers and European Commission met in Brussels on the 14th of November. The next day the ERN coordinators had an internal meeting and parallel the Board of the Member States met. High on the agenda was the call for new members launched at 30 September 2019 and closed on 30 November 2019. A large number of new HCPs will submit their application and will result in a large expansion of all ERNs across Europe.

Expansion of the RITA network

Also the RITA network expects over 50 applications for its network. After an extensive application and assessment procedure the RITA network will grow considerably.

In addition to new full members, the RITA network already welcomed 14 affiliated partners from Estonia, Latvia, Lithuania, Austria, Croatia, Denmark, Hungary, Malta and Slovakia.

■ RITA annual event 17-18 January 2020

ERN RITA will organise a two day event for all members on 17 and 18 January 2019 in Utrecht. ERN RITA is developing and growing and this event is aimed at strengthening the network, engaging more network members, enhancing collaboration within and among the 11 Working Groups, the 4 Disease Streams, HCPs, professional societies and patient groups.

■ Molecular testing WG: Second 2-day workshop on Human Phenotype Ontology (HPO)



The 17-18th of October ERN RITA molecular testing WG in collaboration with the ESID WP of Genetics organised the second 2-day workshop on Human Phenotype Ontology (HPO) for immune mediated diseases. The workshop was hosted by Kaan Boztug, Julia Pazmandi and Matthias Haimel, from the Ludwig Boltzmann Institute for Rare and Undiagnosed diseases / St. Anna Children's Cancer Research Institute in Vienna.

HPO provides a standardized vocabulary to describe phenotypic (clinical) abnormalities observed in human diseases. However to date, rare diseases of the immune system lack a complete and accurate description using standardised clinical terms. An accurate vocabulary of phenotypic description helps to separate similar disease subtypes and provide

patients with the correct treatment. The initiative started with the first HPO workshop in Vienna last year and aims to fill this gap in order to provide a revised and complete phenotype ontology for immunodeficiencies.

Over the two days, more than 20 experts from the fields of bioinformatics, medicine, immunology, and genetics worked together to further expand the description for different immune mediated disorders in HPO.

There are currently 4 groups working on HPO terms addressing the IUIS based subgroups of Immunodeficiencies affecting cellular and humoral immunity; autoinflammatory diseases; diseases of immune dysregulation and predominantly antibody deficiency. If you would like to join a group or start working on a different IUIS subgroup, [please let us know](#).

■ Transition Care WG: a survey performed across Europe

A survey across Europe was performed to have a global vision of the current situation of transition care in PID and autoinflammatory diseases across Europe, to detect differences between countries and centres and to identify potential global needs that could be addressed from the ERN.

44 centres responded. Preliminary data are available and could be partially shared. The working Group has planned for a second distribution to increase coverage across European countries.

As a result of this work, the working group aims at releasing a manuscript and at creating European guidelines.

FROM RITA STREAMS

■ PID - IPOPI IPIC*: keynote presentation from Dr Enrique Terol, Senior Policy Officer at European Commission, in charge for ERNs



ERN-RITA actively participated in the 4th IPIC with a booth where congress participants were able to get informed about the activities of the network.



Dr Enrique Terol,

*From left to right:
Markéta Filipíková,
Zdeněk Doležal, Martine
Pergent, IPOPI, Dr
Enrique Terol,
Johan Prévot (IPOPI),
Leire Solis (IPOPI)*

Dr Enrique Terol, Senior Policy Officer at European Commission and in charge of European Reference Networks provided a keynote presentation in the opening session of the 4th International Primary Immunodeficiencies Congress (IPIC2019). IPIC2019 was organised by IPOPI and held in Madrid, Spain on 6-8 November 2019. The congress brought together 750 clinical and scientific experts in the field of primary immunodeficiencies alongside patient organisation leaders, expert nurses, industry representatives and other healthcare professionals. Among challenges/future goals for the ERNs, Dr Terol highlighted the integration of ERNs into national healthcare systems, effective support at hospital level, long term organisational and financial sustainability, enlargement of the diseases and geographical coverage, the development of a research activities ERN framework as well as the monitoring and

evaluation process of ERNs to demonstrate their added value. In exploring the ERNs long term sustainability issue, Dr Terol presented the next budget scenario indicating there will be a multiannual financial framework with a new approach to health as an investment (European social fund plus and link to other financial mechanisms: European Semester, InvestEU fund) and the new Research & Innovation funding programme (Horizon Europe). Specific sectorial budgets will also be available.

