

# HEMO•NEWS

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## Who are we?

**EFAPH is a European network of Patients' Associations currently covering 10 countries:**

Belgium (HBA), France (FFAMH), Germany (HVD), Hungary (HBE), Ireland (IHA), Italy (AplSdE-M), Norway (NHF), Spain (AEH), UK (Haemochromatosis Society UK), Switzerland (Hämochromatosen selbst-hilfe Group Weinfelden).

### *An International Network*

EFAPH is a founding member of the International Alliance of Haemochromatosis Patients Associations (IAHA) created in 2011 at Vancouver.

### *Objectives*

To raise the awareness of Hereditary Haemochromatosis (HH) in all European countries among both health professionals and the general public.

To promote information and to help GPs in the early diagnosis of the disease, to implement good practices

and to support further research on the genetic diseases of Iron metabolism.

### *Goals*

To initiate new national associations in Europe, to support their develop-

ment, to create common tools and to promote collective actions such as The European Haemochromatosis Week (first introduced in 2010).



*Representatives at the 8th.AGM in front of the Hepatology Dpt. of the Pontchaillou Hospital, Rennes, France.*

*1st row, 3rd/4th from the left: Pr Graça Porto (Scientific Committee, President), Jean-Daniel Kahn (Treasurer); 2sd row from the left: Dr Barbara Butzeck (President), Dr Françoise Courtois (General Secretary), Pr Pierre Brissot (Honorary President).*

## What should be known about Hereditary Haemochromatosis (HH)

### *HH is an iron overload disorder:*

The daily iron intake amounts to 1 to 2 mg (of which 70% are used to

produce haemoglobin and 30% for other cells). People who absorb too much iron accumulate it in the liver, the pancreas, the heart, the joints... resulting thus in haemochromatosis. If

not treated the complications can be quite serious: cirrhosis, liver cancer, diabetes. Other difficulties such as: osteoporosis, arthropathy, hypogonadism may also occur in the long term.

## Early diagnosis is essential:

before complications occur. The first symptoms, being non specific, can result in an incorrect or late diagnosis: abnormal and chronic fatigue, joints pains, cardiac arrhythmia, diabetes, sexual difficulties, grey pigmentation of the skin. A simple blood test is sufficient to measure the ferritin (iron storage) and the transferrin saturation (iron transportation in the blood)..

## Diagnosis

Any anomaly must lead to:

- a genetic test to detect possible mutations of the gene HFE (HH type 1, the most frequent),
- treatment.

In most cases the complications occur around the age of 45 for men and 50 for women. Hence the importance of an early diagnosis.

## A genetic disease

Type 1 HH is the most frequent (the prevalence is 1 out of 300 in Europe, i.e. 2,2 millions people have a genetic predisposition), but all will not have the disease. The prevalence is higher in French Brittany ( 1 out of 200) and in Ireland (1 out of 80). Rare forms can develop not only for adults but also for children or teenagers; the corresponding genetic mutations are different (4 types), are most often very severe and can be observed worldwide.

## A simple treatment

Periodic blood letting, known as venesection, (initially 400 to 500 ml) will progressively offset the iron overload. If a young patient (say 30 years old, before the appearance of complications) the main symptoms will fade off.

## Hepcidine: a key element in iron metabolism

This hormone is the one specific to iron metabolism; it monitors the iron intake and output of the organism. A full understanding of its action should in time be a very helpful discovery.

# Highlights 2011/2012



From left to right: Dr. Françoise Courtois (EFAPH, General Secretary), Margaret Mullet (IHA President), Bernard de la Villardière (Journalist and moderator), Dr. Barbara Butzeck (EFAPH, President), Antonya Parvanova (MEP, ENVI Committee) Pr. Pierre Brissot (Hepatologist, CHU Rennes-France, EFAPH Honour Vice President), Dr. Rolland Lemye (Vice President of Permanent Committee of European Medical Practitioners - CPME).

## I - I. A lunch-debate organized at the European Parliament (Brussels, 09/20/2011)

The preparation of this event took one year with the objective of raising the awareness of Authorities and of

Health Professionals at EU level. The results were:

### A "written question"

signed by MEPs of the ENVI Committee and addressed to the European Commission. The question was: "Does the Commission consider:

- taking initiatives to promote the early diagnosis of HH (programmes for raising awareness and information of the general population and health professionals – general practitioners)?
- promoting research on genetic diseases?"

## **“Reply from the European Commissioner Mr. Dalli”**

(February, 2012),

considered as very qualified by the concerned MEPs:

- The Commissioner agreed that HH is indeed underdiagnosed,
- The knowledge of the prevalence and penetrance of HH must be improved by clinical and epidemiological research,
- The role of the Commission is not to issue guidelines for the early diagnosis of HH,
- There should be room in the future European financing programmes (FP7 and FP8, 2014-2020).

