

MINUTES OF THE 12TH ANNUAL GENERAL ASSEMBLY Innsbruck – April 10th 2016

Report : Françoise COURTOIS, Barbara BUTZECK, Jean-Daniel KAHN, May 20th, 2016

Agenda: *Appendix 1*

In Attendance: ABELE Maria (Hungary), BRISOT Pierre (France), BUTZECK Barbara (Germany), COUESLANT Béatrice (France), COURTOIS Françoise (France), DISTANTE Sonia (Norway), DON Howard (UK), EVANS Robert (UK), EVANS Patricia (UK), GIRELLI Domenico (Italy), KAHN Jean-Daniel (France), LOREAL Olivier (France), MARRIS Ben (Australia), MILMAN Nils (Denmark), PINEAU Brigitte (France), PORTO Graça (Portugal), SANCHEZ Mayka (Spain), SANTOS Paolo (Brasil), SEVERN John (UK), SORRILL Robert (Italy), STAKVIK Dag Erling (Norway), TEIXEIRA Emerenciã (Portugal), TOSKA Ketil (Norway), VANCLOOSTER Annick (Belgium)), WHITE Desley (UK), ZOLLER Heinz (Austria).

Apologies: ALTES Albert (Spain), BARDOU-JACQUET Edouard (France), DE MAEGHT Stéphane (Belgium), DELWART Bernard (Belgium), FINZEL Stephanie (Germany), HEAD David (UK), HORN SCHLEGEL Gerda (Switzerland), Mc GRATH Ann (Ireland), MULLETT Margaret (Ireland), RIALLAND Jean (France), RYAN John (Ireland).

Invited : SWINKELS Dorine (Netherlands), VAN DEURSEN Cees (Netherlands).

Welcome

The 2016 meeting is a joint meeting to EFAPH and HI (Haemochromatosis International). Barbara Butzeck and Ben Marris welcome the attendees from Europe, Australia and Brazil. They express their thanks to Pr Heinz Zoller for the excellent logistics of the meeting and, generally speaking, for the EIC Congress. Barbara points out that the attendance to all meetings has been strong. Both scientists and patients have been numerous and very active. She mentions that, parallel to the AGM, a session will be dedicated to the Non- HFE Registry (Pr Heinz Zoller) at the end of the morning.

1. EFAPH Activity Report October 2015 - April 2016 : Dr Françoise Courtois, General Secretary (*Appendix 2*)

Françoise comments a slide show. Highlights are as follows: participation to the EHA managed ERN for Haematological Rare Diseases, launching of an International working group on HH arthropathy, international rollout of the GPs awareness program, promising contacts with La Jolla Pharmaceutical Company (USA) and continued cooperation with EFS (French Blood Establishment). Patricia Evans is welcome as member of the HemoNews EU editorial committee. Issue 4 is now available in English and French (*Appendix 3*)

The activity report has been unanimously approved

2. EFAPH Financial Report – Dag Erling Stakvik, Treasurer (*Appendix 4*)

The total revenues (01/01/2015-31/12/2015) amount to 15,464 €, of which:

- membership fees	2,214
- collects and grants	6,200
- financial income	199
- charity concert	6,851

The total expenses (same period) amount to 10,558 €, of which:

- travel	8,073
- printing/communication	1,974
- mail/telecom	67
- banking fees	444

Profit for the year 2015 amounts to 4,906.-€. This is definitely an improvement over the previous years. The music event largely contributed to our profit, but this is not a recurring income.

Dag Erling expresses his thanks to Béatrice Coueslant and Jean-Daniel Kahn as volunteer assistant treasurers.

The Financial report is approved unanimously

3. Hemochromatosis News : Pr Pierre Brissot

Therapeutic aspects.

1:1. About bloodletting.

A) A prospective controlled study, by the Maastricht team, of 30 C282Y/C282Y patients concludes to the beneficial effect of using oral proton pump inhibitors in order to decrease the number of venesections during maintenance therapy.

B). A recent study, by the Rennes team, shows in 122 C282Y/C282Y patients, the interest of using an hemoglobinometer at bedside for checking haemoglobin levels during venesection therapy. It is an easy, quick, reliable, although somewhat expensive, procedure. A similar technique is also available for Ferritin measurement.

