

HEMO NEWS EU

Bulletin n°7, April 2019 | The newsletter of the European Federation of Associations of Patients with Haemochromatosis

GENERAL SECRETARY'S MESSAGE



Dr Françoise Courtois

Last year's Hemonews (bulletin n° 6) reported highlights of the 2017 and 2018 EFAPH AGMs. The coming AGM in May, 2019 in Heidelberg will see significant changes to the personnel of the EFAPH Board. In particular, our President of 10 years Barbara Butzeck will be stepping down and I would like to reduce my workload and to begin to train a new general secretary. We already have a new member society in Sweden - the 15th EFAPH-member and se-

veral new volunteers are helping the EFAPH community: Dag Erling Stakvik from the Norwegian Society is the new patient ePAG representative (EuroBloodNet) and Howard Don from the HUK is responsible for EFAPH's dealings with EPF.

We have already begun consultations towards a closer partnership between EFAPH and Haemochromatosis International (HI) which will continue at the Heidelberg Meeting in May when both EFAPH and HI members will be present. The in-coming President of HI - Dr Dianne Prince - will be introduced at this Meeting which will also see probable changes to the classification of the various forms of haemochromatosis. There will be a special session on this topic during the International Bio-Iron Meeting managed by Prof Domenico Girelli. The Heidelberg Meeting promises to be very interesting and rewarding. A huge year ahead for EFAPH! ■

A GOOD ADVERT TO COPY!

Haemochromatosis

is a common genetic condition causing too much iron.

Tricky to say
Easy to treat
Simple to test
Tragic to ignore



Find out if your iron level is right.

This poster was designed by Haemochromatosis Australia (HA) and powerfully expresses much about haemochromatosis in few words. It could be translated into other languages with permission from HA (admin@ha.org.au)!

EFAPH HONOURS



Dr Barbara Butzeck
President



Patricia Evans
HI Secretary

This year we say good-bye to our President Dr Barbara Butzeck who is stepping down: her tireless efforts have helped to shape EFAPH into the successful organisation it is today. As well as being chairperson of the German Haemochromatosis Association (HVD), Barbara has been President of

EFAPH for over 10 years. She has invested so much of her free time to further the well-being of haemochromatosis patients both within EFAPH and as a committee member and trustee of Haemochromatosis International. As a consultant radiologist, Barbara has had a huge influence on the development of HARI which is extending the frontiers of haemochromatotic arthropathy awareness, diagnosis and research. This interest will continue as will Barbara's presidency of the German Association. We will miss her energy and commitment greatly but wish her well in any future enterprises she undertakes. EFAPH thanks her profoundly for her devoted and loyal service. ■

Vielen Dank, Barbara !

Calendar 2019

April 4th, 2019: ERN-EuroBloodNet 5th SSB meeting, Brussels

May 5-9th, 2019: International Bio-Iron Meeting, Heidelberg, Germany

May 9-10th, 2019: Joint EFAPH and HI AGM, Heidelberg, Germany

May 17-18th, 2019: EURORDIS membership meeting, Bucharest, Romania

June 3-9th, 2019: World Haemochromatosis Week

June 14th, 2019: International Blood Donor Day

June 14-16th, 2019: EULAR (HARI), Madrid, Spain

November, 2019: ERN-EuroBloodNet 6th SSB meeting, Paris

December, 2019: Council of European Federations (CEF), Paris

September 2nd-5th, 2020: European Iron Club (EIC), Oxford, UK

HAEMOCHROMATOSIS IS OVER 5000 YEARS OLD!



Skull of **BALLINAHATTY** farmer, Northern Ireland

A team of geneticists from Trinity College, Dublin led by Professor Dan Bradley and archaeologists from Queen's University Belfast, has sequenced the first genomes from ancient Irish humans, and the new information is already answering key questions about the origins of Ireland's people and their culture. A genome is an organism's complete set of DNA, including all of its genes. Evidence of massive migration to Ireland thousands of years ago has emerged from the sequencing of the first genomes from ancient Irish humans. These genetic influxes are likely to have brought cultural changes including the transitions from hunter-gatherers to agricultu-

ral workers, bronze metalworking and may even have provided the origin of the western Celtic language. The team sequenced the genome of an early farmer woman, who lived at Ballinahatty near Belfast some 5,200 years ago, and those of three men from a later period, around 4,000 years ago in the Bronze Age, after the introduction of metalworking. **Amazingly, the H63D haemochromatosis mutation was found when sequencing the genome of the woman farmer and the C282Y mutation was found in the Bronze Age men who lived over 4000 years ago on the Rathlin Islands, close to the Northern Ireland coast.** Whereas the early woman farmer had black hair, brown eyes and more resembled southern Europeans, the genetic variants circulating in the three Bronze Age men from Rathlin Island had the most common Irish Y chromosome type, blue eye alleles and the most important variant for haemochromatosis. The latter mutation is so frequent in people of Irish descent that haemochromatosis is sometimes referred to as a Celtic disease.

