HEMO NEWS EU

Bulletin n°6, May 2018 | The newsletter of the European Federation of Associations of Patients with Haemochromatosis

THE PRESIDENT'S MESSAGE

Barbara Butzeck

EFAPH was delighted to include the Dutch association (HVN) as a new member association in 2017 and to welcome back the Swiss group in February 2018 with Dr. Sarah Ersözlü as their new representative. Additionally, EFAPH is looking forward to the inaugural meeting of a new Swedish Haemochromatosis Association, held in Stockholm at the end of April. Thanks a lot to Ketil Toska from Norwegian HH Association (NHF) for his tremendous work in preparing this meeting. After fruitful co-operations between EFAPH and HI to develop international recommendations on HFE-Haemochromatosis for patients, adopted at the BioIron Meeting in Los Angeles in May 2017, we are happy to announce another joint project: World Haemochromatosis Week 04.-10. June 2018. ■

MÜNSTER AND ZÜRICH AGM HIGHLIGHTS: JUNE 25th, 2017 FEBRUARY 11th, 2018

The Münster 13th AGM, held June 25th 2017 saw strengthening relationships between EFAPH and HI[1], EURORDIS, EUROBLOODNET and HARI. Amongst the various topics discussed was a questionnaire on 'Quality of Life'. The 14th EFAPH AGM held after the European Iron Club meeting in Zürich showed the diversity of interests developed by EFAPH. A very lively AGM discussed recent scientific advances in HH reviewed by prominent scientists and clinicians in the field. New members of EFAPH include the Netherlands and discussions are ongoing to form associations in Sweden and Romania. HI has published their definitive patient guidelines[2] for treatment of HH which are simple, objective and practical for patients, and based on published scientific studies and guidelines. Additionally, HARI has also published advice for patients, available in several languages for the treatment of HH-arthropathy.[3] ■

1 HI: Haemochromatosis International, **2** Hepatology International, Adams, P., Altes, A., Brissot, P. et al. Hepatol Int (2018). https://doi.org/10.1007/s12072-018-9855-0
3 See EFAPH and Association websites

EFAPH HONOURS

the European Iron Club
For Professionals in Biomedical Inorganic Iron

After two successful European Iron Club (EIC) meetings in Münster and Zürich, HemoNews salutes not just the dedicated army of researchers, physicians and patients who have contributed greatly to progress in the treatment of haemochromatosis (HH), but also a younger generation who will no doubt continue this progress. These relative newcomers gave exhilarating and crystal clear presentations at the meetings. The union of meetings of EIC and EFAPH has been tremendously rewarding. Now so much more is known about the etiology of HH thanks to research presented at EIC meetings, at the biennial International BioIron Meetings and in international journals. Thanks to the discovery of hepcidin, a small protein which controls iron metabolism, there have been enormous advances in our understanding of the disease and new possibilities for treatment in evolution. ■

Calendar 2018

February 11th, 2018: EFAPH AGM, Zürich, Switzerland
April 5th, November 8th 2018: EuroBloodNet, Brussels, Paris
April 21 th, 2018: Inaugural Meeting Swedish Haemochromatosis Association, Stockholm
April 29th, June 16th and October 2018: HARI Meetings (London, Amsterdam, Freiburg)
May 10-12th, 2018: European Conference on Rare Diseases Meeting (ECRD), Vienna, Austria
June 4-10th, 2018: World Haemochromatosis Week
June 13-14th, 2018: International Blood Donor Day
December 11-12th, 2018: Council of European Rare Disease Federations, Paris or Brussels.
May 5-10th, 2019: International BioIron Meeting, Heidelberg, Germany (EFAPH and HI AGM probably May 11th, 2019)

EFAPH
European Federation of Associations of Patients with Haemochromatosis

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CONTROVERSY!

Should we treat HH patients with ferritins 300-1000 microgm/L?

A recent clinical trial* (Ong SY et al, Lancet Haematology 2017, 4, e607-614, Comment: page e569) addressed this important question. A total of 94 blinded patients participated (50 control and 44 treated). Patients in the treatment group were erythrocytapheresed (removal of red cells from the blood) every three weeks until their ferritin levels declined to 300 microgm/L (upper limit of normal), whereas the control patients were plasmapheresed (removal of blood plasma only). The number of treatments for the control group was calculated from predictions of how many procedures would be required to reduce their ferritin level to 300 microgm/L if they were being venesected. The aim of the study was to establish whether decrease in ferritin to 300 microgm/L causes improvement in non-specific symptoms such as fatigue, lethargy, generally feeling unwell, and in hepatic fibrosis markers. A modified disease impact scale (MDIS) and hepascore (a measure of liver fibrosis) was used to assess patients prior to and at the end of the study to determine any improvements. MDIS consists of a questionnaire which probes cognitive, physical and psychosocial functions in individuals. At the end of study, there was a significant decrease in MDIS in the treated group relative to the untreated, both in total and in the cognitive section related to mental well-being. A lowered MDIS score indicates an improvement in well-being. Hepascore also decreased slightly in the treated groups and there was lowered lipid peroxidation in the plasma of the treated group. Other parameters studied included Arthritis Impact Measurement, Hospital Anxiety and Depression Score and the Medical Outcomes Study Health Survey. Generally scores in these surveys showed patient improvement in the treatment relative to the control group, however, these improvements were not statistically significant.