C) A work (Maastricht) has compared, in a prospective controlled study, erythrocytapheresis and "classical" phlebotomy in 44 C2828/C282Y patients. The main conclusion is that, although more complex and more expensive, erythrocytapheresis is well tolerated and more efficient.

1.2. Apart from bloodletting.

A) A study (Oxford) points out the potential therapeutic interest of a class of compounds that activates hepcidin production. Another team (Bonn) works on compound suppressing hepcidin inhibition.

B). Some HE gene history...: The C282Y mutation has been found by studying the DNA of prehistoric Irish individuals. This is the first demonstration of a Mendelian disease variant in prehistory and a confirmation of data that previously concluded that the HFE gene goes back to at least 3000-4000 years BC.

4. Moving forward on International Guidelines to Practical expert advice: Ben Marris

Ben Marris points out that guidelines are in demand all over the world; a lot is already available yet there is a need for convergence and harmonization.

1. Comparative analysis of existing guidelines : Annick Van Clooster (HVV) (Appendix 5)

Annick presents a comparative analysis of the North American, European and Dutch guidelines by a team of 7 experts from 5 disciplines (Appendix 5). One aspect of particular interest is the stakeholder's involvement (patients' viewpoint) for which only the Dutch score is satisfactory. Involving the patients is necessary because of the importance in terms of applicability. Interestingly, the total score of the North American guidelines is insufficient compared to the European and Dutch ones with regard to editorial independence, rigor of development...

2. Dutch HFE-HH Guidelines: Assessment of critical issues than need update : Pr Cees Van Deursen (Zuyderland Medical Centrum, Heerlen, The Netherlands)

Cees completes Annick's comments with the following remarks: the lack of evidence-based facts and of experts' opinions. The following points need to be improved: GPS's and specialists' awareness, diagnostic process, treatment and follow-up, general organisation and information. Reaching a consensus on biological indicators, the scope and limit of liver biopsy, liver MRI, HH penetrance and family screening is of the utmost importance. Further treatment indications and levels, phlebotomy vs erythrocytapheresis and life style changes (physiotherapy, ergotherapy, psychosocial aspects...) need to be discussed. Assessing organ damage (liver, heart, pancreas...) is necessary. Additionally, from an organisational viewpoint, costs, staff and blood donation problematics need to be reviewed. (Implication of Sanquin).

The discussion concluded that creating an expert group that would combine efforts with HI is necessary.

3. Input from national HH associations about major issues (Emerencia Teixeira, HI)

The survey covered 25 countries worldwide from the guidelines point of view (10 replies). Significant divergences can be noted with respect to the indicators. Pierre Brissot insisted on the need to come up with very simple and practical guidelines: "Treatment: who, when and how?" and to involve patients.

It is decided to form a working group composed of: Pierre Brissot, Paolo Santos, Sonia Distante, Graça Porto, Emerencia Teixeira, Brigitte Pineau, Ian Hiller (Canada), Domenico Girelli.

5. Non HFE Registry (joint meeting): (Pr Heinz Zoller- Innsbruck Hospital and University, Austria)

Heinz Zoller recapitulates the discussion of the separate session that took place in the morning. It has been agreed to develop a registry for non-HFE Patients in Europe. Several centres will join. Heinz discussed the need for informed consent of the patients to sign in the registry, with the patients' representatives. Approaches differ at the national level. It depends on which data are collected, e.g. year of birth. Heinz explains Ferroportin Disease Type A and B. Pierre Brissot indicates that the clinical definition is published on Orphanet.

Barbara suggests posting information about the Rare Forms (non HFE) and the related Expert Centres on EFAPH and HI websites. She also requests that experts should not forget patients with the classical Type 1 HFE-mutations C282Y and H63D. Domenico Girelli responds that a registry even when it starts as non-HFE could be a chance for the majority of HH-Patients.

