EFAPH AND RARE DISEASE DAY
FEBRUARY 29th, 2015

The 2015 Rare Disease Day was celebrated by scores of merry and colourful demonstrations worldwide. Quite naturally EFAPH took an active part through its 12 European member countries. We cannot go about much detail for each of them but here are 2 representative examples, one in Germany and one in France.

See www.eurordis.org/fr/content/rare-disease-day-2016-theme-and-slogan

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SILVER AWARD 2015!
A radiant HH patient...

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Calendar 2016

March 2016: Launch of ERN on Rare Haematological Diseases (with EHA)
April 7 – 9 2016: Innsbruck (Austria)
European Iron Club Meeting
April 10 2016: Innsbruck AGM
(EFAPH + HI)
May 26 – 28 2016: Edinburgh (Scotland) EUCERD Meeting
June 2016: European HH week
June 2016: Patients Empowerment Campaign (EPF) European Parliament, Brussels

Otorgación de premio de plata 2015!
Una paciente HH radiante...

Margaret Mullett, presidenta de IHA (Asociación Irlandesa de Hémochromatosis), ganó el premio Silver Surfer 2015. Desde hace 20 años, bajo el liderazgo de Margaret, IHA ha establecido un equivalente de una red multidimensional utilizando la última tecnología para promover la conciencia sobre la hemochromatosis hereditaria. Más de 20,000 personas han sido diagnosticadas en este país, que tiene la prevalencia más alta en la UE. ¡Felicidades Margaret!

Calendario 2016

Marzo 2016: Lanzamiento del ERN sobre enfermedades hematológicas raras (con EHA)
7-9 de abril 2016: Innsbruck (Austria)
Conferencia del Club de Hierro Europeo
10 de abril 2016: AGM de Innsbruck
(EFAPH + HI)
26-28 de mayo 2016: Edimburgo (Escocia) Reunión EUCERD
Junio 2016: Semana de HH Europea
Junio 2016: Campaña Empoderamiento de Pacientes (EPF) Parlamento Europeo, Bruselas
As an introduction to EFAPH and Haemochromatosis International (HI, formerly IAHA) AGMs, a scientific Conference, open to the public, was dedicated to “Reflections on clinical and practical aspects of HH”. International speakers, invited by Dr. Barbara Butzeck attended. Patients groups and experts worldwide are working together to reduce the damage done by haemochromatosis.

The European General Meeting (EFAPH’S AGM)

- The statutory activity and financial reports as well as some changes in the Statutes were unanimously approved.
- The Board was renewed (11 members of whom 2 newcomers: Maria Abele, Chair of the Hungary Association and Annick Van Clooster, Treasurer of HVV, Belgium) and the Executive Committee was appointed.
- The projects and activities were reviewed, particularly the opportunity to join one of the European Reference Networks (ERNs).

The international annual meeting (HI’s AGM)

Haemochromatosis International (HI) is a worldwide alliance aiming at federating HH patients’ organisations in 5 continents: North America, South America, Africa, Australia and Europe. Founded in Canada (Bioiron 2011, Vancouver), structured in London Bioiron 2013 (Rob Evans President - UK), HI is now fully operational (Ben Marris President - Australia). HI and EFAPH joined forces to have both their AGMs in Cologne, taking advantage of the excellent logistics provided by HVD. It has been the opportunity for intense exchanges between the 2 groups and some EFAPH representatives have been elected as HI trustees: Barbara Butzeck (Germany), Emerencia Teixeira (Portugal), Howard Don (UK), Rob Evans (UK), together with Paulo Santos (Brazil) and Desley White (UK). Besides Ben Marris as President, Rob Evans is past-President, Paulo Santos is Vice-President elect and Desley White is Secretary Treasurer.

HI is very active and will review its progress at the Innsbruck meeting.

The scientific session

Sixty attendees (patients, experts, nurses) from 15 countries discussed HH patient’s problems and learned something about progress in research. Working groups were set up on several topics. Significantly two large topics stood out orienting the short-term activities:

- **The blood-donation future** has been largely discussed and still is, under EFAPH impulse. Pr. Graça Porto, head of EFAPH’s Scientific Committee, insisted on the variety of national/regional practices and on the necessity to promote harmonization. The survey on blood-donation practices in Europe, conducted by the Portuguese team, has to be concluded (1,800 HH patients from 12 countries were included) and lobbying from governmental authorities, like in Spain (Cataluña), has to be strongly pursued. Blood safety is also a major concern for scientists as HH red blood cells might present morphology abnormalities. A working group has been set up to carry out some scientific studies on the matter. The french Blood Establishment partially supports these actions.