These results show significant short-term benefits in the treatment of patients with ferritins 300-1000 microgm/L, some of whom were asymptomatic. Normalising serum ferritin at these levels would prevent progression to levels in excess of 1000 microgm/L associated with the more severe complications of haemochromatosis. Whether the short term benefits of reducing serum ferritin to below 300 microgm/L are similar to the findings of this trial is at present unknown. The data suggest that patients with moderate iron overload benefit from normalisation of serum ferritin. These findings surely augment the case for introducing screening for haemochromatosis into the community.

EFAPH Comments

This is an important Australian study showing some clinical benefit of treating HH patients as soon as the ferritin level exceeds the upper limit of normal (300 microgm/L). It provides insights into the debate over the threshold value of ferritin at which treatment is initiated, especially as some authors have proposed to wait for a level of 1000 microgm/L before starting venesection therapy. The conclusions of Ong SY et al are in full agreement with the therapeutic recommendations published by an expert panel from Haemochromatosis International, in the Journal "Hepatology International".

Adams, P., Altes, A., Brissot, P. et al. Hepatol Int (2018). https://doi.org/10.1007/s12072-018-9855-0

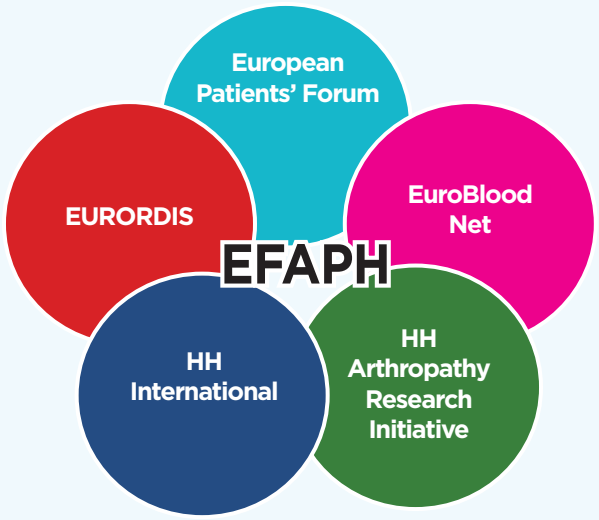
A strong patients' voice

A strong patients' voice to drive better health in Europe is the slogan of the European Patients Forum (EPF). One of their programs 'Taking action – A roadmap to achieving universal health coverage for all patients by 2030' was further explained by a session in the European Parliament in December 2017. Five key-topics were highlighted, namely:

1. Ending discrimination and stigma
2. Encouraging affordability of healthcare products and services
3. Implementing access to a holistic range of health and related services
4. Providing access to quality of care
5. Committing to sustainable investment in health

All topics were explained with tips and tricks. The entire document can be found on http://www.eu-patient.eu/globalassets/campaign-on-access/taking-action-a-roadmap-to-achieving-universal-health-coverage-for-all-by-2030.pdf. ■

EFAPH: Your voice in Europe



Representatives:

HARI: Barbara Butzeck,
HI: Robert Evans,
EuroBloodNet: Graça Porto/Dorine Swinkels,
EURORDIS: Maria Abele/Françoise Courtois,
EPF: vacant at present!

EFAPH NEEDS MORE VOLUNTEERS, CAN YOU HELP US IN ANY WAY?

EXPERTS AND PATIENTS COME TOGETHER TO PUSH HH BOUNDARIES IN BIRMINGHAM

Dr Patrick Kiely (London) receiving a glass momento marking the inaugural Fernau Lecture 2017

The Haemochromatosis Society UK organized a stimulating medico-scientific conference with five broad topics as below (New Understanding of Genetic Haemochromatosis Meeting, March 2017):

The Role of Transferrin Saturation (TS) Testing: Hepatologist Professor Pierre Brissot (Rennes, France), Haematologist Professor John Porter (UCL, London) agreed that the role of TS testing was critical in a diagnosis of iron overload, and crucial for monitoring patients in the maintenance phase. During de-ironing, monitoring TS is not essential as transferrin remains close to 100% saturation until storage iron reaches normal levels - indicated by serum ferritin level. TS testing can quickly identify when patients are starting to reload iron. New guidelines from the British Society for Haematology are currently available (see UK website) and include testing for TS which should be maintained at or below 70%, ideally below 50% in the maintenance phase. Prof Brissot referred to the 50/50 rule, maintaining ferritin and TS at 50 microgm per litre and 50% respectively.