This discovery therefore marks the first identification of an important

disease variant in prehistory, according to the researchers. Its persistence into modern populations in these areas is probably due to the selective advantage of a greater ability to absorb iron in childhood in populations relying mainly on dietary cereal crops with low iron bioavailability. It has also demonstrated what a powerful tool ancient DNA analysis can provide in answering questions which have long perplexed academics regarding the origins of the Irish. ■

*Professor Dan Bradley, Professor of Population Genetics at Trinity College Dublin.
Bradley et al. Proc Natl Acad Sci U S A. 2016 Jan 12;113(2):368-73.*



Modelled head of farmer

Eurordis: always new initiatives



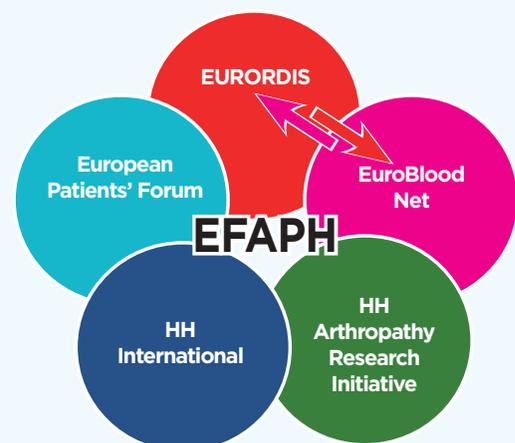
Dag Erling Stakvik from Norway

EURORDIS is a non-governmental federation representing 837 rare disease patient organisations in 70 countries. Most of these countries have a rare disease alliance encouraged to participate in European Reference Network (ERN) decision-making processes. Haemochromatosis has been included in the rare haematological diseases

ERN (EuroBloodNet) for 2 years. To ensure that patient voice is heard, a European Patient Advocacy Group (ePAG) has been created to improve health-care, quality of life and clinical tools. EFAPH now has a new ePAG volunteer, Dag Erling Stakvik from Norway. Dag participated for the first time in the Council of European Federations (CEF) Workshop in Paris, December 2018.



EFAPH: YOUR VOICE IN EUROPE



Representatives:

HARI: Barbara Butzeck,

HI: Dianne Prince,

EuroBlood Net: Dag Erling Stakvik/Graça Porto/Dorine Swinkels

EURORDIS: Maria Abele/Françoise Courtois,

EPF: Howard Don

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

HAEMOCHROMATOSIS CHALLENGES THE HOUSE OF COMMONS!



To raise awareness of the condition among front line clinicians and the public, with the ultimate goal of promoting earlier diagnosis, Haemochromatosis UK (HUK) organised a large meeting at The House of Commons, Westminster, London on 31st October 2018. A patient survey of 1,998 people (“LIVING WITH THE IMPACT OF IRON OVERLOAD: Report from a large survey of people with haemochromatosis”) was presented by David Head, chief executive of HUK. A further objective was to inspire individuals and organisations to improve the lives of people with haemochromatosis, urging adoption and further development of clinical guidelines, more research into the chronic aspects of haemochromatosis and the establishment of an All Party Parliamentary Group (APPG, being formed in January 2019) for Iron Overload. The event was attended by policymakers, clinicians, scientists, senior representatives from NHS trusts, Ministers and Members of Parliament (MPs) and other key influencers.

Corinna Towers (president of HUK) gave a powerful and influential account of her long struggle for diagnosis and treatment.

The survey found that many respondents experience psychological difficulties, sexual and skin problems, fatigue and arthritis or joint pain, forgetfulness or ‘brain fog’ and depression. The findings support the argument that HH needs to be perceived as a chronic disease. In looking at the diagnosis of the condition and the response of healthcare professionals, over one third of respondents said that their GP’s knowledge of haemochromatosis was inadequate or poor: half of people with haemochromatosis are not diagnosed when initially discussing symptoms with their doctor. The report highlights the extent to which chronic symptoms affect quality of life for haemochromatosis patients. There is also a strong correlation between delay in diagnosis and onset of more serious symptoms. ■

See free link of the report and accompanying Calls to Action document: <http://haemochromatosis.org.uk/2017-survey>
HUK has high hopes of raising awareness of HH through the APPG.



UK, HARI and EFAPH members at the commons

Breaking news from France: victory at last!