*International Primary Immunodeficiencies Congress

>>Learn more about IPIC 2019: <https://ipopi.org/ipic2019-welcomed-750-participants-in-madrid-spain/>

■ PID - New Webinar at disposal: “IG Therapies: From good to best practice”



IPOPI held 3rd Clinical Care Webinar on 26 September, on Immunoglobulin therapies: From good to best practice. The invited speaker was Professor Troy Torgerson from University of Washington and Seattle Children’s Hospital, US. The webinar was clinically oriented but suitable for a non-medical audience as well.

In the 30 min webinar, Prof Torgerson explained how to aligning IgG treatment approach with patient needs. Thus, he showed several examples of patients and their treatment choices based on medical outcome and personal choice. He demonstrated how personalized care is very important for primary immunodeficiencies (PID) patients in need of immunoglobulin therapies and that a large range of immunoglobulin therapies are necessary to ensure that PID patients receive the best possible care.

>see the recording of the webinar on IPOPI TV: <https://tv.ipopi.org/>

■ PID – IPOPI PID Forum launched a newborn screening for rare diseases Call to action



IPOPI’s 13th EU PID Forum dedicated to “Newborn screening for rare diseases. A PID perspective” took place on December 4, 2019 at the European Parliament, Brussels.

4 Members of the European Parliament (MEPs) co-chaired the forum, namely, Dr Manuel Pizarro (Social-Democrats, Portugal), Ms Tilly Metz (Greens, Luxembourg), Ms Irena Joveva (Renew Europe, Slovenia) and Ms Sirpa Pietikainen (European People’s

Party, Finland). Dr Tudor Ciuhodaru MEP (Social-Democrats, Romania) actively participated in the meeting.

The meeting set up the scene for a strong political debate on newborn screening for severe combined immunodeficiencies as well as other severe forms of PIDs (such as complete Di George syndrome) and launched a **Call to action**, highly supported by the MEPs present and by the Forum participants. The need for the development and implementation of overarching guidelines in the field of newborn screening for rare diseases and the creation of a European newborn screening standing committee was observed. This would facilitate the exchange of best practices and recommendations on newborn screening and allow national decision-makers to better access information and solid evidence from other Member States.

Newborn screening for rare diseases, using SCID as an example, has been the topic of an EU-wide campaign IPOPI is currently working on in close collaboration with the International Society for Neonatal Screening (ISNS) and the European Society for Primary Immunodeficiencies (ESID). IPOPI is grateful to

ISNS representative, Dr Peter Schielen, and to ESID President, Prof Isabelle Meyts, for their presence and contributions to the Forum.

■ **AUTO-IMMUNITY - European Vasculitis Society (EUVAS) Cambridge 2019 course**

The EUVAS Cambridge 2019 Vasculitis course took place between 23rd and 25th Sep. As vasculitis is a key disease group in the RITA autoimmune strand, Mark Little attended and gave a lecture on vasculitis pathogenesis. The goal of the course, which was over-subscribed threefold and had attendees from all over Europe, was to provide an in depth understanding of the aetiology, epidemiology, pathology and clinical management of vasculitis affecting adults and children, with exploration of the organ specific manifestations. This highlights the multi-disciplinary aspects of the disease; trainees were empowered to consider ways in which to plan vasculitis services in their countries. There was a focus on the ability to dissect and analyse complex clinical presentations and management dilemmas and to promote the ability to offer a tertiary sub-specialist service in vasculitis. Building of collaborator physician networks between medical specialties and geographical areas was emphasised, using ERN-RITA as a mechanism to achieve this.



■ **AUTO-INFLAMMATORY - Annual Behçet's study day**

Doctors, dentists, trainees and students annual Behçet's study day aimed at giving information and taking feedback. Moreover it addressed diagnosis of Behçet, since new criteria have been introduced.

■ **AUTO-INFLAMMATORY - Annual Patient - listening Day**

September had an enjoyable and informational patients' day aimed at updating patients on disease specific management and receiving patient feedback on improving service. The meeting was also a great opportunity to see patient input from last year meeting (patient video for children). Patients are discussing further information videos. This has been a very successful meeting.

AGENDA

January 2020

Neuroinflammation meeting in January 2020. More info once the program is finalized. Info contact: Kumaran Deiva

May2020

Paediatric Infectious diseases - ESPID 2020 to be held in Rotterdam 25-30 may 2020.

[>>Learn more about](#)

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