6. EFAPH Projects update:

a) HH-Arthropathy working group (Barbara Butzeck):

Further to the Cologne meeting, a project team has been formed with P. Guggenbuhl, S. Finzel, P. Kiely, A. Braner, J. Zwerina. In November 2015 Stephanie and Patrick compiled a preliminary agenda: literature review, "unmet-needs", patients' survey on life quality due to iron-overload related arthropathy, imagery and immunological aspects of arthropathies, patients' involvement for early detection, genetic defects... A working meeting will take place in June 2016, on occasion of the EULAR meeting in London.

b) European Survey on Blood Donation (Emerencia Texeira): (Appendix 6)

In the context of preventive treatment of apparently healthy HH subjects, the question arises whether they are accepted as volunteer blood donors or not. In Europe, some countries accept them as such and others don't, thus raising problems of inequity among subjects. EFAPH conducted a survey among fourteen national Patients' Associations inviting their members to respond to an online questionnaire aimed to evaluate knowledge and attitudes regarding blood donation and the impact of decisions on the effectiveness of their preventive treatment. 1850 validated answers were obtained from a total of 1915 respondents, 1414 self-declared as HH patients.

Results confirmed that there is not a common policy regarding eligibility of HH subjects as blood donors amongst different European countries and showed a clear discrepancy between published blood bank policies and HH patients' perceptions in some countries (Denmark, Germany, Italy and The Netherlands). 86% of patients declared not feeling that refusals to donate blood have impact on their health, despite the impact on their feeling of beneficence and self-esteem. There is no evidence that patients claim to be blood donors for the purpose of healthcare. In contrast, a considerable proportion of respondents who were volunteer blood donors prior to their HH diagnosis, were subsequently refused. This highlights the inappropriateness of the most commonly used argument to refuse blood donations from HH subjects, which is that they cannot be considered volunteers.

c) World Blood Donor Day June 14th and Eurordis News:

c-1: World Blood Day (F. Courtois) :

Françoise emphasizes the importance of this event. It will take place this year on June 14th and is organized by the English and Welsh National Blood Institutes. It is an opportunity for EFAPH's National members to start a blood donation project (if not yet existing) as well as to make known the existence and activities at national, European and international levels) with the National Institution of their country and to lobby for blood check.

c-2: Register for Rare Barometer Voices to make your voice heard (F. Courtois):

Françoise refers to the initiative by EURORDIS aiming at registering 10 000 individuals affected by a rare disease.

In fact, concerning HH, it only refers to Non- HFE patients. The individuals concerned should refer to rare.barometer@eurordis.org to contribute to the effort in order to collect individual experiences about life quality and RDs.

d) News about the European reference networks – ERNs (Graça Porto): (Appendix 7)

Graça Porto has given an update on the European Reference Networks (ERNs) project and on the participation of EFAPH. The aim is to link highly specialized HCPs (Health Care Providers) across the EU so that the expertise is closer to Patients. The partners are the EU National Health Authorities and the European Commission, the Patients Organizations (monitored by EURORDIS, European RDs Platform) and the HCPs ("applicants"). The overall project is structured in 21 thematic Groups and EFAPH has been co-opted in the Rare Haematological Diseases Group; the common ground is the importance of research on iron metabolism in this field, the link with other haematological disorders and pre-existing regular communication between EFAPH and other Patients Organizations in the frame of the EHA Patients Advocacy Group (EHA-PAG). EFAPH is involved in screening candidate reference centres and as a member of the EHA-PAG (Coordinators: Prs. JL Vives Corrons, Barcelona and Pierre Fenaux, Paris). Current initiatives include to reply to the EU multidisciplinary questionnaire and to secure the endorsement of national associations, who in turn will seek the endorsement of the Member States Governments.

Conclusion and announcements for future meetings

Barbara Butzeck closed the meeting, thanking all participants for their engagement and lively discussions.

1. **June 26th/27th, 2017 in Muenster, Westfalen, Germany Next joint meeting + non-HFE Registry**
2. **May 7 – 11, 2017 BioIron: Los Angeles UCLA**
3. **February / March 2018 Zurich: EIC + AGM (joint meeting)**
4. **2019 BioIron Heidelberg Germany**
5. **2020 Barcelona (suggested by Mayka Sanchez)**

Annexes:

1. Agenda
2. EFAPH Annual Activity Report 2015-2016
3. Hemo News EU n°4
4. EFAPH Financial report
5. Comparative analysis of existing guidelines : Annick Van Clooster
6. European Survey on Blood Donation- Poster (Emerencia Texeira)
7. News about the European reference networks – ERNs (Graça Porto)



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