Haemochromatosis Arthropathy (HA) as a marker for HH: Delay in diagnosis of HH leads to serious health consequences with early diagnosis crucial. In the inaugural Fernau Lecture, Dr Patrick Kiely presented his research clearly identifying HA as an early marker for HH with the potential to reduce the average age of diagnosis by 5 years or more. Explanations of how HA manifests itself led to debate about the level of understanding and awareness that could be expected of healthcare professionals such as GPs.

New Technologies for Measuring Liver Iron and Damage: Three eminent scientists Professor Tim St Pierre, (Australia), Consultant Radiologist Dr Barbara Butzeck (Germany) and Dr Rajarshi Banerjee (UK), discussed MRI scanning technology as a diagnostic tool for HH. The UK Haemochromatosis Society advocates the use of MRI to measure liver iron concentration (LIC) and liver damage as an alternative to biopsy which must however continue where other clinical information is needed from the liver tissue itself.

The Complex Genetics of HH: Ms Patricia Bignell (Oxford) introduced the wide range of genes and mutations known or suspected to cause iron imbalance, presently being investigated by Next Generation Sequencing (NGS). In some cases of HH there could be more than one mutated gene and the definition of HH as a simple recessive monogenic condition is now being questioned. However, there are still concerns about the costs and necessity for NGS, as the genetic cause of iron overload is largely academic because treatment is simple and based on blood analyses rather than molecular diagnosis.

The Role of Blood Donation: Consultant Haematologist Dr Naim Akhtar (UK) discussed difficulties in UK blood service in the management of HH patients experienced by clinicians and patients. ■

IRISH HH PATIENTS FORCED TO PAY HUGE SUMS FOR VENESECTION!

The Irish Government has introduced a charge of €80 per venesection in certain hospitals (maximum charge of €800/year). This unjust charge affects patients without a medical card (for those on low incomes) or private health insurance. Some patients have cancelled vital appointments. The Irish Haemochromatosis Association (IHA) has written several times and sent a petition to the Irish Health Minister, Simon Harris. IHA also led a brief European survey showing a unique situation. EFAPH wishes to help the IHA to remove this unacceptable charge. ■

EFAPH and HI join forces for:
World HAEMOCHROMATOSIS Week
June 4-10th, 2018

Hungary: A Lucky Coincidence!

Hungary's Heroes: Maria Mörtl, Maria Abele and Prof. Judit Várkonyi

After a 10 year campaign mainly by Prof. Judit Várkonyi, the honorary president, HH patients are now accepted as voluntary blood donors in Hungary. The main reason for non-acceptance was that the principle of voluntary donation was violated when the donation is through therapeutic venesection. A lucky coincidence occurred when a journalist wrote an article about the Hungarian Society for a medical website and a widely read newspaper whilst there was a dramatic shortage of blood in Hungary. The new leadership of the Blood Bank Service then agreed that HH patients could donate blood five times per year under the same conditions as non-HH donors. Patients have to declare their HH disease and if regular requirements of donation are fulfilled, their blood can be used for transfusion purposes. Donors now receive a thank you SMS from the Blood Bank every time their blood is used!.

Two Useful Danish Initiatives:

1. The incidence of detection for the HFE mutation C282Y in Denmark has been extremely low for many years: in 1951-1975 about 4.5/ million persons/ year. However, this has recently risen from 18.7 in 2009-2010 to 74.4 in 2017. Thus in 2017 a total of 330 persons were newly diagnosed with HH. Although a very positive achievement for our patients, the Danish Haemochromatosis Association (DHA) is still far from its goal to identify the 20 000 Danes who are C282Y homozygous!

2. DHA has striven to increase awareness of haemochromatosis among health personnel since 2009. Some blood banks in Denmark have now started to measure serum ferritin in blood donors, a service which should be routine in all blood banks!

Two important initiatives from HARI

Haemochromatosis Arthropathy
HARI
Research Initiative

• HARI experts have prepared simple, practical advice on life style choices for patients with arthropathy, translated into several languages and available both on the EFAPH and National Association websites. EFAPH recommends affected patients to visit these websites.

• A document is in the process of validation by HARI experts to aid GPs in the early diagnosis of HH using the characteristic arthropathy which could bring diagnosis forward by some 7 years avoiding many of the later complications of HH.

EFS
ÉTABLISSEMENT FRANÇAIS DU SANG
Du donneur aux patients

EFAPH welcomes Netherlands !

HVN
Hemochromatose Vereniging Nederland

Don't forget!!!

World Haemochromatosis Week
04 – 10 June 2018

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Join us on www.efaph.eu

We need your support to raise funds to continue our work on your behalf!

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