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Europa Gen Ass  
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## IN THIS ISSUE:

- VHL 2017 General Assembly
- EURORDIS Membership Meeting
- ERN
- Propranolol
- News and Agendas from our members.

## VHL Europa 2017 General Assembly

On May 18th in Budapest.

Most of countries were present (Germany, Netherlands, France, Italy, Hungary) or participated via Skype (Spain, Greece, Denmark).

The president of the Board presented the important activities done or in progress:

- **Young Adults VHL Symposium** in Berlin on next June; 16/17 participants from all Europe have registered for this event; they will have a 2-3 days meeting with lectures and workshops and also time for relaxing and getting to know each other. This will be our second experience after Utrecht 2015 and we believe very much in it; a similar event will be held soon also in USA;

- **Turkey** is now in contact with us; they have a new Facebook group and the VHL handbook (German version) is translated and available in Turkish.

- **A VHL Symposium in Greece** will be held next November, organized by the Greek national alliance. Most of the speakers will come from Europe and even USA, to raise awareness among doctors and families

- **Handbook for young children** (a new project from France) the draft of the book is finished and France is waiting for a final authorization; the idea is to have a book that children could read with their parents' help at first, with easy

language, pictures, etc.; an important team of doctors worked on it . After the meeting, the handbook has been sent to all VHL Europa members. The Netherlands intends to translate it into English. In the next coming months, we will discuss the possibility to translate it in different languages.

- **List of doctors for second opinion** is still in progress; the goal is to share this list for all

- a draft of **Policy on Funding Research** at European level was presented by the Board and approved from the Assembly. Now we have the rules to manage research projects funding at European level.

- **Education of doctors** is a topic we want to finalize. We cannot financially support meetings around Europe, in addition to the local ones, and so the idea is to study how to organize webinars for physicians.

- **Propranolol**, Spain shared its project to launch a clinical trial.

The General Assembly accepted a new member: **Romania**

## EURORDIS MEMBERSHIP MEETING 2017 (a little summary)

270 persons had registered in order to attend.  
Among them: 23 were EURORDIS staff.  
EURORDIS members were 187 in 2000, 743 in 2017.  
European Federations were 20 in 2000, 58 in 2017.

### Friday 19, General Assembly

New policy/strategy for next 3 years in the EU is under discussion. We do worry that Health is not any longer in high priorities, in spite the fact that surveys show that European people place Health, Jobs and Education high in priorities. Even if we are afraid of the storm that is shaking the EU and accordingly the decline of interest and opportunities for RD, we must trust ourselves: we have already achieved a lot and we shall have as many new opportunities as well as new challenges, as those we had in the past 20 years.

### Saturday 20, Workshop n°2: "Hope and happiness as part of the treatment."

SIDE EFFECTS OF MEDICINES. Consult the appropriate authority from the list of national medicines regulatory authorities in the EEA for information on how to report a side effect.

EURORDIS is actually trying to draw attention of the EU Commission to NATC (Natural Alternative Traditional Therapies) in order to include some of them in clinic trials.

We do not have any information on risks and benefits of NATC on RD.

For instance, in VHL, symptoms are very different as well as affected organs and there are many different mutations of the gene. Which one of the NATC might be efficient to treat the different mutations or to treat the different organs symptoms? Investigating would be complex.

How can we transform all patients' experience of NATC into clinical trials: properly record it and utilize all this knowledge?

Many health professional are reluctant: by education, they prefer to rely on scientific evidences, some are more open, a few are crooks. Nevertheless, many NATC being practiced by doctors, we should preferably consult these one.

Using NATC products should be monitored and supervised by a health professional in case of interaction with classic medicines and taking into account the risk of serious side effects.

Closely check the manufacturers claims try to find scientific articles/information, ask your doctor.

Be careful with the "mouth to ear". What works with one, might not fit with you.

## Join us!

Whether you are a VHL expert, doctor, national VHL organization, patient or other interested person or party, we need your support!

We have 3 types of memberships:

- 1) Full members, being national VHL organizations recognized in their country as non profit associations within Europe, independent of authorities, political parties, the pharmaceutical industry and commercial organizations
- 2) Affiliated members, are individuals from European countries where no National Association for VHL is in existence. An affiliated member may attend all VHL-Europa meetings as observer without the right to vote.
- 3) Sponsoring members, members can be individuals who support the goals of VHL Europa. They may attend all VHL-Europa meetings as observer without the right to vote.

### Promote us!

### Saturday 20, Workshop n°4: "Social Revolution"

#### BALANCING CARE AND DAILY LIFE

It focused on the preliminary results\* of the INNOVCare/Rare Barometer Survey on the impact of rare diseases in daily life.

\*Detailed results and press release are now available on EURORDIS website <http://www.eurordis.org/news/3000-rare-disease-patients-carers-voice-difficulties-balancing-care-life>

Possible solutions were presented to the plenum in the plenary session:

1. Coordinator of care, linking health, social, mental issues
2. Case manager, someone takes it all
3. Career coach for young adults & children

An interesting and inspiring workshop.

It would be interesting to learn how the results would be just for VHL affected! If many of us would have joined the study, results were already available...

**You can find all details and presentations on the EURORDIS website**

## ERN

### **What does ERN mean?**

An acronym for European Reference Network (for Rare Diseases).

The goal of ERN is the improvement in the overall quality and management of care of a single rare disease (RD) or a group of RDs with similar health care needs by complementing, supporting and providing added-value to the existing services and expertise at the national level. ERN provides a clear governance structure for knowledge sharing and care coordination across the Europe. Patient organisations are not legally required to participate in the governance and evaluation of ERNs, but ePAGS (European Patients Advocacy Groups) are under construction (see at the end of this article).