Haemochromatosis patients meeting blood donor criteria can now donate blood in the 124 EFS blood bank sites nationwide: a widening of access resulting from the signing of an amendment to the French blood bank regulations after months of close cooperation between FFAMH (French Federation for Haemochromatosis) and the French Health Authorities. ■



HARI marches on



• Two new papers were adopted in November 2018:

1. Could it be the arthritis that occurs in haemochromatosis ?
2. Osteoporosis and Genetic Haemochromatosis (GH)

These can be accessed on the EFAPH and HI websites

- HARI has two new members: Prof. Helen Keen (Australia) and Dr. György Nagy (Hungary).
- HARI was accepted as a study group at the ACR (American College of Rheumatology) in 2017.

Furthermore the HARI group is accepted as a EULAR (European League against Rheumatism) study group for

Haemochromatosis-Arthropathy and will attend their next conference in Madrid in June 2019, with the prospective of building a future task force for HH-Arthropathy.

The group is very proud of this progress in recognition. ■



Dr Graeme Carroll, Dr Patrick Kiely, Dr Stephanie Finzel, Dr Barbara Butzeck, Dr Axel Braner, Pr Pascal Guggenbuhl.



New swedish society forms!



Marten, Eleonor and Ing Marie

On Sunday 9th of December, 2018, Hemokromatosförbundet - the Swedish Haemochromatosis Association - was founded. The Association is organized as a federation of regional associations, so at the same meeting, the first regional group - the Stockholm group - was also founded. The board of both organisations consists of Mårten Fjällström (Chairman), Ing-Marie Bohmelin (Vice Chairman), Tobias Antin (Treasurer) and Eleonor Bergkvist (Secretary). The organization is still very new, but a recruitment drive and lectures are planned for the spring, with the hope of forming more regional groups in the future. The group hopes to gain enough size to qualify for regional (and then national) funding.

EFAPH wishes them the very best in the future!

New Information Leaflet HI/EFAPH

Diagnosis
Biochemical Test: A simple blood test called a serum ferritin measurement can:
 - Transferrin Saturation for iron transport protein.
 - A ferritin measurement above 200 is very suggestive of Haemochromatosis (HH).
 - Serum Ferritin (fast storage protein). An elevated result may be due to iron overload. The result should be interpreted in combination with transferrin saturation.
 - Iron overload through the blood.
 - Iron storage in the organs.
 A genetic test on blood or saliva will confirm the diagnosis. The genetic test for the HFE mutation C282Y/C282Y in HFE gene will detect the most common form of Haemochromatosis.
 Check the name of the HH specific mutations can be tested for in a specialised laboratory or a National Reference Centre.

A family case
 If the test is positive, it is the responsibility of the patient to inform their relatives and to encourage biochemical and genetic testing.

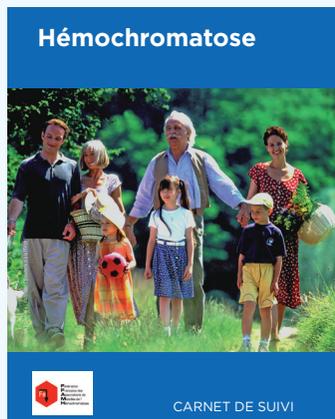
Objectives
 - To raise awareness of Haemochromatosis amongst the Medical Profession and the General Public to improve early diagnosis and treatment.
 - To assist in setting up national patients associations.
 - To promote international guidelines so that patients have equal opportunity for access to the best treatment worldwide.
 - To stimulate and to participate in research.

EFAPH is being member of Haemochromatosis International
HI Haemochromatosis International
 E-mail: info@haemochromatosis-international.org
www.haemochromatosis-international.org

The previous leaflet has been updated to produce a combined publication between HI and EFAPH. As there will be no information stands in Heidelberg, this leaflet will be included in the information pack of each Bio-Iron and HI/EFAPH participant.



A new FFAMH follow-up booklet in circulation



FFAMH has designed a new edition of its haemochromatosis follow-up booklet to meet a constant demand from both patients and health professionals. A double page devoted to osteo-articular complications has been added at the request of a French rheumatologist involved in the Hari Group. It will no doubt be much appreciated by patients who have to live with these complications.

Don't forget!



Labels are now available for this year's World Haemochromatosis Week. The labels are available in 11 languages as either transparent PNG or EPS versions
<http://haemochromatosis.org.uk/whw-labels/>
 (password "international")



Two important communication initiatives from the portuguese association (APH)

Two symposia with 150 GPs were held last year where the topics discussed were iron, haematology and clinical cases. World Haemochromatosis Week was covered on Portuguese national radio and TV giving maximal exposure.



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