

## THE PRESIDENT'S MESSAGE



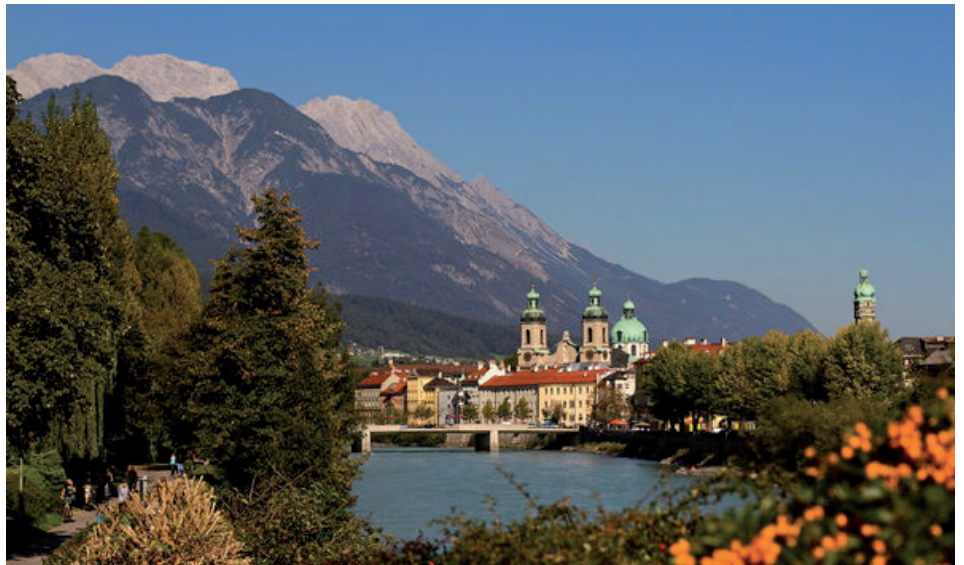
Barbara Butzeck

2016 saw progress in several EFAPH ongoing projects: the Rheumatology Initiative on HH-Arthropathy HARI, launched in Cologne in 2015,

had two face to face meetings.

Prof Graça Porto did an amazing job implementing Haemochromatosis Reference Centres in EuroBloodNet. Unfortunately EFAPH lost Jean-Daniel Kahn as senior advisor, and myself and the board are delighted that Betty Coueslant is now handling all financial aspects. The board attracted more collaborators: Annick Vancloster from HVV will represent EFAPH in EPF; Dr Patricia Evans, UK has become editorial associate of the HEMO NEWS. ■

## INNSBRUCK : AGM NEWS



Accompanying the annual meeting of the European Iron Club, a joint AGM for EFAPH and HI (Haemochromatosis International) was held in Innsbruck, Austria in April, 2016 to discuss current topical issues.

Highlights included discussion of the development of international guidelines for treatment, a European survey on blood donation from HH patients (funded by EFS) and an introduction to the new non-HFE registry project headed by Prof Heinz Zoller (see later).

## EFAPH HONOURS



Dr Margaret Mullett at her graduation

**Margaret Mullett**, (Chairwoman of the Irish Haemochromatosis Association) was awarded the honorary degree of LLD (Doctor of Laws) in 2016 for her role in the early diagnosis and care of patients suffering from HH. Thanks to Margaret, a formidable integrated network of patients has been created throughout Ireland and membership has increased over 50-fold. This is a highly appropriate honour for a lady who has devoted much of her time to raise the profile of HH both in Ireland and internationally against a background of her own personal suffering. ■



Prof Pierre Brissot: brilliant academician and physician !

**Pierre Brissot** has devoted much of his working life to the treatment of patients with genetic haemochromatosis whilst still finding time to conduct internationally acclaimed research into iron metabolism. In recognition for his great contributions in these fields, he has been elected to membership of the Académie Nationale de Médecine (December, 2016), the highest honour which can be awarded to a doctor in France. EFAPH and haemochromatosis patients worldwide profoundly thank Pierre for his huge contribution. ■