### **A little bit of history**

In July 2004, an EU Commission High-Level Group on Health Services and Medical Care was established to bring together experts from all Member States to work on practical aspects of collaboration between national health's systems in the EU.

Since 2004, EUCERD (the EU Committee of Experts on Rare Diseases) is mandated to assist the European Commission in formulating and implementing the Community's activities in the field of Rare Diseases, to foster exchanges of relevant experience, policies and practices between the Member States and stakeholders.

2006 December, the committee of experts on RD report outlines... the importance of identifying centres of expertise and the roles that such centres should fulfil.

2009 June, 08 EUCERD issues a recommendation to EC: "EUCERD Recommendations on European Reference Networks for Rare Diseases". The recommendations were elaborated to feed into the work of the Cross-Border Healthcare Expert Group aiding the European Commission implement the Cross-Border Healthcare Directive, adopted on 28 February 2011.

From 2014, "EUCERD Joint Action" replaces EUCERD and supports the activities of the European Commission Expert Group on Rare Diseases.

- Enhancing the visibility and recognition of RD
- Contributing to the development and dissemination of knowledge on RD, from specialised research, to the support of the healthcare professionals and the empowerment of patients;
- Contributing to improvements in access to quality services and care, from diagnosis, through to care and social support and innovative therapies

2015 June, EUCERD makes public an addendum to the previous recommendation, insisting on the grouping of RD into thematic networks and the necessity of a patient-centred approach to RDs.

2015-2016: reducing to 20-25 groups of Rare Disease (from 6,000) in order to simplify complexity (see below). The EC will cannot, even with an increased budget, support a

big number of ERNs for RDs. So, ERNs must gather a critical mass of patients and data in order to support research and improve research capacities. The proposed structuring into a limited number of ERNs (25) will allow this critical mass to exist and will facilitate interoperability of health information systems across the EU. The system of ERNs which will be created should aim at reducing drastically the number of undiagnosed patients. Each ERN will cover a broad range of diseases, gathered by therapeutic areas rather than by distinct disease or limited group of diseases, creating coherent clusters of RDs; e.g. there would be an ERN on "rare pulmonary diseases" instead of one ERN on cystic fibrosis, etc.

Here is the updated list (2016 Nov) of the 25 ERN:

ERN	Number of health care providers (HCP)
Rare immunodeficiency and auto-inflammatory and autoimmune diseases	24
Rare bone diseases	38
Rare adult cancers	67
Rare paediatric cancer	54
Rare cardiac diseases	24
Rare connective tissue diseases	26
Rare complex epilepsy	28
Rare malformations and developmental anomalies and rare intellectual disabilities	23
Rare endocrine diseases	71
Genetic tumours	23
Rare eye diseases	30
Rare hepatic diseases	28
Rare gastrointestinal diseases	20
Rare gynaecological and obstetric diseases	0
Rare haematological diseases	65
Rare craniofacial anomalies and ENT (ear, nose and throat) disorders	28
Rare hereditary metabolic disorders	69
Rare multi-systemic vascular diseases	31
Rare neurological diseases	61
Rare neuromuscular diseases	61
Rare malformation & development anomalies	37
Rare pulmonary diseases	61
Rare renal diseases	38
Rare skin disorders	56
Rare urogenital diseases	29
Transplantation in children	20

**At the moment, while editing, we do not know in what ERN VHL is going to be.**

Each Thematic RD-ERN should ensure continuity of care for patients by bridging the transition from paediatric through to adult care; therefore, it is highly recommended that a single ERN is established in each domain to meet the needs of both paediatric and adult patients, wherever possible.

EURORDIS suggests to approach all RDs as a whole, and to recommend that each Network to be supported with significant funding.

#### Agenda

16 November 2016	Independent Assessment Body completes the technical assessment on ERN applications
15 December 2016	Board of Members States approval of success of ERN applications
January 2017	EC announces the list of successful ERN applications

8-9 March 2017	3 <sup>rd</sup> ERC Conference in Lithuania: ERNs launched
March 2017	Members State endorsement of affiliated partners, collaborative centers and national centers as part of ERNs
2017 onward	ERNs open application for new HCP providers

In parallel, EURORDIS is developing a EURORDIS **Patient Advocacy Group (EPAG)** for each ERN disease grouping. These ePAGs will bring together elected patient representatives from EURORDIS member organisations and will ensure that the patient voice is heard throughout the ERN development process. This structured approach towards patient representation in ERNs will enable EURORDIS to support patient engagement, mutual support between representatives and cohesiveness.

## Propranolol - First drug to get orphan drug designation

During its meeting in mid January the Committee for Orphan Medicinal Products (COMP) has given Propranolol orphan drug designation.

The Committee agreed that the condition, von Hippel-Lindau (VHL) disease, is a distinct medical entity and meets the criteria for orphan designation. The intention to treat the condition with the medicinal product containing propranolol hydrochloride was considered justified based on preliminary clinical data showing a stabilisation of retino-haemangioblastomas in patients with VHL and who were refractory to laser therapy.

The sponsor - Consejo Superior de Investigaciones Cientificas (CSIC) - has also established that there exists no satisfactory method of treatment that has been authorised in the European Union for patients affected by VHL. A positive opinion for propranolol hydrochloride, for treatment of von Hippel-Lindau disease, was adopted by consensus.

## News and agendas from or members

### Agendas

- 30 September: Celebration of 20th anniversary of the Dutch VHL Organization
- 07 October: Denmark, General Assembly
- 14 October: Italy, VHL Annual Meeting in Rome
- 13-15 October: Germany, VHL Meeting in Leipzig
- 04 November: Greece, VHL Medical Symposium

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