- **Arthopathy**, is known to play a major role in the life quality of HH patients. A working group comprising international experts in rheumatology and HH-patients was set up under the leadership of Dr. Barbara Butzeck. Noteworthy are Pr. Pascal Guggenbuhl (France) - working on the efficiency of Anakinra in reducing pains (THERA project) -, Dr. Patrick Kiely (UK) and Dr. Stephanie Finzel.

EFAP has joined Eular since the Cologne meeting.
NEW! EUROPEAN REFERENCE NETWORKS (ERNs) ON THE MOVE!

Further to the Directive from the European Commission on patients’ rights in cross-border healthcare (March 2011), EURORDIS is managing a European project with a view to establishing Rare Diseases (RD) European Reference Networks (ERNs). Two international Conferences (Brussels 2014 and Lisbon 2015) have resulted in defining the frame and objectives of the ERNs. Further, EURORDIS promoted 21 “Groups” to avoid the dispersion due to the very numerous Rare Diseases. EFAPH volunteered and was co-opted by the Rare Haematological diseases set around a wider patients’ advocacy group in the domain of hematological disorders and supported by the European Haematological Association (EHA). The implementation will be based on 5 stages:

- Call for interest from the European Commission for ERNs (March 2016)
- Submission to the European Commission
- Application reviewed by the European Commission and the Independent assessment Body (IAB)
- Assessments and reports by the IAB, then review by the European Commission
- Final approval by the Board of Member States

Due to EFAPH small size, the integration into the group supported by EHA provides the adequate environment for HH to be covered. EFAPH is represented through its Scientific Committee (Pr. Graça Porto, Pr. Domenico Girelli and Pr. Pierre Brissot). Some national precedents exist within EFAPH and the endorsement of national authorities, including patients organizations and caretakers. This will be done by using a model letter proposed by the group formed around EHA.

As far as EFAPH is concerned, action will be taken to activate its 12-14 national Associations. The whole project will be an intense and quite complex process.

HH AWARENESS

The international rollout of the project to obtain a diagnosis of HH as early as possible is still on. Here are 2 interesting approaches.

PORTUGAL: HH IN TEXTBOOKS!

- 14-15 year-olds will study hereditary haemochromatosis (HH) as an example of genetic diseases over the next few years. A chapter titled “How to study the transmission of hereditary diseases” has been inserted in a science textbook on the Portuguese Haemochromatosis Association initiative. It features some basic information about HH and also some practical questions about the disease.
- HH has also been presented in a teachers’ course in Portugal major cities; none of the 600 attendees met with any difficulties using HH as an example of autosomal recessive diseases. This is an effective and low cost HH awareness strategy, one of EFAPH’s major objectives.

FRANCE: INTERREGIONAL ACTION

- Following the Yvelines Pilot project, which targeted 900 GPs (2013/14), a similar campaign was successfully implemented in French Brittany (3,000 GPs, 2014/2015) and is currently underway in the Paris Region. In all, 14,000 GPs will be involved before the end of 2016. Intense lobbying from French and European Patients’ Federations targeting the Paris Regional Health Agency will gradually lead to the setting up of a genuine HH network of Competence Centres and phlebotomy venues (hospitals, French Blood Centres, independent nurses...). This is essential to the implementation of the GPs awareness campaign in this region.
A meeting was held in Paris October 2, 2015 between EFAPH representatives and those of La Jolla Pharmaceutical Company (San Diego, California). This company produces synthetic hepcidin and showed great interest in the actions developed by EFAPH.

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A charity concert was organised in Paris on 23rd November 2015 to raise funds for EFAPH, with the internationally renowned pianist Yves Henry and the young violinist virtuoso Hildegarde Fesneau. After a masterclass around a Mazurka of F. Chopin, the 2 concert performers interpreted a Sonata of W.A.Mozart followed by a “whim” of P. de Sarasate. The concert was splendidly ended by several romantic opuses of F. Chopin interpreted by Yves Henry whose talent, culture and kindness made a conquest of the public.

An exceptional evening!

In 2015, Maria Abelé replaced Pr.Judith Varkonyi, who remains honorary President of the Association. Maria and her team are very active particularly this year in various events to raise the awareness of General Practitioners and of the public at large.

AEH invited Pr. Pierre Brissot and Dr. Barbara Butzeck to this special event.

Norway, Iceland, Denmark, Sweden and Finland are now sharing the same Facebook page.

Nice initiative of inter-nations!

An exceptional evening!