## Calendar 2017

- May 7 - 12<sup>th</sup> 2017:** Biolron meeting , HI AGM, Los Angeles USA
- May 19 - 20<sup>th</sup> 2017:** EURORDIS AGM in Budapest
- June 2017 (Country dependent):** European Haemochromatosis Awareness Week
- June 14<sup>th</sup> 2017:** World Blood Donor Day
- June 24 - 25<sup>th</sup> 2017:** EFAPH AGM + EIC Muenster, Germany
- June 22 - 25<sup>th</sup> 2017:** EHA (Eur Haematology Ass) EuroBloodNet Meeting, Madrid
- March 2017 and October 2017:** HARI meetings
- October 26 - 27<sup>th</sup> 2017:** Council of European Rare Disease Federations, Paris
- February 8 - 11<sup>th</sup> 2018:** EIC Meeting, Zurich

## DID YOU KNOW...



Elite sport is an arduous pursuit with heavy physical and mental demands on the human body. Heavy exercise induces haemolysis of red blood cells and bleeding through the gut leading to anaemia. Hence many elite athletes take iron supplements but the effects of this on performance have not been generally investigated. However, at least two scientific papers have been published suggesting that increased body iron may enhance athletic performance. In a Spanish study of 65 male elite athletes (cyclists and endurance runners), there was a statistically significant difference in the prevalence of C282Y and H63D mutations compared to non-athletic controls. Another study, this time using data from French

Nordic skiers, rowers and judo fighters (129 altogether), showed a frequency of mutation of 80.4% among athletes reaching the top three places in international competitions. HFE mutations could increase parameters involved in oxygen delivery to muscles such as haemoglobin concentration and haematocrit. Higher iron bioavailability might also influence rates of repair of damaged muscle tissue and resistance to fatigue. Thus the presence of an HFE mutation may confer an advantage in elite endurance sport. Questions remain about the health of these individuals when retired from competitive sport. This also provokes further examination of other possibly favourable effects of the HFE mutation. Why has this mutation persisted when its effects are so disastrous? The ability of individuals to absorb greater amounts of iron impacts on growth and cognitive development markedly in populations suffering from iron deficiency anaemia. High absorbers would have had a selective advantage in producing offspring perpetuating the mutation whilst the limitation of human life span in previous centuries probably meant that individuals died before the

onset of life-threatening symptoms of iron overload. Thus the mutation would confer an advantage during childhood and the reproductive years causing it to persist. ■

*Do you have an article or a story of interest to contribute?*

*We'd love to hear from you. Please contact us via the website.*

### Main source papers used

1. **Iron supplementation in athletes- first do no harm.** H. Zoller & W. Vogel. *Nutrition* 20, 615-619 (2004)
2. **Mutations in the hereditary haemochromatosis gene HFE in professional endurance athletes.** J.L. Chicharro and others. *British Journal of Sports Med* 38, 418-421 (2004)
3. **80% of French sports winners in Olympic, World and European Competitions have mutations in the HFE gene.** O. Hermine and others. *Biochimie* 119 1-5 (2015)
4. <https://www.ucbl.nlm.nih.gov/pubmed>

## Update on Haemochromatosis International



HI, formed in 2011, is committed to improving the health of people with haemochromatosis worldwide. Most of our business is conducted by e-mail and Skype with Committee members originating from Australia, Brazil, England, Germany and Portugal. Members have identified three key areas where HI can make a useful contribution:

- facilitating the sharing of resources ([www.haemochromatosis-international.org](http://www.haemochromatosis-international.org))
- co-operating to raise awareness of HH worldwide
- developing international recommendations for the treatment of HH. HI hopes to extend these ideas in the next AGM in Los Angeles. ■

## HARI - a promising initiative in Haemochromatosis Arthropathy

HARI was launched at the EFAPH AGM in Cologne in 2015. The group comprises 7 rheumatologists from UK, France,



Germany, Austria, Canada and Australia and Barbara Butzeck as patient representative. After 3 meetings in 2016/17, the group is ready to publish advice for patients about treatment. Subsequent documents will show bone lesions and the pattern of HA to raise awareness of GPs and orthopaedic surgeons. The design of a study is in discussion to correlate genetic mutations, degree of iron overload and level of pain. ■





**Prof Graça Porto,**  
Scientific Committee  
President

The European Commission (EC) has been preparing a program to establish European Reference Networks (ERNs) on Rare Diseases since 2012. A call for interests was recently opened (March-July 2016) resulting in the approval (on the 15 December) by the ERN Board of Member States, a total of 24 network applications on different groups of diseases involving 370 hospitals and almost 1000 highly specialized units from 26 countries.