A real opportunity for EFAPH! Hence some medium and long term projects.

## **II - The Annual General Meeting (AGM) of EFAPH and the 8th. European Iron Club (EIC), Rennes 2012**

As every year the EFAPH AGM took place at the end of the EIC Congress which was held this year in Rennes between August 29th. and September 1st. This congress, organized by Pr. Pierre Brissot (heading the Liver Diseases Dpt.- Rennes CHU) and by Dr. Olivier Loréal (Research Director at INSERM-Rennes), was attended by 150 researchers. Some of them are members of EFAPH's Scientific Committee: Pr. Graça Porto – President (Portugal), Pr. Robert Evans (UK), Dr. Mayka Sanchez (Spain), Dr. John Ryan (Ireland), Dr. Olivier Loréal (France).

## **The EFAPH AGM included representatives from several countries.**

Guest: Pr. Odile Kremp (Orphanet-INSERM US14, France) presented the EU recommendations with respect to Reference Centres for Rare Diseases (ERN).

### **Main achievements:**

1. Findings of the 1st. European survey of the genetic information provided to patients (Pr. Graça Porto)
2. Pilot project of an Observatory composed of GPs made aware of HH in the Yvelines Département, France (Dr. Françoise Courtois)

### **Main proposals for projects:**

1. **Implementation of an European Network of Reference Centres (ERN) for iron metabolism diseases,** headed by Pr. Graça Porto (Chair of EFAPH Scientific Committee). The situation is currently very diverse in Europe; the European regulation is being finalized (end of 2012). In France the Centre National de Référence des Surcharges en Fer Rares d'origine Génétique (National Reference Centre for Rare Iron Overloads of Genetic Origin) is located in Rennes; it is headed by Pr. Pierre Brissot (CHU Rennes) and coordinates 9 French competence centres as well as genetic diagnostic labs. These centres are closely related to patients and their associations.

Within EFAPH only 3 countries have a National Reference Centre (France, Portugal and Norway); the most structured is the Rennes Centre. In Spain there is a coordination of expert centres and in 6 other member countries there are several uncoordinated expert centres: Italy, UK, Ireland, Germany, Hungary and Belgium.

This project will be very time consuming for EFAPH Scientific Committee; it implies close coordination with each member country, so that in the long term there will be a national reference centre in each country.

### **2. An epidemiological and clinical research programme on haemochromatosis related arthropathies is contemplated.**

It would be managed by the Scientific Committee of EFAPH in cooperation with Pr. Pascal Guggenbuhl (Rennes CHU).

### **Actions in progress: EFAPH expansion**

The national associations of Germany and Norway are pursuing contacts with a view to setting up national associations in Austria and Denmark in 2012-2013.

*Dr B. Butzeck speaking at the EIC Congress – Medical Faculty – Rennes University (France)*



# Future events

## • Friday October 5th., 2012

2nd. session of the ProAm Sailing and Golfing competition in Cicé-Blossac, France in support of EFAPH



## • Thursday, February 21st., 2013

Charity piano recital in support of EFAPH by the international artist Anne Queffelec. Salle Cortot, Paris,



## • April 14th to 18th, 2013

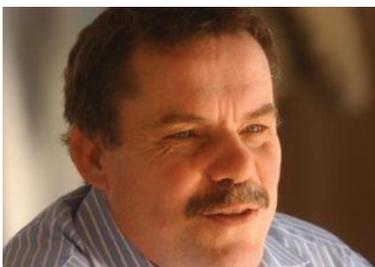
The World Congress of the International Bioiron Society (IBIS) at the University College of London (UK). The Annual General Meeting of EFAPH will take place on **April 19th, 2013**.

# A enthusiastic sponsor for EFAPH: Anne Quéméré

Anne is a well-known sailor, born in Quimper (French Brittany). She holds many sailing records (in 2002 and 2004). She has rowed across several oceans alone (first world records in 2006 and 2011). Very generous and enthusiastic, and interested in the haemochromatosis cause, she has agreed to be the international face of EFAPH.



# Death of Pr Jean Paul Moisan



Pr. Moisan, a specialist in medical genetics, was the first in France to develop genetic prints for forensic medicine. A universally recognized NDA specialist, he created in 2003 a forensic expertise laboratory, Institut Génétique Nantes Atlantique (IGNA).

He died accidentally last August off the Noirmoutier island (Loire-Atlantique). A co-founder of EFAPH, he was always very interested in the evolution of the Federation, and with his advice and expertise encouraged its development.