The launch of the Networks is foreseen during a major conference organized on 9 March 2017 in Vilnius. One of the approved ERNs was the EuroBloodNet, a network linking 66 highly specialized centres. (from 15 countries) on rare haematological diseases (RHD), including oncological and non-oncological (including haemochromatosis) disorders, organized in two hubs coordinated respectively by Prof Pierre Fenaux (Hôpital St Louis, Paris) and Prof Béatrice Gulbis (Laboratoire Hospitalier Universitaire de Bruxelles). They submitted the application in a joint venture with the participation of ENERCA (a network for rare haematological disorders led by Prof Vives-Corrons) and the EHA (European Hematology Association) Advocacy Group with

a strong input from the Patient Associations (EFAPH included). The main objectives of EuroBloodNet are to: 1) improve equal access to highly specialized healthcare delivery across Europe 2) promote the best practices in prevention, diagnosis and safe clinical care across Europe 3) disseminate cutting-edge knowledge and facilitate continuous medical education 4) provide inter-professional consultation by sharing of expertise and safe exchange of clinical information and 5) foster European cooperation in highly specialized procedures for diagnosis, innovative treatments and research. The role of EFAPH in this process should be highlighted for the longstanding efforts to promote the inclusion of hereditary hemochromatosis (HH) in the network and fostering the participation of well recognized HH centres, 7 of which (from France, Italy, Netherlands and Portugal) succeeded in being included in the EuroBloodNet first call. We consider this as a first step to make haemochromatosis more visible and assure that the voice of HH patients is heard and that they are not excluded from this tremendous ERNs effort to improve the patients' healthcare and overall quality of life and to develop more evidence-based clinical tools and cost-effective approaches. Next steps are aimed at including more centres in the network and assuring an active participation of HH centres in EuroBloodNet activities. ■

## REGISTRY STUDY FOR NON-HFE HAEMOCHROMATOSIS



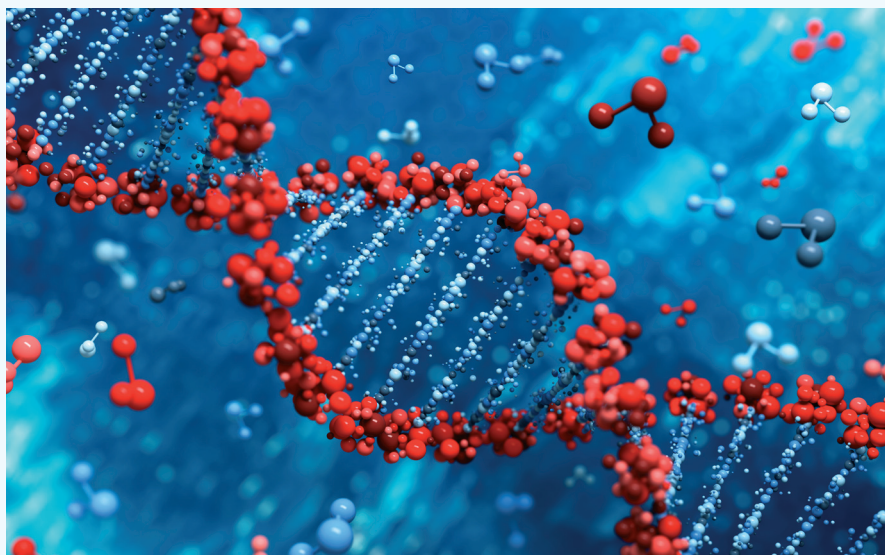
**Prof Heinz Zoller,**  
Medical University of  
Innsbruck, Austria

Identification of the HFE protein has transformed diagnosis of haemochromatosis and thereby

helped patients to access life-saving therapy at pre-symptomatic stages of haemochromatosis. However, around 15 % of patients with clinical haemochromatosis do not carry a mutation in the HFE gene but have mutations in other genes affecting iron metabolism which lead to iron overload. This group of diseases is known as "non-HFE haemochromatosis". One example is ferroportin disease where there is a mutation in the gene encoding the only known cellular iron exporter. In contrast to HFE-associated haemochromatosis, which is caused by a single point mutation, non-HFE haemochromatosis is caused by a variety

of mutations in different genes. Simultaneous testing for mutations in multiple genes now allows non-invasive and rapid detection of such mutations. Although this technological breakthrough holds great potential to further improve patient care, the implications of novel variants are often unknown. The aim of this international "non-HFE registry" study is to collect infor-

mation on the clinical presentation, biochemistry, radiology, family history, genetics and histology of patients with non-HFE haemochromatosis. This will help us to understand any clinical effects of each disease variant and to determine the effect of specific interventions and hence ultimately improve patient care. ■  
see: [www.non-hfe.com](http://www.non-hfe.com)





## Important changes in Denmark



**Dr Nils Milman vice chairman of the Danish Haemochromatosis Association**

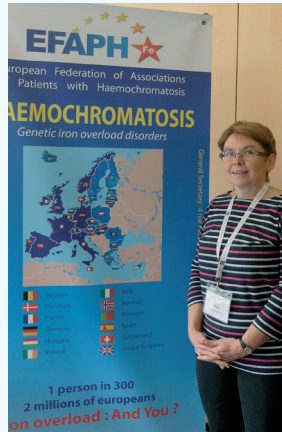
In a population of 5 million ethnic Danes and 0.5 million individuals belonging to other ethnic groups, approximately 20,000 are C282Y homozygous and 0.5 million are C282Y heterozygous. During 2016, the DHA was reorganized and promoted awareness of haemochromatosis in Denmark by sending a recruitment flyer to all GPs. Previously the treatment of haemochromatosis was through "general internal medicine", meaning that all doctors within the specialty

of medicine, including GPs, were authorized to treat the disease, which has proved highly unsatisfactory. Following negotiations with the National Board of Health, the Danish Society of Gastroenterology and Hepatology (DSGH) was accepted as the coordinator for Haemochromatosis rather than Haematology. The Danish Haemochromatosis Association consider this achievement to be a great step forward for diagnosis and treatment of these patients. EFAPH congratulates DHA on this hard-won achievement.



## Focus on Hungary

The Hungarian Association (HBE) has around 30 members, a small, but active group! Most members live in the capital, Budapest. Prof Várkonyi (a leading haematologist) along with this association are fighting for transferrin saturation to be used as a general screening indicator. However, not all GPs can perform this test through social insurance. There are no guidelines for treatment and phlebotomized blood is prohibited from use in transfusions. In terms of awareness, leaflets and posters were available to all participants of the Hungarian GPs' Conference last year. New patients are also briefed on some HealthCare Days and each newly diagnosed patient receives the patient information leaflet. Additionally, HBE has close and very helpful contacts with the Hungarian Rare Disease Group. The most difficult thing is to get haemochromatosis recognized as in other countries.



**Maria Mörtl, HBE secretary**



## Ground-Breaking Meeting of the UK-HS

The Haemochromatosis Society UK organized an international conference entitled 'New Understanding of Genetic Haemochromatosis' in Birmingham in March 2017. Highlights included a debate on the role of transferrin saturation as a diagnostic and monitoring tool, the Fernau Lecture on haemochromatotic arthropathy and advances in the genetics of haemochromatosis. There was also a focus on MRI technology in the diagnosis of iron overload. Highlights will be reviewed in the next edition of HemoNews. Don't forget to get your copy!!!!



**DETECT/FER**  
Semaine nationale de l'Hémochromatose

## A fourth awareness campaign completes the French Federation initiative at the national level!

After the creation of haemochromatosis health care channels in the Paris area along with the launching of a pilot regional awareness campaign with the backing of the regional Health agency (FERIF), FFAMH is initiating a promising nationwide campaign (DETECT/FER) targeting GPs and specialist doctors involved in haemochromatosis. Approaching every stakeholder - biologist unions, MG unions, specialist doctors unions, health ministry and scientific societies up to 12-14 months ahead of kick-off should ensure the success of this action. Indeed, the setting up of a steering committee involving every player cannot but be a win-win. FFAMH is so far benefiting from their expertise, their financial support and in due course will benefit from their communication network. As for their partners these are committing themselves to a project which will help the patient community and will no doubt be quite rewarding for them. Let's keep our fingers crossed!



## Thanks Jean-Daniel!



A thousand thanks to Jean-Daniel Kahn for the huge and diverse contribution he has made to our federation. We hope that he will continue to give us precious advice when needed. We will miss him greatly.

Join us on  
[www.efaph.eu](http://www.efaph.eu)



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